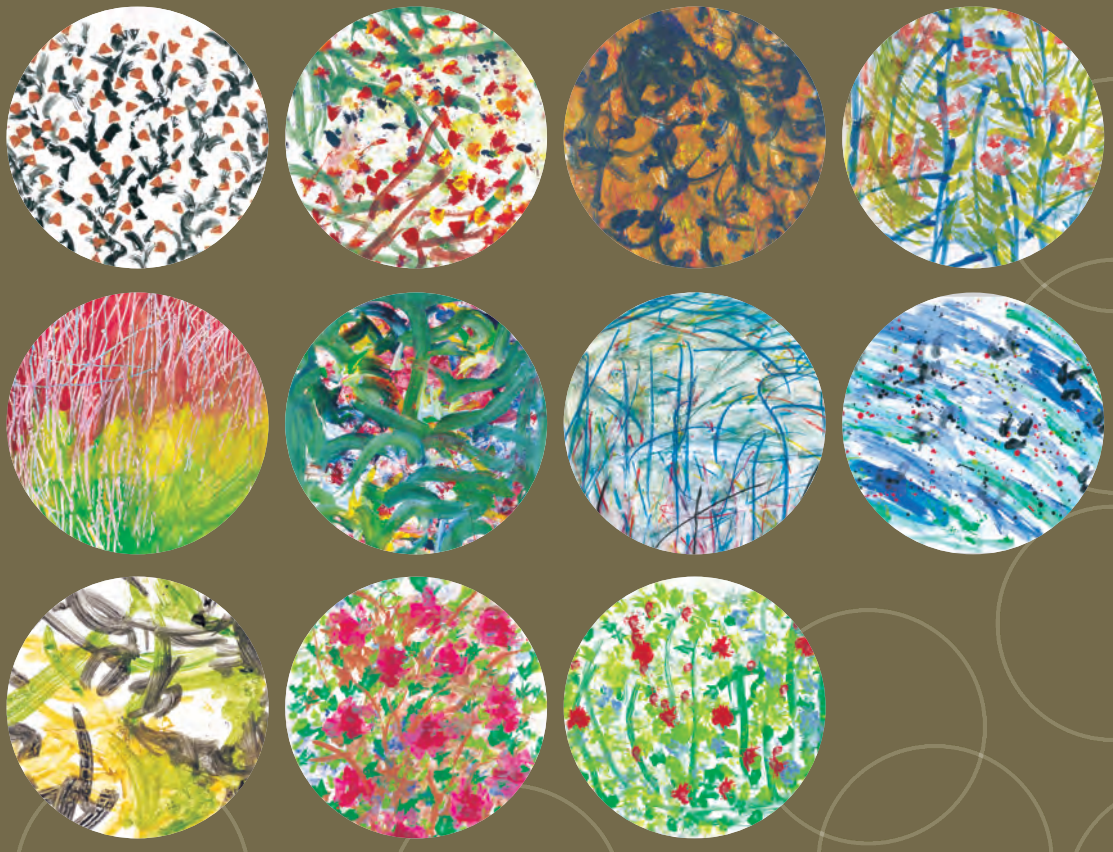
BIRTH DEFECTS IN SOUTH-EAST ASIA
A PUBLIC HEALTH CHALLENGE

SITUATION ANALYSIS



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A public health challenge

Situation Analysis

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The paintings used in this document are made by Mr Nitin, who is afflicted with Down syndrome.

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Contents

Abbreviations

ABBREVIATIONS

AIIMS	All India Institute of Medical Sciences, New Delhi
ANC	Antenatal care
ART	Assisted reproductive technology
BCG	Bacillus Calmette–Guérin
CMV	Cytomegalovirus
CRS	Congenital rubella syndrome
CVD	Cardiovascular diseases
DCR	Day-care room
DQ	Development quotient
DTP	Diphtheria-tetanus-pertussis
G6PD	Glucose-6-phosphate dehydrogenase
GDP	Gross domestic product
Hb	Haemoglobin
Hb CS	Haemoglobin Constant Spring
HBV	Hepatitis B virus
Hib	Haemophilus influenzae type b
HKI	Helen Keller International
HMIS	Health management information system
ICD10	Tenth Revision of the International Classification of Diseases
ICMR	Indian Council of Medical Research
IDD	Iodine deficiency disorder
IGMH	Indira Gandhi Memorial Hospital, Male
IMCI	Integrated management of childhood illness
IMR	Infant mortality rate
LMIC	Low- and middle-income countries
MDG	Millennium Development Goal
MMR	Measles-mumps-rubella
MOD	March of Dimes
NBE	National Board of Examinations, India
NE	Neonatal encephalopathy
NGO	Nongovernmental organization

NIP	National Immunization Programme
NPD	Neonatal-perinatal database
NTD	Neural tube defects
PEM	Protein energy malnutrition
PKU	Phenylketonuria
RMNCH	Reproductive, maternal, newborn and child health
RT-PCR	Reverse transcription polymerase chain reaction
SEA	South-East Asia
SEAR	WHO South-East Asia Region
TGP	Total goiter prevalence
TGR	Total goiter rate
UCI	Universal child immunization
UNESCAP	United Nations Economic and Social Commission for Asia and the Pacific
UNFPA	United Nations Population Fund
UNICEF	United Nations Children's Fund
USA	United States of America
USAID	United States Agency for International Development
WHA	World Health Assembly
WHO	World Health Organization
X-ALD	X-linkedadreno-leukodystrophy

1. INTRODUCTION

There has been a significant decline in infant and childhood mortality rates in most countries in the past two decades. This has primarily been due to extensive and successful use of immunization, control of diarrhoeal disorders, acute respiratory tract infections and improvement in health-care services through a focus on primary health care. As a consequence, birth defects are responsible for a greater proportion of infant and childhood mortality (World Bank, 1993). Indeed in developed countries birth defects cause 30–35% of perinatal, neonatal and childhood mortality. In developing countries, they contribute to about 5–7% of mortality, and this proportion is progressively increasing.

In 2010, the World Health Assembly (WHA), vide Executive Board agenda items EB125, 126 and 127 (WHO, 2010a), expressed concern about the high number of stillbirths and neonatal deaths occurring worldwide, and the large contribution of neonatal mortality to under-five mortality. It recognized the importance of birth defects as a cause of stillbirths and neonatal mortality, and that the attainment of MDG 4 on reduction of child mortality will require accelerated progress in reducing neonatal mortality, including prevention and management of birth defects. The Secretariat was therefore requested by the Member States to carry out the following activities to:

- raise awareness among all relevant stakeholders, including government officials, health professionals, civil society and the public, about the importance of birth defects as a cause of child morbidity and mortality;
- set priorities, commit resources, and develop plans and activities for integrating effective interventions that include comprehensive guidance, information and awareness-raising to prevent birth defects, and care for children with birth defects into existing maternal, reproductive and child health services and social welfare for all individuals who need them;
- record surveillance data on birth defects as part of national health information systems;
- develop expertise and build capacity on the prevention of birth defects and care of children with birth defects;
- strengthen research and studies on aetiology, diagnosis and prevention of major birth defects and to promote international cooperation in combating them; and
- promote the collection of data on the global burden of mortality and morbidity due to birth defects, and to consider broadening the groups of congenital abnormalities included in the classification when the International Statistical Classification of Diseases and Related Health Problems (Tenth Revision) is revised.

It was recommended that Member States should be supported in developing national plans for implementation of effective interventions to prevent and manage birth defects within their national maternal, newborn and child health plans. Support should also include strengthening health systems and primary care (including improved vaccination coverage such as for measles and rubella), food fortification and other preventive strategies of birth defects, promoting equitable access to such services, and strengthening surveillance of birth defects.

These important recommendations in the 2010 resolution WHA63. 17 (WHO, 2010a & b), form the basis of initiatives by the WHO Office for the South-East Asia Region for the prevention of birth defects.

Why have birth defects not received the attention they deserve to date from policy-makers, funding organizations and health-care providers? This is probably due to the misperception that these disorders are rare. In fact there is no nationally representative data in any of the Member States on the magnitude of birth defects and their contribution to foetal loss and newborn or infant mortality.

Another myth is that birth defects require expensive and high technology interventions for their care and prevention that are beyond the health budgets of low- and middle-income countries (LMIC). On the contrary, it has become apparent that simple technologies and strategies are at hand for the prevention of many birth defects.

Birth defects: a public health challenge

According to March of Dimes Reports on Birth Defects (MOD Foundation), every year more than 8.14 million children are born with a serious birth defect, due to genetic or environmental causes. Hundreds of thousands more are born with serious birth defects of post conception origin, including maternal exposure to environmental agents (teratogens) such as alcohol, rubella, syphilis and iodine deficiency that can harm a developing foetus. Serious birth defects can be lethal. For those who survive, these disorders can cause lifelong mental, physical, auditory or visual disability (Christianson, Howson and Modell, 2006)

This high toll of birth defects was only appreciated after infant mortality rates came down. In the low and middle income countries (LMIC), the burden of birth defects is much higher than in high-income countries. This is due to sharp differences in maternal health and other significant risk factors, including poverty, a high percentage of older mothers (in some countries), a greater frequency of consanguineous marriages etc. In LMIC, birth defects cause a tremendous drain on national resources, and urgent focus in these countries should therefore be on prevention. The urgency is clear at the sight of a child bound to a wheelchair because of being born with spina bifida, or a child with mental retardation due to hypothyroidism, or congenital rubella, or a family with two children with muscular dystrophy. Every child who has a preventable birth defect is a failure of medical care and public health systems that ignore available preventive measures. The failure to prevent birth defects is caused, in large measure, by the lack of organized effort and political will that are required to implement the necessary interventions.

Realizing the paucity of nationally representative data on birth defects in Member States of the Region, WHO-SEARO commissioned this situation analysis report on birth defects including burden of the problem and existing opportunities for prevention. A standard questionnaire was developed that was sent out to the countries to collect the information. In The Regional Programme Managers' meeting held in March 2012 the participating national programme managers from ministries of health presented the national data and information. Existing literature - published as well as unpublished (but significant) from all the Member States has been extensively reviewed to compile data and information related to birth defects.

It is hoped that the situational analysis would effectively highlight the public health importance of this neglected area of birth defects and help strengthen appropriate response for surveillance and prevention of common birth defects in the countries of South-East Asia Region.

2. DEFINITION AND CAUSES OF BIRTH DEFECTS

Definition

According to the Tenth Revision of the International Classification of Diseases (ICD10), congenital anomalies include congenital malformations, deformations and chromosomal abnormalities, but exclude inborn errors of metabolism. An expanded definition of birth defects, as stated by MOD, covers abnormalities of structure or function, including metabolism, which are present from birth. In this report the expanded definition of birth defects is used.

Causes and types

Congenital malformations are multifactorial in origin, i.e. they are caused by the interaction of both genetic and environmental factors. These comprise 65–70% of all birth defects. Birth defects may be due to chromosomal, single-gene or multifactorial causes. The frequency of these disorders is: chromosomal, 1 in 263 births; single-gene disorders, 1 in 81 births; and congenital malformations, 1 in 27 births. Environmental factors are responsible for 5–10% of total birth defects, and include nutritional deficiencies, infectious diseases, maternal medical conditions, teratogenic medications, alcohol, recreational drugs, and teratogenic pollutants.

Structural birth defects are related to a problem with body parts. Range of such defects includes neural tube defects (NTDs) such as spina bifida, and related problems of growth and development of the brain and spinal cord, cleft lip with or without cleft palate, heart defects, and abnormal limbs. Functional birth defects are related to a problem in working of a body part or system. These often lead to developmental disabilities and can include disorders such as:

- **Nervous system or brain problems –** learning disabilities, mental retardation, behavioural disorders, speech or language difficulties, convulsions, and movement trouble. Some examples of birth defects that affect the nervous system include autism, Down syndrome, Prader-Willi syndrome, and Fragile X syndrome.
- **Sensory problems –** blindness, cataracts and other visual problems, and varying degrees of hearing loss including deafness.
- **Metabolic disorders –** involving a body process or chemical pathway or reaction, such as conditions that limit the body's ability to get rid of waste material or harmful chemicals. Two common metabolic disorders are phenylketonuria (PKU) and hypothyroidism.
- **Degenerative disorders –** conditions that might not be obvious at birth, but develop intellectual delay that gets worse progressively. Examples are X-linked adreno-leukodystrophy (X-ALD), Rett syndrome, muscular dystrophy, and lysosomal storage disorders.

The causes of common birth defects, with examples, are listed in Table 1.

Table 1. Causes, classification, and examples of selected birth defects

Cause	Classification	Birth defect examples
Genetic	**Chromosomal**	**Down syndrome, Trisomy 18, Trisomy 13**
	Single gene	**α and β thalassemia, sickle-cell disorder, G6PD deficiency, albinism, cystic fibrosis, PKU, haemophilia A and B**
Environmental (teratogenic)	Infectious diseases	Congenital rubella syndrome, congenital cytomegalovirus, toxoplasmosis
	Maternal nutritional deficiencies (folic acid, iodine)	NTDs, iodine deficiency disorders
	Other maternal illnesses	Insulin-dependent diabetes mellitus, PKU
	Medications: thalidomide misoprostol, anticonvulsants, anticoagulants	Deformities of limbs
	Recreational drugs: alcohol	Several Neurological damage, foetal alcohol syndrome
	Pollutants: organic mercury	Neurological damage
Complex genetic and unknown	Congenital malformations involving single-organ systems	**Congenital heart disease, NTDs, cleft lip and/ or palate,talipes (clubfoot), developmental dysplasia of the hip**

Source: *Modified from Bale, Stoll and Lucas (2003).*

3. GLOBAL SCENARIO

As per March of Dimes (MOD) estimates, every year 6% of children worldwide are born with a serious birth defect/congenital disorder due to genetic or environmental causes. Based on the annual births data of 2010 (163 million,as per the World Health Statistics, 2012 Report) the estimate would be 9.78 million children. Globally, the most common serious birth defects of genetic or partially genetic origin are *(Christianson, Howson and Modell, 2006):*

- Congenital heart defects (1 040 835 births);
- Neural tube defects (323 904 births);
- Haemoglobin disorders, thalassemia and sickle-cell disease (307 897)
- Down syndrome – trisomy 21 (217 293 births);
- G6PD deficiency (177 032 births)

Combined, these five conditions account for about 25% of all birth defects.

According to the World Health Statistics 2012, about 7% of all under-five deaths globally are caused by congenital anomalies (WHO, 2012).

Regional differences range from 5% in the African Region, 7% in the South-East Asia (SEA) Region to 19% in the European Region. An estimated 11% of neonatal deaths are due to congenital anomalies in the People's Republic of China. These percentages are likely to be underestimated because they rely on data from verbal autopsy studies, thereby resulting in some probable misclassifications of deaths with disorders that need specialist diagnosis such as congenital heart defects getting missed. Viewed together, these figures indicate that, in the context of achieving the MDG 4 target, the issue of birth defects needs to be urgently addressed. Although congenital anomalies account for a smaller percentage of neonatal deaths in LMIC than in the wealthiest countries, more than 95% of all child deaths due to congenital anomalies occur in these settings, indicating that congenital anomalies impact all populations and represent a significant challenge to public health.

The distribution of infants born with birth defects annually by income of country is depicted in Table 2.

Table 2. Estimated number(millions)and percentage of annual total birth defects, early deaths due to birth defects, and under-5 deaths for low-, middle- and high-income countries

Countries	Low-income	Middle-income	High-income	Total millions
Annual total birth defects	4.75 (60%)	2.64 (34%)	0.49 (6%)	7.9
Annual early deaths	2.38 (72%)	0.79 (24%)	0.14 (4%)	3.3
Annual under-5 deaths	8.8 (80%)	1.8 (16%)	0.6 (4%)	11.2

Source: *Christianson A, Howson CP, Modell B, editors. March of dimes global report on birth defects: the hidden toll of dying and disabled children. New York: March of Dimes Birth Defects Foundation, White Plains, 2006.*

Estimates of infants born with serious birth defects in different regions of WHO are given in Table 3.

Table 3. Minimum estimates of infants with serious congenital disorders by WHO region

WHO Region	Population in millions,1996	Births/year, in millions, 1996	Congenital malformations/1000	Chromosomal disorders/1000	Single-gene disorders/1000	Total congenital disorders/1000	Annual affected live births
Eastern Mediterranean	506	18.1	35.7	4.3	27.3	69	1 237225
African	540	23.0	30.8	4.4	25	61	1 412427
South-East Asian	1,401	38.2	31.0	3.9	14.7	51	1946 606
European	867	10.8	31.3	3.7	12.4	49	522832
American	782	16.2	30.9	3.8	11.9	48	774235
Western Pacific	1,650	31.3	30.6	3.5	11.4	47	1464067
Total	5,746	137.6	31.5	3.9	16.8	53	7357392

Source: *World Health Organization. Primary health care approaches for prevention and control of congenital and genetic disorders: report of a WHO meeting, Cairo, Egypt, 6–8 December. Geneva: WHO, 1999a*

It is observed that birth defects cause both early mortality and chronic problems that contribute to a heavy cost for the family and society. It is estimated that almost 30% of live births with defects die early, about 27% have chronic problems, and about 43% are cured (Table 4).

Table 4. Prevalence and outcome of birth defects

Group of conditions	Birth prevalence /1000 live births	% of early mortality	% of chronic problems	% Cure	Early mortality /1000 live births	Chronic problems /1000	Cure/1000
Congenital malformations	36.5	22	24	54	8	8.8	19.7
Chromosomal disorders	3.8	34	64	2	1.3	2.4	0.1
Genetic Risk factor (rhesus)	2.6	0	0	100	0	0	2.6
Single gene disorders	12.3	58	31	11	7.1	3.8	1.4
Total	55.3	29.8	27.2	43.0	16.4	15.0	23.8

Source: *Christianson, Howson and Modell. March of dimes global report on birth defects: the hidden toll of dying and disabled children. New York: March of Dimes Birth Defects Foundation, White Plains, 2006 : Example from population of European origin.*

The prevalence of specific conditions varies widely in different populations. In countries where basic public health services are not available, the prevalence of serious birth defects is generally higher than in developed countries. As infant mortality rates fall, birth defects are responsible for an increasing proportion of infant mortality and morbidity (Modell and Kuliev, 1989; WHO, 1985, 1996, 1999b). In the majority of Latin American and Middle Eastern countries that have reduced infant mortality to less than 50 per 1000 live births, the infant mortality due to birth defects is as high as 25% (WHO, 1996), which is similar to the proportion in developed countries.

Post conception damage

Comparable data are difficult to derive for birth defects due to post conception damage caused by maternal exposure to teratogens, and to alcohol, tobacco, drugs, some infections and a number of toxic environmental agents. The limited data that do exist suggest the highest toll results from the following four post conception birth defects:

- Foetal alcohol spectrum disorder
- Birth iodine deficiency disorder
- Congenital rubella syndrome, and
- Congenital syphilis.

Together, these disorders account for hundreds of thousands of affected births. As with birth defects of genetic or partially genetic origin, post conception birth defects are more common in LMIC where the potential for exposure to teratogenic agents is greater and fewer preventive measures are in place than in high-income regions.

The average prevalence at birth of recognizable congenital malformations in developed and developing countries alike is 2–3.5%. NTDs have high prevalence at birth in China, Egypt, India, Mexico, and Central America, whereas cleft lip and palate are more prevalent in Asian populations. Advanced maternal age and reduced availability of prenatal diagnosis lead to a higher birth prevalence of chromosome anomalies (6 per 1000) and particularly of Down syndrome –2 to 3 per 1000 (WHO, 1985, 1996; Modell and Kuliev, 1989; Modell, Kuliev and Wagner, 1992). Major single-gene disorders in the developing world have a global incidence similar to that of industrialized countries, approximately 3.5 per 1000 (WHO, 1985), although the frequency of individual conditions vary. Haemoglobin disorders (sickle-cell disorder and thalassemia) constitute a major proportion of genetic conditions internationally, but particularly in the developing world which has the least resources for coping with the problem. Table 5 gives the causes of birth defects in high-income countries.

Table 5: Percentage of birth defects by cause in high-income countries

Cause	%
Preconception	
Chromosome disorders	6
Single-gene disorders	7.5
Multifactorial malformations	20–30
Postconception	
Teratogens	7–8
Intrauterine abnormalities	2
Subtotal	10
Unknown cause	50

Source: *Turnpenny and Ellard, 2005.*

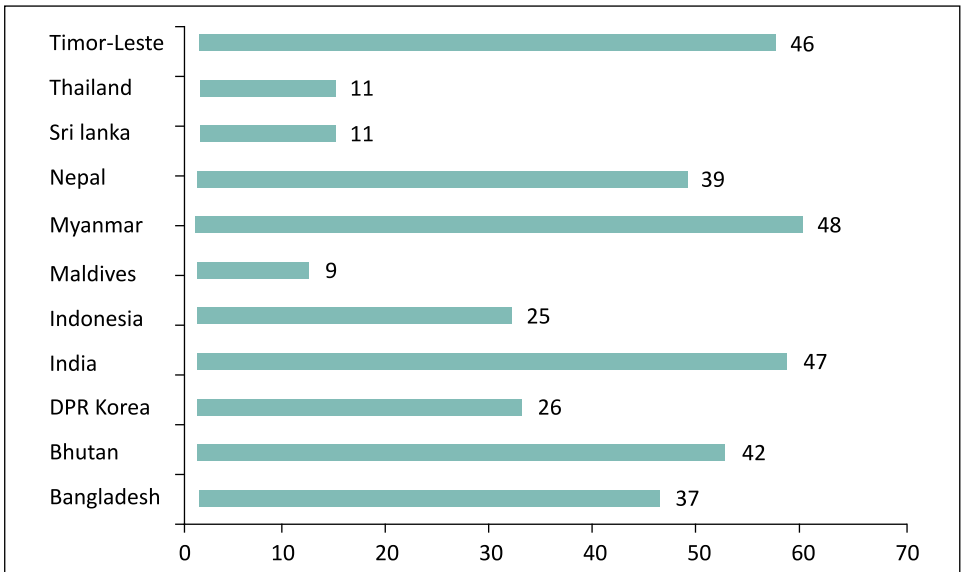
4. REGIONAL SCENARIO

Member States in the Region are witnessing a transition in disease pattern, and are grappling with the double burden of communicable and non communicable diseases (NCDs). Since communicable diseases have been brought under reasonable control, more attention is being focused on NCDs.

While there has been a progressive decline in child mortality in the Region, progress is not uniform as Member States are in different stages of development. The following graphics present the mortality rates across the Region.

Infant mortality rates in the countries of SEAR are summarized in Figure 1. The lowest IMR is observed in Maldives, while the highest is observed in Myanmar.

Figure 1. Infant mortality rate per 1000 live births in the South-East Asia Region

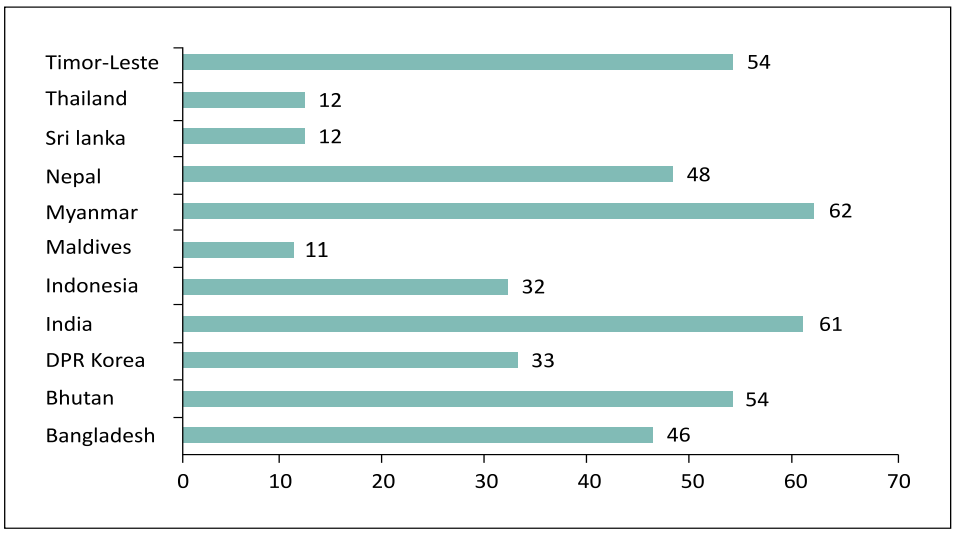


Source: *United Nations Inter-Agency Group for Child Mortality Estimation, 2012.*

Under-five mortality rates in SEAR are depicted in Figure 2.

The highest rates are in Myanmar, and the lowest in Maldives.

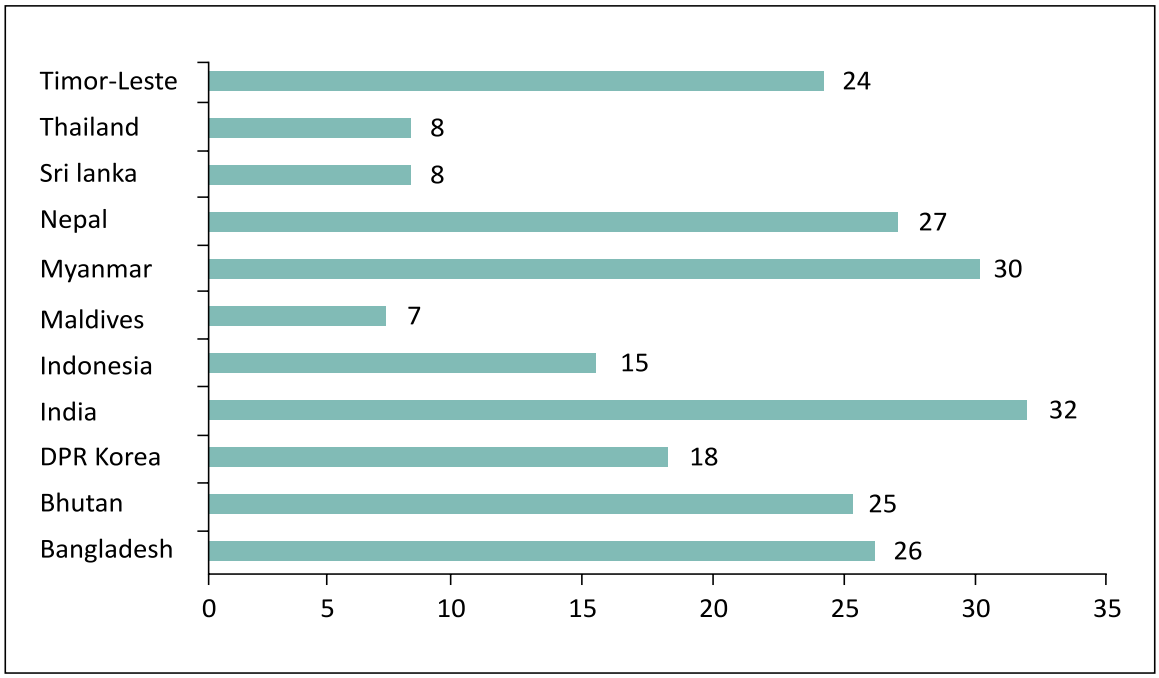
Figure 2. Under-five mortality rate per 1000 live births in the South-East Asia Region



Source: *United Nations Inter-Agency Group for Child Mortality Estimation, 2012*

Neonatal mortality rates in countries of the Region are shown in Figure 3. These are highest in India and lowest in Maldives.

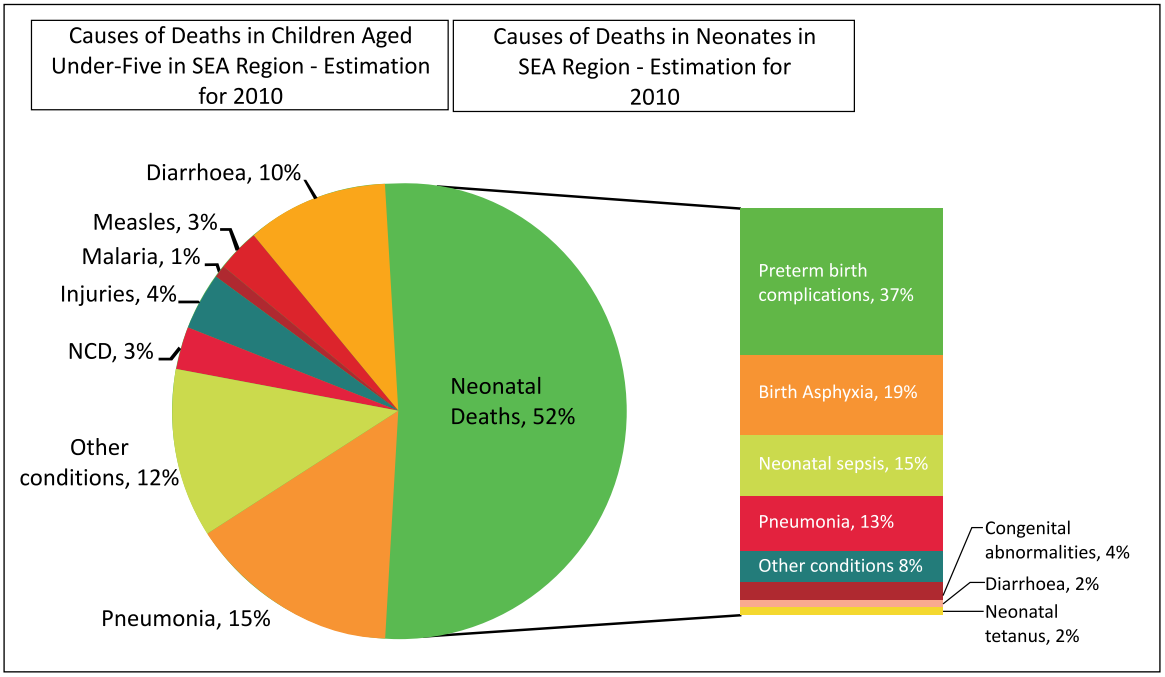
Figure 3. Neonatal mortality per 1000 livebirths in the South-East Asia Region



Source: *United Nations Inter-Agency Group for Child Mortality Estimation, 2012.*

Major causes of deaths in under-five children and in the neonatal period are depicted in Figure 4.

Figure 4. Causes of deaths in under-fives and neonatal period



Source: *WHO Global Health Observatory(http://www.who.int/gho/child_health/en/index.html)accessed Nov 2012.*

Although congenital abnormalities are responsible for an estimated 4% of neonatal deaths, it must be emphasized that birth defects contribute to a huge burden of foetal losses (abortions, medical terminations and stillbirths), the exact extent of which remains unknown. As the proportion of deaths due to infections and malnutrition decreases, birth defects will become a more and more important cause of newborn and child mortality. In addition, birth defects are known to contribute to lifelong disabilities with an enormous economic and social burden on society. It is important to remember that services for birth defects and genetic disorders are but a part of the general health services.

There is little doubt that birth defects cause enormous harm in settings where risk factors for many conditions are significant and resources for health care are limited, as in developing countries (Penchaszadeh, 2002). In some Member States, including those that have entered the phase of epidemiological transition, the burden of birth defects is yet to be recognized. Their significance is veiled by the continuing prevalence of infectious diseases and malnutrition. This, associated with limited diagnostic capability in clinical genetics, unreliable health records and statistics, infant and early childhood mortality of affected individuals, results in the lack of documentation of the majority of deaths from birth defects, which become absorbed in the general mortality statistics and thus are not acknowledged for what they truly represent.

Table 6 displays the demographic characteristics of countries in the South-East Asia Region.

Table 6. Demographic characteristics of countries in South-East Asia Region

Member State	Total population (thousands), 2010	GNI per capita (PPP int. $), 2010	Life expectancy at birth (years), 2009	Total adult literacy rate (%), 2005–2010*	Congenital anomalies as cause of death in children < 5 years (%), 2010
Bangladesh	148692	1810	65	56	7
Bhutan	726	4990	63	53	7
DPR Korea	24346	0	70	100	11
India	1 224614	3550	65	63	7
Indonesia	239871	4200	68	92	9
Maldives	316	8110	75	98	30
Myanmar	47963	1950	64	92	5
Nepal	29959	1210	67	59	5
Sri Lanka	20860	5010	71	91	30
Thailand	69122	8190	70	94	27
Timor-Leste	1124	3600	67	51	5

Source: *World Health Organization. World Health Statistics 2012. Geneva: WHO, 2012*

Some birth defects are clinically obvious at birth; others may only be diagnosed later in life. Spina bifida is one example of a structural defect that is obvious at birth. The bleeding disorder haemophilia is a functional defect that usually not clinically obvious until infancy or childhood. The frequency of birth defects in countries in SEAR is summarized in Table 7.

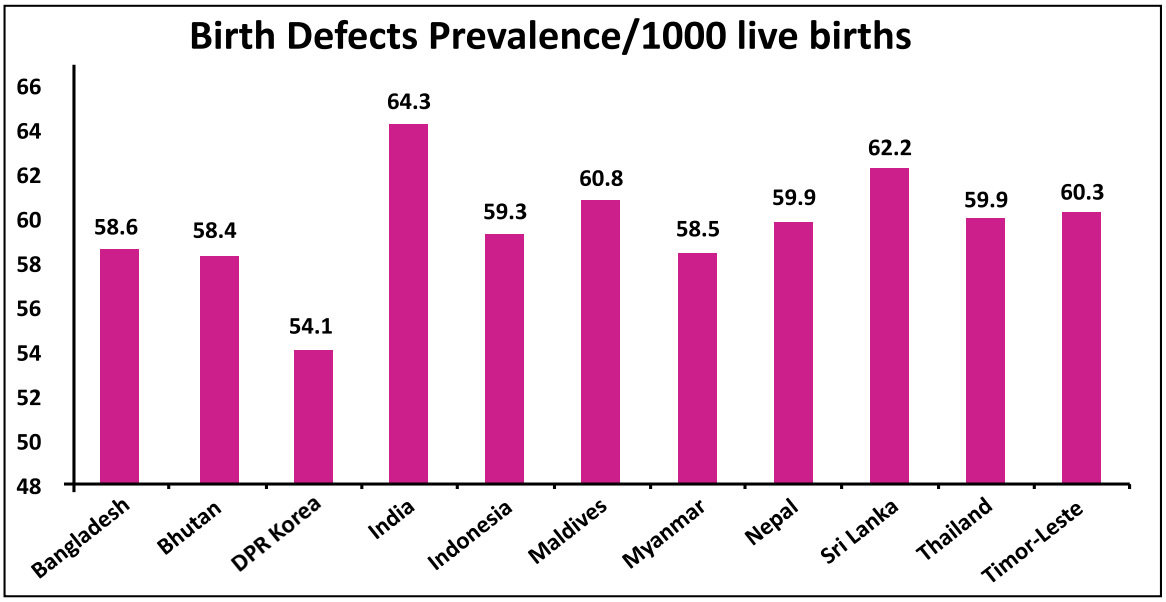
Table 7. Estimated Prevalence of common birth defects in the South-East Asia Region

Member State	Children born with birth defects annually	2009 annual births (000s)	Birth defects of the cardiovascular system	Neural tube defects	Haemoglobin syndromes	Down syndrome	G6PD deficiency	Total of birth prevalence with no known preventive strategies in place
		Per 1000 live births						
Bangladesh	199 299	3401	7.9	4.7	0.7	1.6	1	58.6
Bhutan	876	15	7.9	4.7	0	2.1	2.1	58.4
DPR Korea	17 691	327	7.9	4.7	0	0.8	0.1	54.1
India	1722404	26 787	7.9	4.7	1.2	1.6	2.4	64.3
Indonesia	260 090	4 386	7.9	0.7	0.8	1.4	0.9	59.3
Maldives	365	6	7.9	2	6.4	1.7	1.7	60.8
Myanmar	59 436	1 016	7.9	0.7	4	1.7	3.1	58.5
Nepal	43 727	730	7.9	4.7	0.2	2.1	3.4	59.9
Sri Lanka	22 641	364	7.9	2	0.6	1.9	1.9	62.2
Thailand	58 522	977	7.9	0.7	5.6	1.5	3.6	59.9
Timor-Leste	140	46	7.9	0.7	1	2.1	10.5	60.3

Source: *Estimated from the March of Dimes Report (Christianson, Howson and Modell, 2006), using annual births of 2009 as given in the statistical tables of the United Nations Children's Fund (UNICEF) (accessed 7 November 2011).*

The prevalence of birth defects per 1000 births in SEAR, as per the March of Dimes Report, is displayed in Figure 5.

Figure 5. Birth defects prevalence in the South-East Asia Region/1000 live births



Source: *Christianson, Howson and Modell. March of dimes global report on birth defects: the hidden toll of dying and disabled children. New York: March of Dimes Birth Defects Foundation, White Plains, 2006.w*

5. COUNTRY SCENARIOS

SEARO undertook an initiative to collect information related to birth defects in Member States in the Region. A standard questionnaire was developed to collect information on the burden and common types of birth defects, existing policies and guidelines related to well-known birth defect prevention interventions, opportunities to integrate such strategies in existing programmes, challenges in this area and support that countries may need to mount birth defects prevention programmes. This questionnaire was sent to WHO Country Offices that helped collect the required data from ministries of health and other related sources

WHO organized a Regional Experts Group Meeting and a Regional Meeting of Programme Managers on Prevention and Control of Birth Defects. Invited participants were requested to collect information on birth defects in their countries to share in the meetings. Published literature was also reviewed to obtain additional information from the Member States.

Countrywide information related to birth defects is presented in this section under the following headings:

1. **Demographic information**
    a. Causes of neonatal and child mortality
2. **Information related to birth defects**
    a. Congenital malformations
    b. Perinatal infections
    c. Micronutrient deficiency disorders
    d. Thalassemia
    e. Other genetic disorders
    f. Disabilities
    g. Consanguinity
    h. Birth defects prevalence as per the March of Dimes Report
3. **Noncommunicable diseases**
4. **Country response to birth defects**
    a. Surveillance programmes
    b. Genetic services
    c. Screening programmes
    d. Prevention programmes
    e. Micronutrient fortification with folic acid
5. **Services for care of people with birth defects**
6. **Further opportunities for prevention of birth defects**

BANGLADESH

Bangladesh

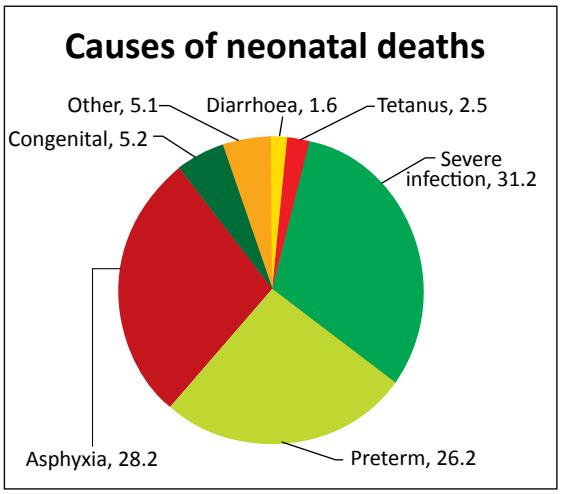
1. Demographic information

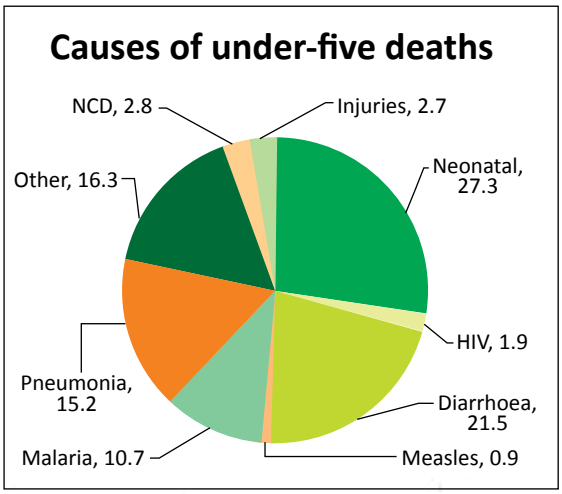
Total population ('000)	148 692
Estimated annual births ('000)	3 038
Under-five mortality rate (per 1000 live births)	46
Infant mortality rate (per 1000 live births)	37
Neonatal mortality rate (per 1000 live births)	26
Perinatal mortality rate (per 1000 live births)	50
Number of stillbirths	232
Complete immunization coverage by 12 months (%)	83
Complete immunization coverage at any time between 12–23 months (%)	86
Measles vaccination coverage by 12 months (%)	84
Measles vaccination coverage at any time between 12–23 months (%)	88
Vaccination against rubella:	Yes
Antenatal care (ANC) coverage (any) (%)	68
First ANC visit before 4 months (%) (data from 2007)	24
ANC visits: one visit (%)	15
ANC visits: 4 or more (%)	26
Iron folic acid supplementation coverage (%) (data from 2007)	55
Institutional deliveries (%)	29
Deliveries assisted by skilled birth attendants (%)	32

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *National Institute of Population Research and Training (NIPORT), Mitra and Associates, and ICF International. Bangladesh Demographic and Health Survey 2011. Dhaka, Bangladesh and Calverton, Maryland, USA: NIPORT, Mitra and Associates, and ICF International, 2013.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010.*

2. Information related to birth defects

Data and information on birth defects are not available at national level. A number of studies have been published reporting the frequency of birth defects in defined populations and geographic areas.

a. Congenital malformations

Rashid (2002) scanned 5841 obstetric patients over a period of 34 months. Of 41 cases of congenital anomalies, seven were hydrocephalus, seven hydronephrosis, five were mild hydrocele (which could be a physiological variant), five anencephaly, five foetal ascites, three omphalocele, two small biparietal diameter, two short-limbs, and one each of gastroschisis, renal cyst, hepatosplenomegaly, and pleural effusion with oedema (hydrops fetalis). The higher detection of hydrocephalus and anencephalus were due to the fact that these were clinically suspected by the obstetrician and referred for a scan.

Khanum, Noor and Kawser (2004) studied prospectively 11 680 consecutive deliveries for congenital malformations,the overall incidence of which was 2.3%. Musculoskeletal was the most commonly involved system by NTD.

Begum et al. (2007) determined the causes of perinatal death using Wigglesworth classification in all-live and stillborn babies delivered at BIRDEM over a five-year period (January 2000 to December 2004). The majority of deaths were in the group "macerated stillbirths" (63.5%). Perinatal asphyxia was responsible for 13.6% of deaths, lethal congenital malformation was found in 11.2% and immaturity and "others" each accounted for 6.1%.

Peto et al (unpublished, 2012) carried out a community-based birth defects surveillance in rural Bangladesh (Gaibandha and Rangpur Districts), from 2001-2007 as part of maternal micronutrient supplementation trials (JiVitA Project, Johns Hopkins University). Local field workers were trained for first level screening through home-visit to examine the infants age 1-3 months whose mothers had been enrolled in the study. Any abnormalities of any part of the body were noted on standardized forms and digital pictures taken. The findings were reviewed by the study physician for confirmation. Among 46,851 live born infants that were surveyed, 9686 infants with abnormalities were identified by trained field workers of whom 1,173 birth abnormalities were confirmed and digitally photographed and documented. Among major abnormalities that were identified were polysyndactyly, orofacial clefts, hypospadias, and anotia/microtia. Among minor defects that were identified were minor ear malformations, periauricular ear pits, umbilical hernias and thin upper lip. The experience showed that a medical and digital technology-supported rural birth-defect surveillance system through trained lay workers is feasible. However, as lay workers were asked to identify"anything abnormal" and to overstate the cases, sensitivity was presumably maximized. Specificity was improved by sequential stages of research physician's examination and provisional diagnosis, followed by an expert review of the photographic data.

Ellis et al. (2011) collected information on 31 967 deliveries in 18 unions of 3 districts of Bangladesh from 2005 to 2008, using traditional birth attendants as key informants. Of these deliveries, 26 173 (82%) occurred at home. For home deliveries, the mean cluster adjusted stillbirth rate was 26/1000 births, the perinatal mortality rate 51/1000 births, and the NMR 33/1000 live births. There were 3186 (12.5%) home-born infants who did not breathe immediately. Of 201 fresh stillbirths, 40 (14%) had major congenital abnormalities. Abnormal limbs were reported in 18, anencephaly in 13, spina bifida in 3, and a combination thereof in 4.

Shahidullah (personal communication, 2012) analysed 7000 deliveries in the Department of Obstetrics in Banga Bandhu Sheikh Mujib Medical University from 2007 to 2011. Congenital anomalies were found in 232 (3.31%) cases. The commonest malformations were of the central nervous system, 88 (37.9%), urinary, 50 (21.5%), gastrointestinal, 42 (18.1%), musculoskeletal, 14 (6.0%), cardiac, 4 (1.7%), multiple anomalies, 22 (9.5%), and non-immune hydrops, 12 (5.2%).

Neural tube defects

Dey et al. (2010) reported that NTDs had a frequency of 1.38 per 1000 among 20 267 deliveries in three hospitals in Dhaka. They measured zinc concentration in 32 mothers and their newborns with NTDs as well as 32 mothers with their babies with no known abnormalities as controls. The mean serum zinc level of the case and control mothers was 610.2 µg/L and 883.0 µg/L, respectively ($P < 0.01$). The mean serum zinc level of the case and control newborns was 723 µg/L and 1,046 µg/L, respectively ($P < 0.01$).

Gamble et al. (2005) measured plasma concentrations of homocysteine, folate, and cobalamin and urinary concentrations of creatinine in 1650 adults in Bangladesh. The prevalence of hyperhomocysteinemia in men (mean 11.4 µmol/L) was markedly greater than in women (mean 10.4 µmol/L) – (P 0.0001). Folate was lower (9.8 + 6.5 and 12.3 + 7.6 nmol/L, respectively), whereas cobalamin was higher (281 + 115 and 256 + 118 pmol/L, respectively). Folate explained 15% and cobalamin explained 5% of the variation in homocysteine concentrations.

Khambalia O'Connor and Zlothin (2009) showed that in rural Bangladesh, of 272 nulliparous women, 37% were anaemic (haemoglobin < 120 g/L), and 13% were folate deficient (plasma folate ≤ 10 nmol/L). The Micronutrient Initiative and UNICEF (2004) estimated that approximately 8000 severe birth defects occur annually, including infantile paralysis due to folate deficiency.

Howell, Barnett and Underwood (2001) showed thatwhite women from the United Kingdom were 5.7 times more likely to take folic acid supplements than Bangladeshi women.

b. Perinatal infections

Rahman et al. (2002) analysed 198 hearing-impaired children and 200 children without hearing problems. Rubella antibody was detected in 74% of hearing-impaired children and in 18% of those with normal hearing: this finding correlated with the presence of rubella antibody in mothers (67%) of rubella seropositive hearing-impaired children. In contrast, rubella antibody was observed in only 14% of mothers of children without hearing problems. Consistent with the presence of antibody, 41% of the seropositive mothers who had hearing-impaired children gave a history of fever and rash during early pregnancy. This study indicates a strong association between rubella infection and hearing impairment in Bangladeshi children. In addition, it also indicates that infection by rubella virus is common in Bangladesh: this suggests that priority should be given to implementing appropriate measures for the control of rubella.

c. Micronutrient deficiency disorders

The Micronutrient Initiative estimated that about 750 000 Bangladeshi babies born each year with intellectual impairment are caused by iodine deficiency during pregnancy (The Micronutrient Initiative and UNICEF, 2004).

Eighty four percent of all edible salt is iodized. The preliminary report of the Bangladesh Nutrient Survey 2012, estimated 57.6% of households consume adequately iodized salt.

This coverage has increased from 51.2% since 2004-2005. The median of urine excretion level is 163 µg/L (ICCIDD, 2012).The prevalence of goiter in school-aged children decreased from 50% in 1993 to 6% in 2004/2005 as a direct result of salt iodization. Prevalence of severe iodine deficiencies in school-aged children was reduced from 23.4% in 1993 to 4% in 2004/2005 (Emma et al., 2011).

L*indstrom et al. (2010)* examined the prevalence of anaemia and micronutrient deficiencies as well as their determinants in early pregnancy in Matlab, a subdistrict in rural Bangladesh from 1 January to 31 December 2002. Pregnant women (n = 740) were enrolled in approximately week 14 in pregnancy. Anaemia was present in 28% of the women, 55% were zinc deficient, 46%were vitamin B-12 deficient and 18% were folate deficient. Anaemia was not associated with iron deficiency but rather with vitamin B-12 deficiency. Infestation with Ascaris was highly prevalent (67%) and associated with both folate and vitaminB-12 deficiency. Anaemia and micronutrient deficiencies all varied significantly with season. Ascaris infection is possibly associated with vitamin B-12 deficiency due to malabsorption and inefficient uptake of B-12. Vitamin B-12 deficiency in early pregnancy could lead to adverse pregnancy outcomes and increase the risk of metabolic syndrome in adult life.

d. Thalassemia

Thalassemia is a major problem in Bangladesh and haemoglobin (Hb) E is also fairly common. *Khan, Banu and Amin (2005)* reported the frequency of carriers to be 7%, which means roughly 11.2 million Bangladeshis are thalassemia carriers. It is estimated that about 7483 children are born with the disease every year with expected 374 154 living patients (Bangladesh Thalassemia Foundation). A well-functioning thalassemia centre (ASHA) has been established in Dhaka in the major children's hospital. Attempts are being made to establish prenatal diagnostic services for thalassemia. As in other Muslim countries consanguineous marriages are highly frequent, pointing to the need for genetic diagnostic and counselling services.

e. Other genetic disorders

Apart from thalassemia, other common conditions are Hb E, haemophilia, Wilson disease, Duchenne muscular dystrophy, and G6PD deficiency. Of chromosomal disorders,Down syndrome is the commonest cytogenetic abnormality detected, followed by Turner syndrome, Intersex disorders and Klinefelter syndrome at the Cytogenetics Lab, BSMMU *(Habib et al., 2007).*

f. Disabilities

About 5.6% of people in Bangladesh have a disability of one kind or another: hearing, 18.6%; visual, 32.2%; speech, 3.9%; physical, 27.8%; intellectual, 6.7%; and multiple (more than one type), 10.7% *(Titumir and Hossain, 2005).* In another survey of 10 299 subjects, 2% of children were found to have mental retardation *(Durkin et al., 2000)*. Age standard prevalence of bilateral blindness and binocular lowvision were 1.53% and 13.8%,respectively *(Dineen et al.,2003).*

g. Consanguinity

The practice of marrying biological relatives is common in many parts of Bangladesh *(Ambrus, Field and Torero, 2010).* The Matlab Health and Socioeconomic Survey (MHSS) in1996 reported first-degree consanguineous marriage in 10% of people (Rahman et al, 2001). An earlier household-based survey of the Teknaf region of Bangladesh showed that 17.6% of marriages were consanguineous *(Khan, unpublished, 1997).*

h. Birth defects as per March of Dimes Report (2006)

The March of Dimes Report estimated that 199 299 children were born with birth defects annually in Bangladesh *(Christianson, Howson and Modell, 2006).* These comprise 26 868 children with defects of the cardiovascular system, 15 985 with NTDs, 2381 with haemoglobinopathies (this is an underestimate, according to the Bangladesh Thalassemia Foundation), 5442 with Down syndrome, and 3410 with G6PD deficiency.

3. Noncommunicable diseases

Bangladesh has been facing a dual burden of existing infectious diseases and an escalating rise in NCDs like diabetes, heart disease, stroke, cancer, and chronic respiratory disease. Around 12.5% of deaths are caused by CVD. Approximate prevalence rates are hypertension, 12%; diabetes mellitus, 10% in urban areas and 5% in rural areas;and chronic obstructive pulmonary disease, 3%. Cancer is the major cause of mortality causing about 20% of all deaths (Bangladesh NCD Network, 2011).

To prepare for the challenge of these diseases, a nationwide survey was conducted to determine the distribution of NCD risk factors in the Bangladeshi adult population(WHO-Country Office Bangladesh, 2011).Of 9275 respondents in the survey, 4312 were men and 4963 were women. The mean age was 42.4 years. Around 98.7% of the survey population had at least one risk factor of NCDs, around 77% had two or more risk factors and 28.3% had three or more risk factors. More women were found to have three or more risk factors than men. The report shows no clear differential in risk trend with varying levels of wealth. Smoking was prevalent in 26.2% of people surveyed (mostly men). Smokeless tobacco use represented 31.7% of tobacco use. Low fruit and vegetable intake was present in 95.7%, low physical activity in 27%, obesity in 18%, hypertension in 14.8%, and existing diabetes in 3.9%.

4. Country response to birth defects

Bangladesh has made impressive progress in health. There has been an increase in vitamin A coverage from 82% to 97% and an increase in the use of emergency obstetric care from 27% to 47% *(UNICEF 2010; MoHFW Bangladesh, 2009).* The achievement of an93% immunization rate of three doses of combined diphtheria/pertussis/tetanus vaccine (Upazila, 2010; NIPORT, Mitra Associates and ICF International, 2013) merited an award from the GAVI Alliance. The decline in child mortality merited the United Nations Millennium Development Goal 4 award. Today, there are some 20 million people taking action to improve hygiene and sanitation practices and seeking quality services in health, water and child protection. Since 2006, the country has seen a five-fold increase in birth registration. There is also a close alignment of both the Children's Act 2010, approved by the Cabinet, and the National Policy on Children with the Convention on the Rights of the Child; the government has been supported to develop a monitoring framework on recommendations of the Committee on the Rights of the Child. Equally important are more frequent, meaningful interactions of children with policy-makers. However, high proportion of early marriage, low coverages of antenatal care, delivery at health facilities and assisted birth by skilled providers are still the challenges (NIPORT, Mitra Associates and ICF International, 2013).

a. Surveillance programmes

There are no surveillance programmes for birth defects at present but the country has experience of surveillance in other areas.

- There is an excellent Nutritional Surveillance Project of Helen Keller International, Bangladesh, implemented in partnership with the Government of Bangladesh's Institute of Public Health Nutrition. This is a technically sound surveillance system with rapid turnover of data and findings.
- There is a surveillance network for communicable diseases such as HIV/ sexually transmitted infections, acute flaccid paralysis and poliomyelitis.
- There is a national registry for cancer.

b. Genetic services

- Genetic counselling is provided by the physician/paediatrician involved in patient care. The Department of Medical Genetics and Centre for Genetic Counselling are not yet established. Two trained histopathologists are working as geneticists. There are five cytogenetic, several biochemical and a few molecular genetic laboratories; all are in Dhaka city. Ultrasound studies during pregnancy are easily available and used frequently for prenatal diagnosis.
- Genetic laboratories: there are five cytogenetic laboratories in the University and Dhaka city. There are few biochemical and molecular laboratories. Some of the larger Indian laboratories have opened branches in Dhaka to collect samples and send them for analysis in India.

c. Screening programmes

Currently there is no nationwide genetic screening programme, except for newborn screening in certain parts of the country.

- Antenatal screening for anaemia is being carried out from Upazila and above. Screening for haemoglobinopathies; infectious diseases like syphilis, HIV, rubella, Cytomegalovirus (CMV); toxoplasmosis; Down syndrome; and NTDs is done only in high-risk target groups.
- Antenatal screening of mothers for genetic disorders/malformations is carried out only in target groups at risk (couples having a family history, a previously affected offspring, history of recurrent abortions/intrauterine death) or with high index of clinical suspicion.
- Ultrasound studies during pregnancy are easily available and are used frequently for prenatal diagnosis of birth defects at some centres.
- Newborn screening: the only countrywide programme is for congenital hypothyroidism. The Institute of Nuclear Medicine and Ultrasound, Dhaka with its five regional centres has screened 200 000 newborns countrywide over the last four years by assaying thyroid-stimulating hormone (TSH) in the cord blood. They confirmed 88 cases of congenital hypothyroidism, suggesting an incidence of 1 in 2200 births. The scale of coverage is less than 5% of the whole country (unpublished data, Institute of Nuclear Medicine and Ultrasound, Dhaka).

d. Prevention programmes

- A measles and rubella vaccine has been added to the immunization schedule from June 2012.
- Avoidance of pregnancy after 35 years of age is advised sporadically as a counselling message.
- Avoidance of medications/X-rays/ environmental hazards during early pregnancy (first trimester) is included in the ANC counselling package.
- Detection and treatment of diabetes in women before and during pregnancy is carried out routinely during the antenatal period.
- Prenatal diagnosis is carried out predominantly by ultrasonogram and biochemical markers in maternal serum. Amniocentesis is carried out by a few expert obstetricians on personal initiative. Chorionic villus sampling (CVS) is not being done.
- Menstrual regulation is used to terminate pregnancy to save the life of the mother. It depends on the identification of the foetal abnormality/ period of gestation/ access to health-care facility. It is permitted up to 12 weeks of pregnancy.
- Health care, educational and public facilities have been declared 100% smoke free. It is banned andis punishable by law. [Smoking and Using Tobacco Products (Control) Act, 2005.
- Alcohol is banned by law.

e. Micronutrient/folic acid fortification programme

There is an iron and folic acid supplementation programme for adolescent girls, pregnant and lactating women. No campaign, legislation or national policies/programmes exist for periconceptional folic acid supplementation.

Food fortification by micronutrients is not being carried out. A salt company in Bangladesh has announced plans to fortify salt with iron and iodine. Small-scale efforts to fortify wheat flour have taken place. However, the country has low wheat consumption. There is a salt law for universal salt iodization.

5. Services for care of people with birth defects

Haemophilia facilities for laboratory investigations and treatment with factor VIII and IX are available. There are no community-based rehabilitation programmes. The Bangladesh Protobondhi Foundationconducts school-based programmes (Kalyani Special School) and other activities, but thescope of corrective and rehabilitative surgeries is limited. Parent organizations exist for disorders like thalassemia, haemophilia, childhood cancers and autistic spectrum disorders.Corrective/rehabilitative surgeries are available at both public and private facilities.

6. Further opportunities for prevention of birth defects

Bangladesh has a good surveillance system for nutritional disorders. Therefore, the expertise exists for carrying out surveillance programmes. National Neonatal Health Strategy and Guidelines have been developed with involvement of all stakeholders. A neonatal and perinatal database as part of a SEAR initiative is being developed by a network of selected hospitals. Centres that are part of this perinatal network can be entrusted with the task of evolving a Birth Defects Registry based on the recordingof information in the major neonatal units in the country.

Studies show that NTDs have a high frequency, and therefore prevention of these defects should be a high priority. The focus should be on fortification of food with folic acid and vitamin B12. A good thalassemia centre exists and should be strengthened so that it can perform prenatal diagnosis and thus mount a control programme in the country. This should be a high priority due to the severe burden of the disease in Bangladesh. There is also a well-planned control programme for NCDs. A part of the funds available for NCDs should be allocated to the control of birth defects and thalassemia.

There are many trained and skilled paediatricians and physicians. There is a need to set up a Department of Medical Genetics in the National Children's Hospital, and this can be a forerunner to start similar departments in other universities. This will initiate the process of manpower development in the management of birth defects and genetic disorders.

Country experts have identified main stakeholders as Ministry of Health and Family Welfare; national institutions like Bangabandhu Sheikh Mujib Medical University, Institute of Nuclear Medicine and Ultrasound, Dhaka, Centres of Nuclear Medicine and Ultrasound, Institute of Child and Maternal Health (ICMH), Matuail, Dhaka, Bangladesh Institute of Child Health and Dhaka Shishu Hospital, BRAC (Bangladesh Rural Advancement Committee); professional associations like Bangladesh Paediatrics Association and Bangladesh Neonatology Forum; and international agencies like WHO, UNICEF, United Nations Population Fund (UNFPA), United States Agency for International Development (USAID),Australian Agency for International Development (AusAID), and Save the Children. The existence of multiple stakeholders is useful to advocatefor more government attention to birth defects.

BHUTAN

Bhutan

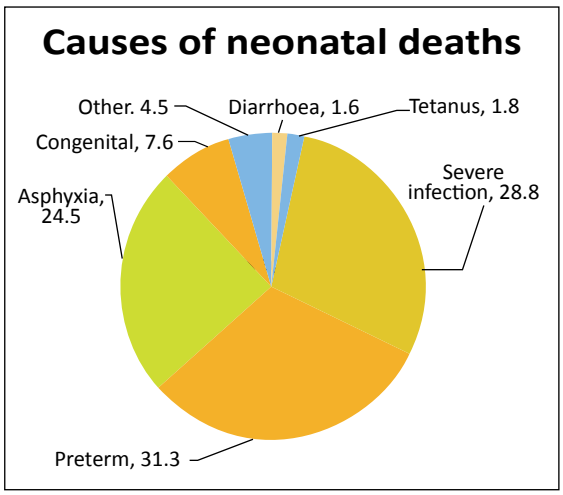
1. Demographic information

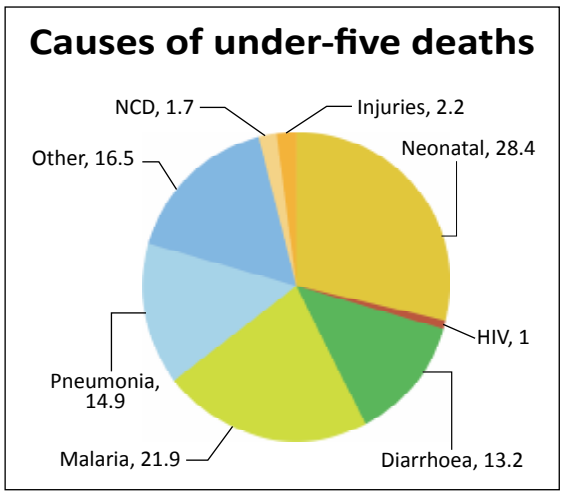
Total population ('000)	726
Estimated annual births ('000)	15
Under-five mortality rate (per 1000 live births)	54
Infant mortality rate (per 1000 live births)	42
Neonatal mortality rate (per 1000 live births)	25
Perinatal mortality rate (per 1000 live births)	No data
Number of stillbirths	No data
Complete immunization coverage by 12 months (%)	91
Complete immunization coverage at any time between 12–23 months (%)	No data
Measles vaccination coverage by 12 months (%)	95
Measles vaccination coverage at any time between 12–23 months (%)	No data
Vaccination against rubella:	Yes
Antenatal care coverage (any) (%)	97
First ANC visit before 4 months (%)	No data
ANC visits: one visit (%)	97
ANC visits: 4 or more (%)	77
Iron folic acid supplementation coverage (%)	No data
Institutional deliveries (%)	63
Deliveries assisted by skilled birth attendants (%)	65

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *National Statistics Bureau, Royal Government of Bhutan. Bhutan Multiple Indicator Survey 2010. Thimphu, Bhutan: National Statistics Bureau, 2011.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

Bhutan is a small country with only 15000 births per year but there is hardly any published data on the number of birth defects and genetic disorders.

a. Congenital malformations

March of Dimes estimated that 876 children are born with birth defects annually in Bhutan (Christianson, Howson and Modell, 2006). These comprise 119 children with defects of cardiovascular system, 71 children with neural tube defects, 32 children with Down syndrome, and 32 children with G6PD deficiency.

b. Perinatal infections

No data are recorded for perinatal infections.

c. Micronutrient deficiency disorders

The first nationwide IDD assessment in 1983 found the total goitre rate (TGR) to be 64.5% and salt iodization almost non-existent. In 1984 the Iodine Deficiency Disorder Control Programme (IDDCP) was established with five major components: salt iodization and distribution, administration of iodized oil injections, monitoring of iodine content in salt, internal evaluation of the programme and community-level education. A salt iodization plant was set up in 1985 in Phuentsholing as one of the key measures to combat IDD. Following these interventions, the second nationwide study undertaken in 1991–1992 found that the TGR had fallen to an average 25.5% and iodized salt coverage averaged 95.8%. A 1996 report found the TGR among school children aged 6 to 11 years to be 14%, salt iodization close to 100% and iodized coverage at 82%(Royal Government of Bhutan nutrition section, et al., 1996).In 1997, His Holiness issued a statement which led to intensification of the iodine deficiency control program so that by 2003 the elimination of iodine deficiency disorders has been sustained. This has been a major success story of elimination of iodine deficiency disorders in the South-East Asia Region (Sithey, 2006). The coverage of households consume iodized salt is almost universal, 96.2%, with median level of urinary iodine excretion at 217 µg/L (ICCIDD, 2012).

d. Thalassemia

There are hardly any cases of β thalassemia. It is expected, however, that there are cases of Hb E, which by itself does not lead to clinical problems. Bhasin (2006) reported the frequency of Hb E trait as 0.01 to 0.05 in Bhutan.

e. Other genetic disorders

In a hospital study of 3512 deliveries, 9.7% occurred in the age group 31–35 years, while 4.5% occurred in the age group above 35 years (Bhandari, unpublished data, 2010)

f. Disabilities

In 2002, a pilot survey on mental disorders carried out in three districts observed the prevalence of mental retardation to be 14%. The 2004 National Population and Housing Census stated that 3.4% of the total population had disabilities (Office of Census, 2005).

g. Consanguinity

No data have yet been published on consanguinity but cases of second-degree consanguinity do exist.

h. Birth defects as per the March of Dimes Report

The March of Dimes Report on Birth Defects estimated that 876 children were born with birth defects annually in Bhutan (Christianson, Howson and Modell, 2006). These comprise 119 children with defects of the cardiovascular system, 71 with NTDs, 32 with Down syndrome, and 32 with G6PD deficiency.

3. Noncommunicable diseases

A National High-Level Multi-sectoral Advocacy Meeting on Non-Communicable Diseases was organized in July 2011. The advocacy meeting was attended by the Minister of Health, Minister for Education, and other top dignitaries of the Government. Several topics were actively discussed, particularly on (1) the very stringent Tobacco Control Act of Bhutan, (2) the recently introduced taxation on alcohol, (3) the effect of rapid urbanization, and (4) changes in lifestyle that were leading to rising trends in morbidity and mortality due to NCDs in the country. The need to emphasize simple interventions that would prevent more than 80% of NCDs was highlighted by many participants; these were health promotion messages, school health programmes and community involvement to ensure healthy lifestyles, including through women's empowerment.

As cases of diabetes, cancer, and heart diseases appear to be on the rise, the Health Department plans to develop a programme for prevention and control of these problems.

4. Country response to birth defects

The positive points include: a small birth cohort, strong birth registration system, a comprehensive school health programme, periconceptional folic acid supplementation nationwide, rubella vaccination, free health care and referral. In addition, institutional deliveries are being improved.

a. Surveillance programmes

There is no surveillance system for birth defects, but the country could develop one using the existing mechanisms.

- Hospital-based health information system is in place.
- Birth Register mentions congenital anomalies.
- Perinatal Medicine Unit exists at Jigme Dorji Wangchuck National Referral Hospital.
- There is a comprehensive school health screening programme.
- Cancer Registry is being developed.

b. Genetic services

Specialized genetic services are not available at present and patients requiring attention are referred to centres outside the country, mainly to India and Thailand. Bhutan looks forward to assistance from more developed countries of the Region and WHO support to initiate activities in this area. There are only a handful of paediatricians who counsel for common genetic disorders like Down syndrome.

c. Screening programmes

Antenatal screening programmes have been introduced. Ultrasound studies are used in district hospitals to confirm gestational age and placental localization. Only at the National Referral Hospital ultrasound studies are used to scan for anomalies. Screening for anaemia during pregnancy is carried out in all health facilities, even in the periphery. Screening for infectious diseases like syphilis, HIV, rubella, CMV, and toxoplasmosis is available only at the district hospital level. Screening for NTDs is done by ultrasound only at the national central hospital in Thimphu.

d. Prevention programmes

A rubella vaccine programme for children and women of childbearing age has been in place since 2006 (nationwide campaign), when it became part of the routine immunization schedule.

Informative hoardings and posters have been put up around the country to sensitize women about healthy pregnancy. For example, such methods educate women about the high risk of birth defects in pregnancies after the age of 35 years. They are also useful resources on the adverse effects of some medications, ultraviolet radiation and other environmental hazards on pregnancy, especially in the first trimester.

Education/awareness programmes for pregnant women and the general population are included in the Maternal and Child Health handbook. Avoidance of pregnancy after 35 years of age,certain medications,X-rays,and environmental hazards during early pregnancy are emphasized in the advocacy posters (Figure 6).Detection and treatment of diabetes in women before and during pregnancy is encouraged.

Medical termination of pregnancy is legalized, but can be carried out only if the woman's life is at risk or threatened or if the unborn child is expected to suffer severe physical and mental abnormalities as a result of the pregnancy. To carry out medical termination, the certification of two medical doctors is required.

Alcohol awareness programmes have been introduced on television and through campaigns to help women better understand the adverse effects of alcohol consumption both before and after pregnancy. Counselling sessions have also been arranged for such women in Bhutan. Moreover, high taxation is applied on the purchase of alcohol. Bhutan is perhaps the first country to have banned the consumption of tobacco, for which the country has set strict legislation.

Figure 6. Poster displaying information regarding precautions during pregnancy

e. Micronutrient fortification programme

IDDs have been significantly reduced through the commercial distribution of iodized salt. Bhutan imports the majority of its flour and has little or no domestic milling industry. Therefore, food fortification with micronutrients would be a challenge. However, means should be explored for fortification of food and rice with folic acid and vitamin B12; as a small country, implementation should be easier.

5. Services for care of people with birth defects

The health sector established a community-based disability and rehabilitation programme in early 1997. Zangley Muenselling School for the Visually Impaired was set up in Khaling, decades ago. Corrective/rehabilitative surgery for cleft lip/palate repair is done twice a year.

6. Further opportunities for prevention of birth defects

As there is a shortage of paediatricians, the few that exist should be a given a refresher course in genetic counselling. Some nurses or social workers could be trained in genetic counselling to help families with genetic disorders.

Folic acid supplementation (on a regular basis) should be advised for expectant mothers in their first trimester. Also, the same should be recommended to lactating mothers and to women planning to conceive. Routine immunization like rubella should be offered to women in childbearing age and to newborns. A comprehensive school health programme allows children to be screened for congenital malformations like cleft lip, heart disease and deafness and provided treatment for the same.

Forms for registration of birth defects could be easily modified to include the presence of a birth defect and its precise diagnosis, and thus provide information on the frequency of birth defects. However in Bhutan 78.3% of deliveries take place at home and only 18.9% take place in health facilities. Deliveries by trained attendants are 72% (WHO, 2011). In the absence of trained birth attendants, it is noteworthy that husbands and mothers/ mothers-in-law play a significant role during delivery. Therefore awareness programmes on birth defects in communities is necessary.

Neonatal units in hospitals should maintain a perinatal database of the causes of morbidity and mortality in newborns. This would provide information about the etiological role of adverse factors and facilitate a strategy for their control.

DEMOCRATIC PEOPLE'S REPUBLIC OF KOREA

Democratic People's Republic of Korea

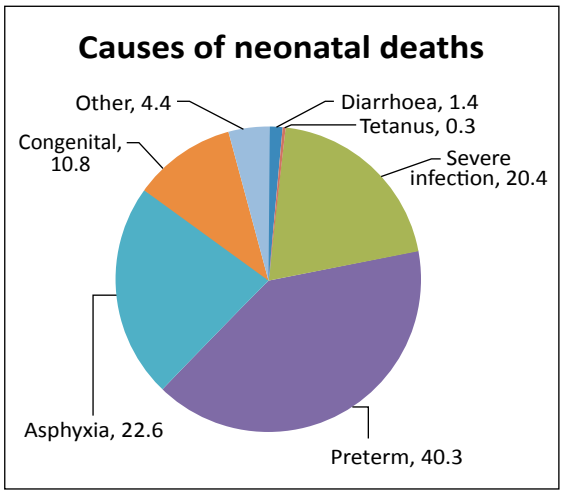
1. Demographic information

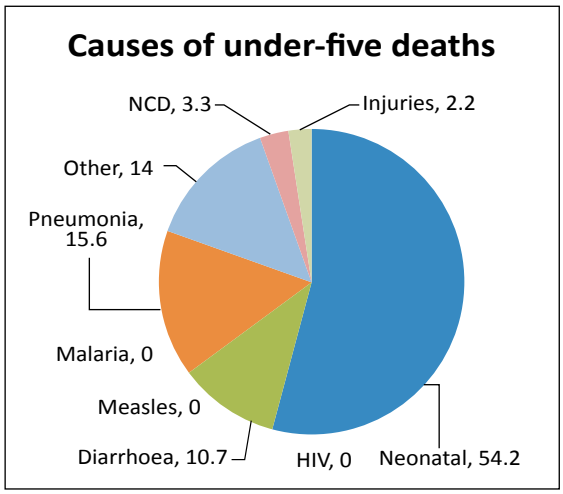
Total population ('000)	24346
Estimated annual births('000)	348
Under-five mortality rate (per 1000 live births)	33
Infant mortality rate (per 1000 live births)	26
Neonatal mortality rate (per 1000 live births)	18
Perinatal mortality rate (per 1000 live births)	No data
Number of stillbirths	No data
Complete immunization coverage by 12 months (%)	93
Complete immunization coverage at any time between 12–23 months (%)	No data
Measles vaccination coverage by 12 months (%)	99
Measles vaccination coverage at any time between 12–23 months (%)	No data
Vaccination against rubella:	No
Antenatal care coverage (any) (%)	100
First ANC visit before 4 months (%)	No data
ANC visits: one visit (%)	100
ANC visits: 4 or more (%)	94
Iron folic acid supplementation coverage (%)	No data
Institutional deliveries (%)	95
Deliveries assisted by skilled birth attendants (%)	100

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *Central Bureau of Statistics (CBS), Democratic People's Republic of Korea. Democratic People's Republic of Korea Multiple Indicator Cluster Survey 2009, Final Report. Pyongyang: CBS, 2010.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

a. Perinatal infections

The high prevalence (estimated at 4.5% in 2003) of blood-borne hepatitis-related morbidity and mortality has long been recognized by the Government of the Democratic People's Republic of Korea (DPR Korea), but the true prevalence of chronic hepatitis B infection is unknown. A vertical prevention and control programme exists from the central to the community levels.

b. Micronutrient deficiency disorders

Coverage of households using iodized salt is low, around 24.5%, and the median urinary iodine excretion is 97 µg/L (ICCIDD, 2012).

c. Thalassemia

No data are available on the frequency of these disorders. However, based on data for the Democratic Republic of Korea there is expected to be a low frequency of carrier rate for thalassemia. This is possibly due to the absence of selection in favour of the β thalassemia genes.

d. Other genetic disorders

No data are available.

e. Disabilities

Various degrees of disability affect 3.4% of the population, of which disability affecting the limbs is a substantial proportion. The majority of people affected by disability reside in rural areas (65%), and males slightly outnumber females in this category. There is a higher prevalence of disability in the older age groups (WHO, 2010b).

f. Consanguinity

No data are available.

g. Birth defects as per the March of Dimes Report

The March of Dimes Report on Birth Defects estimated that 17 691 children were born with birth defects annually in DPR Korea (Christianson, Howson and Modell, 2006). These comprise 2583 children with defects of the cardiovascular system, 1537 with NTDs, 262 with Down syndrome, and 33 with G6PD deficiency.

3. Noncommunicable diseases

These diseases account for an increasing burden of morbidity and mortality. In 2002, heart disease accounted for the major burden of NCD, particularly heart disease caused by rheumatic fever, ischemic heart disease and other related ailments. Cerebrovascular diseases, cancer, chronic respiratory disease and neurological diseases were also prevalent. According to Ministry of Public Health (MoPH) officials, these diseases account for approximately 60% of all causes of mortality (MoPH DPRK, 2007). The three major causes of death in DPR Korea are ischemic heart disease (13%), lower respiratory infections (11%) and cerebrovascular disease (7%). The prevalence of cerebrovascular disease is 17.8 per 10 000 population, of cancer 14.4 per 10 000 population and chronic respiratory diseases 26.5 per 10 000 population

The high prevalence rate of smoking tobacco (54.5%) in the adult male population is also a major contributor to the burden of NCDs (WHO DPRK, 2011).

A strategic priority has been to address women and children's health, and reduce the risk factors that lead to an increasing prevalence of NCDs.

4. Country response to birth defects

a. Surveillance programmes

Control of communicable and re-emerging diseases is one area in which progress has been substantial. This can be easily extended to birth defects and genetic disorders.

b. Genetic services

No data are available.

c. Screening programmes

All pregnant women are encouraged and expected to register for health care within the first three months of their pregnancy, to receive essential screening and monitoring. Postnatal care is well established, during which all women are seen five times. Extensive use is made of mass media, in particular Central TV, education and cultural programmes on television, broadcasting and various newspapers and printed media, for health education and for promotion of key health messages.

d. Prevention programmes

Immunization coverage with all vaccines is high (> 90%). Measles vaccine is included in the schedule (UNICEF, 2010). Vitamin A supplementation is given to 98% of all children under 2 years of age. There is a high proportion of institutional deliveries.

Comprehensive services for children are provided through Ri hospitals, urban polyclinics and the household doctor. Integrated management of childhood illness (IMCI) training adapted to the needs identified in DPR Korea has recently been introduced in the country and will be expanded over time.

Health services for pregnant women,mothers, newborns and children are organized in a comprehensive package that links the Ri hospitals, polyclinics,household doctors, nurses and midwives. Service points are organized so that they are within 30 minutes walking distance to where women and children live.

e. Micronutrient fortification programmes

Universal salt iodization has been achieved. The country receives fortified wheat flour as food assistance, but little is known about its domestic wheat industry and opportunities for fortification.

5. Services for care of people with birth defects

Information is not available.

6. Further opportunities for prevention

The vaccination policy could be extended to cover rubella.

The birth registration form could be modified to collect information on the presence of a malformation at birth. The collection of information on birth defects may be initiated in the neonatal unit in the main hospital in the capital. It should be feasible to set up a surveillance system for birth defects, given the good infrastructure.

INDIA

India

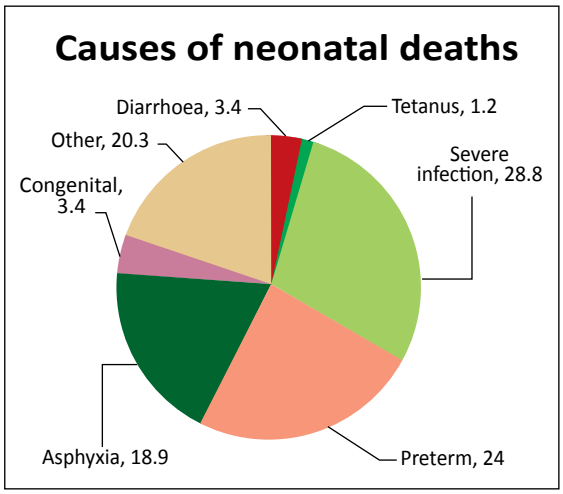
1. Demographic information

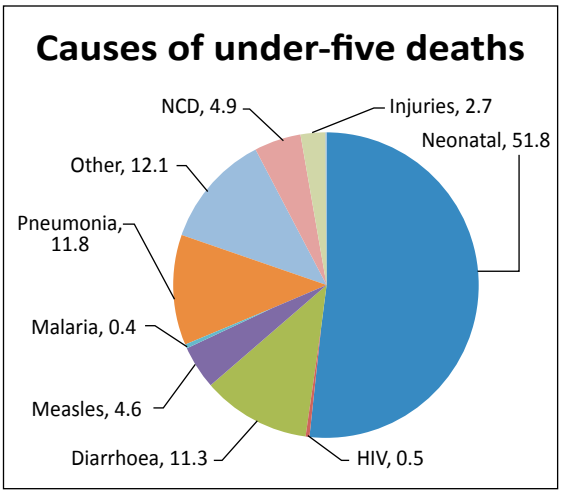
Total population ('000)	1 224 614
Estimated annual births ('000)	27 165
Under-five mortality rate (per 1000 live births)	61
Infant mortality rate (per 1000 live births)	47
Neonatal mortality rate (per 1000 live births)	32
Perinatal mortality rate (per 1000 live births)	48.50
Number of stillbirths	1 105
Complete immunization coverage by 12 months (%)	36
Complete Immunization coverage at any time between 12–23 months (%)	44
Measles vaccination coverage by 12 months (%)	48
Measles vaccination coverage at any time between 12–23 months (%)	59
Vaccination against rubella:	Selected States
Antenatal care (ANC) coverage (any) (%)	76
First ANC visit before 4 months (%)	44
ANC visits: 1 visit (%)	9
ANC visits: 4 or more (%)	48
Iron folic acid supplementation coverage (%)	65
Institutional deliveries (%)	39
Deliveries assisted by skilled birth attendants (%)	47

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012.*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012.*

3. *International Institute for Population Sciences (IIPS) and Macro International. National Family Health Survey (NFHS-3), 2005–06: India: Volume I.Mumbai: IIPS, 2007.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

Genetic disorders and birth defects are relatively common in India. This is due to the fact that many communities marry consanguineously. In South India, marriages among Hindus are consanguineous in 20–30% of cases, especially uncle–niece marriages. Muslims marry consanguineously in 20–30% of cases. Thalassemia and sickle-cell disease also have a high frequency in many states. Moreover, with improvements in health care and intensive care facilities, many infants with birth defects and inborn errors of metabolism now survive. Indeed due to the large number of births per year (almost 27 million), March of Dimes Report estimated that India has the largest number of infants born with birth defects in the world (Table 8) (Christianson, Howson and Modell, 2006).

a. Congenital malformations

Like many other Member countries, India is undergoing an epidemiological transition. Communicable diseases are on the decline due to better living conditions and health care. On the other hand, the relative increase in the prevalence of noncommunicable diseases threatens to be a public health problem in this country. One such group of disorders is congenital malformations. Though several studies have been done on congenital malformations in India since the early 1960s, coinciding with the thalidomide tragedy across the world, no uniform methods have evolved for the surveillance of birth defects. Each study has come out with varying results, not only because of the geographic variation in birth defects but also due to the different standards adopted by each study in data collection, case definition and because of other methodological issues.

In cities, congenital malformations and genetic disorders have become important causes of mortality in the perinatal period. For example, during the 1970s All India Institute of Medical Sciences (AIIMS) Hospital,New Delhi reported fewer deaths caused by congenital malformations during the perinatal period than sepsis. This pattern was reversed in the 1980s and, by the 1990s congenital malformations surpassed immaturity as a cause of perinatal mortality (Hospital records – Unpublished). A somewhat similar trend has been observed in hospitals in other large cities. In a Mumbai hospital that mainly serves the poor, congenital malformations was the third commonest cause (13.2%) of perinatal mortality (Ravikumara,Bhat and Oumachigui, 1996). Likewise, in a nationwide study conducted by the National Neonatology Forum in 1995 of 1460 stillbirths and 1400 neonatal deaths, congenital malformations were the second commonest cause (9.9%) of mortality among stillbirths, and the fourth commonest cause (9.6%) of neonatal mortality.

A study funded by the Department of Atomic Energy was carried out in three cities (Baroda, Mumbai and New Delhi) using a uniform performa (Verma et al., unpublished, 2012). General hospitals were selected, to reflect the overall population. The staff members at all centres were trained in dysmorphology. Major malformations recorded from 94 610 persons enrolled in the study gave a malformation rate of 2.12% (Taksande et al., 2010). The systemic distribution of these malformations is depicted in Table 8.

Table 9 shows the estimated number of infants born annually with various malformations in India, deduced from the data of this study. Such studies can be used to assess the requirement for surgical services, as well as to assess the effect of preventive strategies, e.g. for NTDs (CNS defects). It may be noticed that congenital heart defects, one of the most common defects observed at birth, are under-represented as echocardiography was only performed in infants who became symptomatic.

Table 8. Estimated frequency of birth defects and genetic disorders in India

Disorder	Incidence	Births / year
Congenital malformations	1: 50	678,000
Down syndrome	1: 800	34,000
Metabolic disorders	1:1200	22,477
β thalassemia and sickle-cell disease	1: 2700	16,700
Congenital hypothyroidism	1: 2477	10,900
Duchenne muscular dystrophy	1:5000 (M)	2,700
Spinal muscular atrophy	1:10,000	2,700

Source: *Modified from Verma and Kumar (2012).*

Table 9. The systemic distribution of malformations in the three-centre study

System	No.	Per 10 000	Total / year
Central nervous system	569	60.18	162486
Musculoskeletal	484	51.12	138024
Gastrointestinal	363	38.37	103599
Genitourinary	204	21.58	58266
Ears	141	14.89	40203
Cardiovascular	120	12.70	34290
Respiratory	65	6.87	18549
Eyes	57	6.03	16281
Miscellaneous	238	25.12	67824
Total			493522

The two-year birth defects surveillance study was carried out by the Genetic Research Centre, National Institute for Research in Reproductive Health, Parel, Mumbai in collaboration with the Nowrosjee Wadia Maternity Hospital, as a part of Department of Atomic Energy study mentioned above, but the authors published the data separately (Patel and Adhia, 2005). It was found that, of the 17 653 consecutive newborns examined by paediatricians and geneticists, 294 (1.6%) had major malformations, 1400 (7.92%) had minor malformations, and 328 (1.8%) were stillbirths. Malformations were highest in the stillbirth group. Polygenic traits accounted for 45.1% while chromosomal etiology was found in 4%. A genetic basis was found in 65.4% of cases (Table 10).

Table 10. Rate of selected malformations and total births per annum in India

Malformation	Rate per 10 000	Total / year
Spina bifida/anencephaly	40.42	109134
Talipes	18.60	50220
Cleft lip and cleft palate / Cleft palate alone	15.42	41634
Hydrocephalous alone	11.20	30240
Hypospadias	5.71	15417
Tracheo-esophageal fistula and esophageal atresia	5.08	13716
Intestinal and anal atresia	5.08	13716
Obstructive uropathy	4.23	11421
Diaphragmatic hernia	3.17	8559
Exomphalos	2.75	7425
Anopthalmia and micoropthalmia	2.54	6858
Congenital dislocation of hip	1.59	4293

Neural tube defects

NTDs were the commonest disorders observed in the Department of Atomic Energy study, described above. A previous meta-analysis of 515 500 births from various parts of India revealed a prevalence of anencephaly and spina bifida of 2.5 and 1.5 per 1000, respectively (Verma, 1978a; 1978b). The prevalence was extremely high in north India (more than 4 per 1000 births), especially in the states of Punjab, Chandigarh, Haryana, New Delhi and Rajasthan, with a decreasing trend in frequency towards the east (Bihar and West Bengal) and south (Tamil Nadu and Kerala). The reported high frequency in North Indians is also observed when they migrate to the developed countries (Singapore and Canada). The frequency of NTDs in Sikhs living in British Columbia, Canada, was reported to be 2.86/1000 while the overall rate was 1.26/1000 in that area. A higher frequency has also been reported from Davangere, Karnataka. It was suggested that this may be due to consanguinity *(Kulkarni, Mathew and Ramachandran, 1987; Kulkarni, Mathew and Reddy, 1989).* Recent unpublished data from the Birth Defects Registry of India has identified Visnagar, Gujarat, as another high NTD reporting area.

The frequency in the United States of America (USA) and Europe is reportedly below 1/1000, where there has been a progressive decline through supplementation and fortification of food with folic acid. In cities in India obstetricians do advise their patients to take folic acid periconceptionally. However as many pregnancies are unplanned it would be best to fortify food with folic acid and other nutrients *(Godbole and Yajnik, 2009).*

b. Perinatal infections

All types of intrauterine infections have been described in India (Deka, 2010). Commonly observed ones are cytomegalovirus, rubella, toxoplasmosis, herpes simplex, chicken pox, parvovirus, hepatitis B, hepatitis E and HIV infection. Although there is no official policy on testing for intrauterine infections during pregnancy, most obstetricians arrange for these tests to be done. Once a year, or so, patients who are positive for IgM antibodies against rubella come in a cluster to Foetal medicine unit, indicating an infection in the community. This emphasizes the need for introduction of rubella vaccine in the immunization schedule.

c. Micronutrient deficiency disorders

Anaemia

India continues to have severe burden of anaemia. The NFHS-3 survey (2005/06) estimated that the prevalence of anaemia is high in all age groups: 55.3% in women of reproductive age, 53.2% innon-pregnant non-lactating women, 56% among adolescent girls, and 69.5% in children aged under-five (IIPS and Macro International, 2007). The prevalence of anaemia is higher in India in comparison to that in South-East Asia countries like Bangladesh, Nepal and Sri Lanka.

Anaemia during pregnancy accounts for one-fifth of maternal deaths and is a major factor responsible for low birth weight (Lokare et al, 2012). Recent studies by Viveki et al (2012) and Lokare et al (2012) show that the prevalence of anaemia among pregnant women is very high (82.9% and 87.2% respectively). Previous studies also showed the similarity. For instance, a study carried out by Nutrition Foundation of India in 7 states had observed the overall prevalence of anaemia as 84% among pregnant women *(Agarwal and Agarwal, 2006).* The Indian Council Medical Research (ICMR) Task Force Multicentre Study revealed that the overall prevalence of anaemia among pregnant women from 16 districts was 84.9% *(range 61.0% - 96.8%) (Toteja et al., 2006).*

Iodine Deficiency Disorders (IDD)

IDD is another major nutritional disorder in India (Kapil, 2010). Surveys conducted by the Centre and State governments and other agencies show that not even a single state is free from the problem of IDD. Of 324 districts surveyed, 263 showed a Total Goitre Rate of 10% or more. Coverage of households use iodized salt is low, around 51%, and the median urinary excretion level is 154 µg/L (ICCIDD, 2012).

d. Thalassemia

There are several reports on the frequency of β thalassemia trait from different parts of the country, which vary from less than 1% to 17% (Madan et al., 2010). Most of the earlier studies are in small groups of hospital-based patients and/or population groups. It is well known that frequency is higher in some population groups (Verma, Saxena and Kohli, 2011). A study of 11 090 schoolchildren from Kolkata, Mumbai and New Delhi was carried out as an Indian Council of Medical Research (ICMR) Collaborative Study. The overall gene frequency of carriers of β thalassemia in Mumbai and New Delhi was 2.68% and 5.47% in the two cities, respectively. In Mumbai, the gene frequency was evenly distributed. The majority of children with β thalassemia from Mumbai were from Marathi (38.9%) and Gujarati (25.0%) speaking groups. Gene frequency was > 5% in Bhatias, Khatris, Lohanas and Schedule Castes. In New Delhi, a higher incidence was observed in schoolchildren of North and West Delhi (5.8–9.2%). These children were predominantly of Punjabi origin compared with children in the South of the city. Gene frequency of Hb D was greater in schoolchildren from New Delhi (1.1%) than in Mumbai (0.7%). Hb S trait (0.2%) was observed exclusively in children from Mumbai. A low incidence of Hb E trait (0.04%) was seen in children in Mumbai. A higher incidence was reported from Kolkata. The number of cases studied from the eastern region was small as the data from the East (Kolkata) could not be included in the analysis.

The mean frequency of β thalassaemia carriers in India is 3–4%. Colah et al. (2010) analysed these frequencies in different districts of Maharashtra and Gujarat in western India. A total of 18 651 individuals were screened for haemoglobinopathies and mutations were characterized in 1334 β thalassaemia heterozygotes. The distribution of frequencies was uneven, varying from 1.0% to 6.0% and 0% to 9.5% in different districts of the two states.

A large study on carrier screening was carried out by the Indian Red Cross Society of Gujarat (Maheshwari et al., 1999). Of the total 370 117 subjects screened, 173 112 were students, 45 000 were youths, and 8 377 pregnant women. The carrier rate of β thalassemia was 4.55–5.0% in the different groups.

An important project was carried out under the Prime Minister's Jai Vigyan Thalassemia Control Programme in six cities in India with a high prevalence of haemoglobinopathies: Mumbai (Maharashtra), Vadodora (Gujarat), Dibrugarh (Assam), Kolkata (West Bengal), Ludhiana (Punjab) and Bangalore (Karnataka). Screening was carried out in 29 898 college students and 26 916 pregnant women. The prevalence of β thalassaemia trait was 1.5–3.4% among college students, and 1.3–4.2% among pregnant women. A high frequency of carriers was observed among certain communities, like Vellalas, Sindhis, Aroras, Lohanas, Mandals, Pillais, Jains, Khatirs and Baidyas (Mohanty et al., 2008).

e. Other genetic disorders

Metabolic disorders

Metabolic studies in New Delhi (AIIMS and Sir Ganga Ram Hospital, Mumbai (KEM Hospital), and Bangalore (National Institute of Mental Health and Neurosciences) demonstrated that mucopolysaccharidoses, lysosomal storage disorders (Gaucher disease, Niemann-Pick disease and Pompe disease), Wilson disease, Glycogen Storage Disease and galactosaemia are quite common *(Bharucha*

et al., 1994). A screen of 112 269 neonates for amino acid disorders in the south Indian state of Karnataka showed four disorders to be the most prevalent: tyrosinaemia, maple syrup urine disease, PKU (with a combined frequency of 1 per 2495), and generalized amino acidurias with a frequency of 1 per 1605 (Appaji et al., 1988). In another hospital-based study, biochemical screening of 4400 cases of mental retardation revealed abnormalities of amino acids in 256 individuals (5.7%) (Kaur, Das and Verma, 1994). The four most common amino acid disorders were hyperglycinaemia, homocystinuria, alkaptonuria, and maple syrup urine disease.

Down syndrome

Data on the prevalence of malformations in various studies in India were subjected to meta-analysis, which gave a Down syndrome frequency of 1 in 916 (82 cases of Down syndrome in 75 103 births) *(Verma et al., 1998).* In the more recent three-centre study that specifically investigated Down syndrome, 1 per 1150 births was affected. All of these studies were clinical, although in the latter investigation the diagnosis of Down syndrome was confirmed by cytogenetic analysis. The number of Down syndrome cases born every year given in the March of Dimes report is an over estimate as it uses distribution of births by maternal age data of other countries which is inappropriate for India.

Fragile X syndrome

Fragile X syndrome is the second commonest cause of mental retardation, and in a study on mentally retarded persons, the prevalence was 2.8% in males and 1.8% among patients of both sexes. Among males with non-specific mental retardation, the prevalence was 5.8%. More recently, Fragile X syndrome was observed in 4.5% of children referred to hospitals with mental retardation, and in 2.8% of children attending special schools (Elango and Verma, 1996; Jain, Verma and Kapoor, 1998).

f. Disabilities

According to the 2001 Census, 21.9 million people live with disabilities in India, representing 2.13% of the total population. In 2002, the National Sample Survey Organization (NSSO) estimated the number of persons with disabilities to be 18.49 million, or 1.8% of the population *(Walia, 2010),* an estimate that is widely considered to be more accurate. Distribution by type of disability was: movement (locomotors) 51%, visual 14%, hearing 15%, speech 10%, and mental 10%. The prevalence is greater among males and in rural areas.

The legislative framework for the protection of the rights of disabled people is covered by four acts in India.

1. Mental Health Act 1987. This Act came into effect in all states and union territories in April 1993 and replaced the Indian Lunacy Act of 1912. It consolidated and amended the law to improve treatment and care of mentally ill persons and their property and affairs. Unfortunately it does not differentiate mental illness from mental retardation.

2. Rehabilitation Council of India Act 1992. This regulates the training of professionals in rehabilitation and sets out a framework for a Central Rehabilitation Register.

3. Persons with Disabilities Act 1995. This Act provides 3% reservations for persons with disabilities in poverty alleviation programmes, government posts, state educational facilities, as well as other rights and entitlement.

4. The National Trust Act 1999. This provides for the constitution of a national body for the welfare of persons with autism, cerebral palsy, mental retardation and multiple disabilities.

g. Consanguinity

As noted earlier, consanguineous marriages are common among Hindus in South India, and occur in some 20% of Hindu marriages in Maharashtra. However, unions between biological kin are uncommon in the northern, eastern and north-eastern states because of a general prohibition in the majority Hindu population, and thus occur in only 1–5% of the community. Consanguineous marriage is common in all Indian Muslim communities, without the north–south divide that exists in the Hindu population (Bittles, 2002). Consanguineous marriages are more common in rural areas and an increased incidence of autosomal recessive disorders is reported among those who are married consanguineously (Gulati, 2011).

h. Birth defects as per the March of Dimes Report

According to the March of Dimes Report, 1 722 404 infants are born each year with birth defects, i.e. 64.3 infants per 1000 live births (Christianson, Howson and Modell, 2006). Of these, 7.9 have cardiovascular defects, 4.7 NTDs, 1.2 some form of haemoglobinopathies, 1.6 Down syndrome, and 2.4 have G6PD deficiency.

3. Noncommunicable diseases

NCDs account for nearly half of all deaths in India. Among these, cardiovascular diseases (CVD) account for 52% of mortality, followed by chronic obstructive pulmonary disease, cancer, diabetes mellitus and injuries. NCDs account for 43% of disability-adjusted life years (DALYs) and projections show that without targeted interventions, the burden of NCDs will increase substantially. The potentially productive years of life lost due to CVDs in the age group 35–64 years was 9.2 million in 2000 and is expected to rise to 17.9 million in 2030 (University of Sydney et al., 2004). Since the majority of deaths are premature there is a substantial loss of lives during the productive years compared with other countries. Heart disease, stroke and diabetes are projected to increase cumulatively, and India stands to lose 237 billion dollars during the decade 2005–2015 (WHO, 2005).

Proposed programmes to control NCDs during the 12th Plan fall under three broad categories (MoH Government of India, 2011):

a) **Programmes for lifestyle, chronic diseases and risk factors: cancer, diabetes,** CVD and stroke, chronic obstructive pulmonary diseases, chronic kidney diseases, organ and tissue transplant, mental disorders, iodine deficiency disorders, fluorosis, oro-dental disorders.

(b) **Programmes for disability prevention and rehabilitation:** trauma (including road traffic accidents), burn injuries, disaster response, emergency medical services, musculoskeletal (bone and joint) disorders, physical medicine and rehabilitation, blindness, deafness, health care of the elderly (geriatric disorders), neurological disorders (epilepsy, autism), congenital diseases, hereditary blood disorders (sickle-cell anaemia, thalassemia, haemophilia).

(c) **Health promotion and prevention of NCDs:** tobacco control, prevention and management of nutritional disorders and obesity, National Institute for Health Promotion and Control of Chronic Diseases, patient safety programme, establishment of public health observatories.

[1]It is likely that the incidence of Down syndrome is overestimated as the percentage of women who deliver after the age of 35 is only 2.5%.

4. Country response to birth defects

a. Surveillance services

There is growing interest in starting Birth Defect Registries in India. In Chennai a Birth Defects Registry has existed for several years and currently receives data from 500 hospitals all over India. So far, 580 000 deliveries have been analysed and an alliance has recently been forged with the Federation of Obstetricians and Gynaecologists of India to extend coverage to other hospitals. In addition, the ICMR has plans to initiate a Birth Defects Registry in North India.

b. Genetic services

Various genetic services are available in all major towns. There are 54 genetic counselling centres, 40 genetic laboratories, and 20 prenatal diagnostic centres listed on the national web site (http://geneticsindia.org/). This listing is not complete as many laboratories have not yet registered. The number of laboratories offering various genetic tests is chromosomes (40), biochemical (26), and molecular (28).

The referral system for genetic services available in India, outlined in Table 11, is discussed in detail *by Puri and Verma (2012).*

c. Screening programmes

A major screening study for amino acid disorders in newborns was carried out by Appaji Rao and colleagues in Bangalore. In 2008 the ICMR approved a multi-centric study of newborn screening in five centres in India for two disorders (congenital hypothyroidism and congenital adrenal hyperplasia). This study has now been completed. The State of Goa has a mandatory screening programme for all newborns, while the states of Tamil Nadu, Chandigarh, Maharashtra and Andhra Pradesh have initiated pilot studies of newborn screening in some districts *(Kapoor and Kabra, 2010).*

d. Prevention programmes

For NTDs,a prevention programme is a necessity. However, as mentioned in the discussion on micronutrient fortification, only food distributed under the public distribution system is fortified with folic acid. This needs to be extended to flour being sold in the open market. In cities, most obstetricians prescribe folic acid to be taken 3 months before pregnancy and to be continued for 3 months after pregnancy. However, this remains limited to the small number of women who come for pre-pregnancy counselling.

For thalassemia and sickle-cell disease a widespread programme of screening is being carried out in Gujarat. The ICMR sponsored a study in six centres that screened 29 898 college students and 26 916 pregnant women for thalassemia carrier status and carried out prenatal diagnosis where necessary. This study showed that it is possible to mount a prevention programme in thalassemia in India. A study is being planned for control of thalassemia in the National Capital Territory of Delhi and the Union Territory of Chandigarh. In private hospitals most obstetricians screen pregnant women for carrier status of thalassemia.

For Down syndrome and other chromosomal disorders most women get screened by ultrasound during the 11th to 14th week of pregnancy by the first trimester scan measuring nuchal translucency. Biochemical screening during the first and second trimester is gaining momentum and a large number of women undergo these tests. Amniocentesis is easily available for women who are detected to be at high risk.

Since medical termination of pregnancy is legal in the event of an abnormal foetus, prenatal diagnosis of various genetic disorders is fairly popular. For other genetic disorders such as Duchenne muscular dystrophy, spinal

Table 11. Referral system for genetic disorders in India

Type of centre	Number	Services provided
Medical schools Tertiary care hospitals	350 Many	20 genetic centres Advanced facilities
District hospitals Community hospitals	237 4045	Genetic counselling, screening for thalassemia and medical disorders
Primary health centres (30 000 population)	23458	Identify and refer, advice
Sub-centres (5000 population)	146378	Identify and refer

muscular atrophy and fragile X syndrome, a number of centres are able to provide prenatal diagnosis by chorionic villus sampling and use of molecular techniques.

In some states routine measles-mumps-rubella (MMR) immunization is given. In private practice children are commonly given MMR vaccine, but not women. However various studies show that almost 85% of women entering pregnancy have IgG antibodies to rubella.

There is a strong commitment by the Government to ban tobacco use in public places by enacting legislation. Some states, such as Gujarat and Mizoram, have a blanket ban on selling alcohol.

e. Micronutrient supplementation and fortification programme

India was the first developing country to take up a National Nutritional Anaemia Prophylaxis Program (NNAP) to prevent anaemia among pregnant women in 1970 during the fourth 5-year health plan aiming to reduce the prevalence of anaemia to 25%. The IFA tablets containing 100 mg of Ferrous Sulphate and 500 mcg of Folic Acid is distributed to pregnant women a minimum of 100 days during pregnancy as well as during lactation.

Policy thrust for childhood anaemia is only recent. Preventative efforts have predominantly focused on increasing iron intake, primarily through supplementation. However, the implementation is very poor, only 3.8 – 4.7% of preschool children receive iron-folate supplements (Sachdev and Gera, 2013).

Since 2011, a programme for control of anaemia in adolescent girls has been introduced in thirteen states to reach out to 27.6 million adolescent girls of whom 16.3 million are school-going girls and 11.3 million are out-of-school girls (Aguayo et al, 2013). Building on the critical elements of and lessons learned from the programme, in 2012 the Government of India launched the national Weekly Iron and Folic Acid Supplementation (WIFS) programme to universalize the benefits of anaemia control to the overall population of Indian adolescents. Coverage of iron folic acid supplementation programme for adolescent is poor at present, but for pregnant women, coverage is about 65% (IIPS and Macro International, 2007).

There are no guidelines by any professional body to give folic acid during the periconception period. However in cities most obstetricians are aware that folic acid should be given before pregnancy to prevent NTDs, although no more than 5% of women currently come for pre-pregnancy counselling.

In 1962, the Government of India initiated a 100% centrally assisted National Goitre Control Program. A policy of Universal Salt Iodization (USI) has been adopted since 1983 to ensure that all edible salt for human and animal consumption is iodized. Under the Prevention of the Food adulteration Act (PFA), iodized salt is defined to have a minimum of 30 ppm iodine at the production level and a minimum of 15 ppm at the retail trader level. In 1992, the name of the program was changed to National Iodine Deficiency Disorders Control Program (NIDDCP). Recent country-wise surveys have revealed the success of the program: 86% of districts have median levels of urinary iodine at 100 mcg/L or greater, while 58% of districts had iodine content of salt at 15 ppm or greater at beneficiary level. The sustainability of activities of NIDDCP is vital for elimination of iodine deficiency in India (Kapil, 2010; Kapil and Sareen, 2012).

A number of states have fortification programmes for wheat flour from industrial mills distributed through the public distribution system – Andhra Pradesh, Chandigarh, Delhi, Gujarat, Kerala, Madhya Pradesh, Punjab, Rajasthan, Tamil Nadu, and West Bengal. Discussions regarding fortification are taking place among mills in the states of Bihar, Haryana, Karnataka, and Uttarakhand. One challenge is to expand the fortification programme to flour in the open market, and not just the public distribution system.

Given the wide variation in government engagement as well as cereal consumption, flour fortification policy is developed on a state-by-state basis. Gujarat has state regulations to fortify wheat flour with iron (ferrous sulphate, 30 ppm) and folic acid (1.5 ppm). State governments in Bihar and Madhya Pradesh are considering the Gujarat model along with the possibility of adopting mandatory fortification for the government-sponsored food distribution schemes. While current experience and future planning is based on the use of ferrous sulphate, high extraction (98%) atta flour, which possibly represents an 80% market share for flour in India, requires that a higher bioavailability compound such as NaFeEDTA (sodium iron ethylene diamine tetra acetic acid) should be used. Because fortification is under way with other compounds, there may be a need for an efficacy trial to provide additional evidence for policy-makers and implementers prior to changing iron fortificant compounds. Such a trial would provide an opportunity to add zinc and vitamin B12, important micronutrients given the likelihood of deficiencies among vegetarian populations.

5. Services for care of people with birth defects

For the management of birth defects many states in India are providing free operations for oral clefts, heart disease, talipes and other anomalies, in the government hospitals. There are good paediatric surgeons in India to carry out these surgeries, and in many states have also agreed to provide free factor VIII for treatment of haemophilia A, as well as the medicines required for the care of children with thalassemia.

Family support programmes exist for thalassemia, haemophilia, sickle-cell anaemia, cleft lip and talipes, fragile X syndrome, autism, cystic fibrosis, etc.

6. Further opportunities for prevention of birth defects

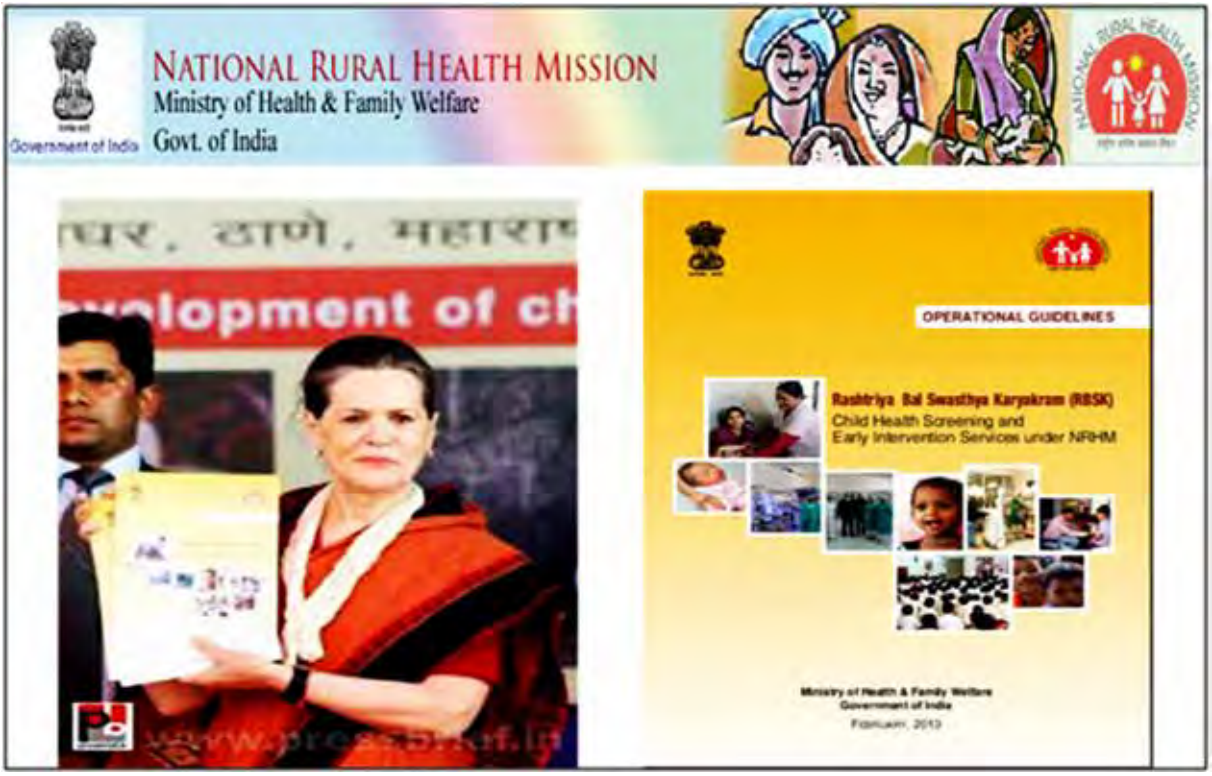
A welcome trend all over India is the increasing registration of births. For example, in Delhi all 325 000 annual births are registered online. It should be easy to modify the registration form for births to include information regarding the presence of birth defects, and their nature. This would be a good way to begin. Additionally the network of neonatal units could be used to record the congenital malformations observed and their contribution to morbidity and mortality.

The project on screening and prenatal diagnosis of thalassemia under the Prime Minister's Jai Vigyan Thalassemia Control Programme has provided the experience for mounting control programmes in the country. Such programmes should be initiated in states that have a high frequency of carriers.

The Planning Commission of the Government of India estimates that there is a 50% shortage of doctors in the country. The shortage of trained medical geneticists is 90%, and there is therefore a tremendous need to increase their number. Correspondingly the Medical Council of India plans to revise the curriculum for undergraduate and all postgraduate courses to include more education in genetics. A postgraduate programme in medical genetics has been approved both at Medical Doctor and Doctor of Medicine levels. The National Board of Examinations (NBE) plans to introduce a two-year fellowship programme for medical doctors. The Department of Biotechnology will initiate a National Course in Medical Genetics in conjunction with the NBE.

In view of the vast population, a practical solution for prevention of folic acid responsive birth defects (especially NTDs and cleft lip and cleft palate) could be food fortification with folic acid and other nutrients. Additional reasons are that many pregnancies are not planned and only a few women come during the preconception period. Based on the experience of food fortification in several states mandatory food fortification could be considered for scaling-up.

In February 2013, National Child Health Screening and Early Intervention Services were launched by Sonia Gandhi to cover 30 health conditions of the children aged 0-18 years through various approaches.



Identified Health Condition for Child Health Screening and Early Intervention Service

Defects at Birth

1. Neural Tube Defect
2. Down's Syndrome
3. Cleft Lip & palate/ cleft palate alone
4. Talipes (club foot)
5. Developmental Dysplasia of the Hip
6. Congenital Cataract
7. Congenital Deafness
8. Congenital Heart Diseases
9. Retinopathy of Prematurity

Deficiencies

10. Anaemia especially Severe Anaemia
11. Vitamin A Deficiency (Bitot spot)
12. Vitamin D Deficiency (Rickets)
13. Severe Acute Malnutrition
14. Goiter

Childhood Diseases

15. Skin Conditions (Scabies, fungal infection and Eczema)
16. Otitis Media
17. Rheumatic Heart Disease
18. Reactive Airway Disease
19. Dental Caries
20. Convulsive Disorders

Developmental Delays and Disabilities

21. Vision impairment
22. Hearing impairment
23. Neuro-motor impairment
24. Motor Delay
25. Cognitive Delay
26. Language Delay
27. Behaviour Disorder (Autism)
28 Learning Disorder
29. Attention Deficit Hyperactivity Disorder

30. Congenital Hypothyroidism, Sickle Cell Anaemia, Beta Thalassemia (optional)

INDONESIA

Indonesia

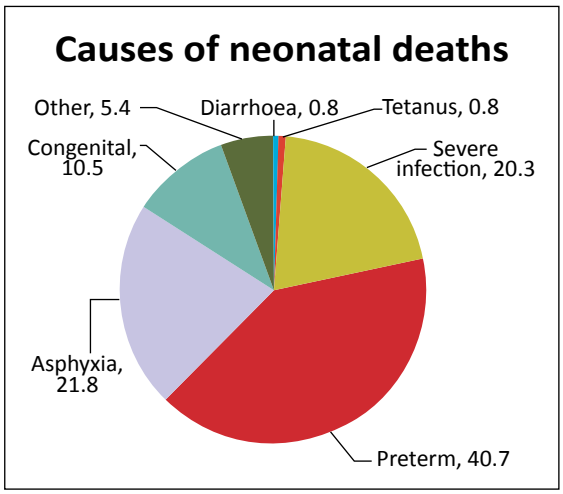
1. Demographic information

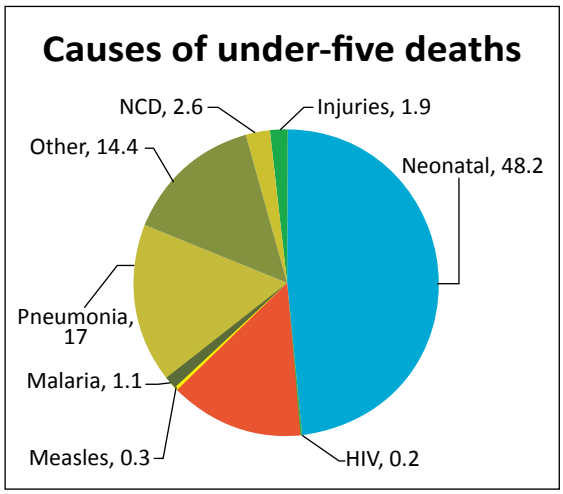
Total population ('000)	239 871
Estimated annual births ('000)	4 372
Under-five mortality rate (per 1000 live births)	32
Infant mortality rate (per 1000 live births)	25
Neonatal mortality rate (per 1000 live births)	15
Perinatal mortality rate (per 1000 live births)	25
Number of stillbirths	174
Complete immunization coverage by 12 months (%)	51
Complete immunization coverage at any time between 12–23 months (%)	59
Measles vaccination coverage by 12 months (%)	67
Measles vaccination coverage at any time between 12–23 months (%)	76
Vaccination against rubella	Yes
Antenatal care coverage (any) (%)	93
First ANC visit before 4 months (%)	75
ANC visits: 1 visit (%)	3
ANC visits: 4 or more (%)	82
Iron folic acid supplementation coverage (%)	77
Institutional deliveries (%)	46
Deliveries assisted by skilled birth attendants (%)	73

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *Statistics Indonesia (Badan Pusat Statistik—BPS) and Macro International. Indonesia Demographic and Health Survey 2007. Calverton, Maryland, USA: BPS and Macro International, 2008.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

a. Congenital malformations

Congenital malformations contribute to 1.4% of deaths in the age group 0–6 days of life; and to 19% of deaths in the age group 7–28 days *(MoH Republic of Indonesia, 2010).* Published articles are not easily accessible as they are published in a local paediatric journal. From a five-year evaluation among newborn infants born at Gunung Wenang Hospital in Manado, Indonesia *(Masloman, Mustadjab and Munir, 1991),* the total incidence of congenital malformations was 0.9%, 0.5% of which were major. The most common malformations were cleft lip and palate, talipes, multiple malformations, anal atresia, omphalocele and congenital heart diseases.

The 2007 National Basic health research *(Riskesdas)* has estimated that congenital malformations constitute 5.7% of infant mortality and 4.9% of under-five mortality *(MoH Republic of Indonesia, 2008).*

Neural tube defects

Green et al. (2007) reported on red cell folate concentrations in women of childbearing age in three cities. The values were highest ($P < 0.001$) in women from Jakarta at 872 nmol/L (95% CI 833, 910) followed by Kuala Lumpur at 674 nmol/L (95% CI 644, 704) and lowest in Beijing at 563 nmol/L (95% CI 524, 601). Accordingly, predicted NTD rates were highest in Beijing at 30/10 000 (95% CI 27, 33), followed by Kuala Lumpur at 24/10 000 (95% CI 22, 25), and lowest in Jakarta at 15/10 000 (95% CI 14, 15).

Bhattacharjee, Chakraborty and Purkaystha (2008) documented that fronto-ethmoidal encephalomeningocele is a rare congenital disease but occurs with a frequency of 1:5000 population in East Asian countries such as Indonesia, Malaysia, Myanmar and Thailand. Other types of NTDs do occur but their frequency is not known.

b. Perinatal infections

Data on perinatal infections is meagre.

c. Micronutrient deficiency disorders

Helen Keller International (HKI) reports that micronutrient deficiencies including iron and folate, iodine, and vitamin A are highly prevalent in Indonesia. HKI carried out the landmark research in the 1970s, establishing the link between vitamin A deficiency and childhood morbidity and mortality, as well as more recent work on the lower bioavailability of vitamin A from vegetables and fruits compared with animal products. HKI's vitamin A supplementation programme currently reaches 70% of children aged 12–59 months in Indonesia's most densely populated provinces.

In a study by the United States Agency for International Development (USAID) (2010) in Indonesia , 27.7% of children aged 1–4 are quoted to be anaemic. However, rates of anaemia as high as 50–85% have been reported among children 12–23 months in the poorest urban and rural areas (HKI, 2012). Vitamin A deficiency is common, being reported in 28.5% of children. Iodine deficiency was reported in 37.7% of children. The coverage of households use iodized salt is 62.3% and the median urinary iodine excretion is 229 µg/L (ICCIDD, 2012).

The Micronutrient Initiative is supporting a twice-yearly supplementation programme through local health services.

d. Thalassemia

This is a major genetic problem in the country. The genetic diversity of the population is reflected in the carrier frequency of β thalassemia (which ranges from 5–10%), haemoglobin E, Hb E, 1–33%), and α thalassemia (6–16%), in the various ethnic

populations. Likewise, the spectrum of underlying p-globin mutations varies widely between regions. Seventeen mutations account for more than 90% alleles in Indonesia. Hb E and IVS1-nt5 are found in all geographical areas, albeit with different frequencies, while other mutations are specific for particular ethnic groups such as Hb Malay and cd 26 (GAG TAG) in the Malay populations of Sumatara, IVS1-nt1 in the Sundanese and cd 35 in the Javanese (both of the Java island), and large β- and α-globin gene deletions in Buginese and Makasarese of South Sulawesi. The relationship between these mutations, their overall genetic background and their clinical expression are also of major interest. The findings have major implications in the management of thalassemia in Indonesia. Variation in the distribution of carriers in the various ethnic populations and the size of the populations resulted in unequal anticipated carrier screening and prenatal diagnosis workload. The diagnostic protocol for carrier screening and for prenatal diagnosis could be specifically designed on a regional basis.

The Government has not accorded a high priority to thalassemia treatment and control. Premarital genetic screening and counselling, as well as public education for community-based prevention and control models are used. Prenatal diagnosis is not carried out. Almost 2500 new thalassemic cases are born year *(Wahidayat et al, 2006).* Some problems in Indonesia are that: manpower is limited; medical treatment is not covered by health insurance; many patients come from poor families and cannot afford proper medical treatment; some parents are uneducated and have difficulties in communication; some live in remote areas far from the provincial general hospital, with limited access. Government support for thalassemia treatment (transfusion and oral chelator) and haemophilia is available in limited facilities on demand.

e. Other genetic disorders

Down syndrome

The Down Syndrome Association of Indonesia estimates that there are almost 300 000 cases in the country. The disorder is largely taboo in Indonesia, and living with the genetic condition can be difficult for those who have it, and their families. Chairperson of the Indonesian Down Syndrome Society (ISDI) and mother of a 19 year-old with Down syndrome, Aryanti Yacub, says having a child with this condition can be very difficult when there is a negative stigma attached to it in Indonesia. “You can feel the negativity on the street. People look differently at you, they try to stay away, in extreme cases they say nasty things, and you can always sense the hostility.”

G6PD deficiency is common, its prevalence varying from 2–14% in the population *(Suryantoro, 2003).*

f. Disabilities

According to the Indonesia Health Profile, the percentage of children aged 24–59 months with disabilities is as follows: blind, 0.09%; deaf, 0.08%; Down syndrome, 0.12%; cerebral palsy, 0.09%; and others, 0.11% (MoH Republic of Indonesia, 2010).

g. Consanguinity

No national data on consanguinity is available. However, *Glinka (1994)* surveyed 970 households in West Timor and reported a consanguinity rate of 17.8%.

h. Birth defects as per the March of Dimes Report

The March of Dimes Report on Birth Defects estimated that 260 090 children were born with birth defects annually in Indonesia *(Christianson, Howson and Modell, 2006).* These comprise 211 617 children with defects

of the cardiovascular system, 125 899 with NTD, 32 144 with haemoglobinopathies, 6 140 with Down syndrome, and 3 947 with G6PD deficiency.

3. Noncommunicable diseases

An epidemiological transition towards NCDs is becoming an additional challenge for Indonesia. This added burden of disease, associated with high levels of morbidity, is not limited to affluent populations in urban settings, but is also affecting poorer people, reducing their earning capacity and as such contributing to further impoverishment.

The 2007 Indonesian Basic Health Research *(Riskesdas)* showed that Indonesia has quite a high prevalence of NCDs in which hypertension accounts for 31.7% of total disease, followed by joint problems (30.3%); accident-related injuries (25.9%); stroke (8.3%); heart problems (7.2%); cancer and tumours (4.3%); asthma (3.5%); and diabetes mellitus (1.1%) (MoH Republic of Indonesia, 2008). Stroke accounts for 15.4% of the total leading causes of deaths, placing it as the most common cause of death among all age groups, followed by hypertension (6.8%); ischemic heart disease (5.1%); and other heart diseases (4.6%).

NCDs are heading to become a major public health problem, requiring sustained prevention and control of the risk factors involved. Indonesia has adopted WHO's global strategy in prevention and control of NCDs. However, the major challenge ahead will be to implement the strategy and to develop multisectoral public policies in support of the strategy.

4. Country response to birth defects

a) Surveillance programmes

There are no systematic birth defect surveillance activities in Indonesia. The South-East Asia Regional Neonatal-Perinatal database (SEAR Neonatal-Perinatal Database, WHO Report 2007–2008) reported that congenital malformations accounted for 19.61% of primary causes of neonatal deaths. Gastrointestinal, cardiac, Down syndrome, hydrocephalus and NTDs were the most common congenital malformations. In stillbirths in whom a cause could be established, malformations were found in 16.2% of the total cases (second most common cause).

b) Genetic services

Molecular genetic testing capabilities are well developed, largely because of the Eijkman Biology Molecular Institution, which has been in operation since 1990 and has facilities for advanced tests of molecular biology. It also conducts research in genetics in the country, although their medical application to genetic counselling and prenatal diagnosis is limited. There also appears to be a shortage of trained medical geneticists. A Master Degree of Genetic Counselling has been offered in the University of Diponegoro since 2007.

Cytogenetic studies are performed at Departments of Biology/Human Genetics in several universities. FISH (fluorescence in situ hybridization) studies for chromosomal disorders and micro deletion syndromes are available only in the Eijkmann Institute. Screening for congenital hypothyroid, G6PD deficiency and OAE (otoacoustic emissions) for testing hearing is performed in selected private hospitals.

c) Screening programmes

The following programmes are available in Indonesia.

- Antenatal screening exists for anaemia, haemoglobinopathies, infectious diseases like syphilis, HIV, rubella, cytomegalovirus (CMV), toxoplasmosis, Down syndrome, and NTDs. Ultrasound studies during pregnancy are not easily available.
- Newborn screening exists for congenital hypothyroid for 11 out of 33 provinces, but is not yet mandatory (Rustama et al., 2003).

d) Prevention programmes

The community health programmes related to birth defects prevention are as follows:

Maternal and child health handbook

Antenatal care includes the following:

- Iron and folic acid supplementation for the first 90 days of pregnancy, covering all pregnant women;
- Advice on avoidance of alcohol consumption, unnecessary traditional medication and tobacco smoking;
- Early detection and treatment of diseases in pregnancy (hypertension, anaemia, etc.).

Classes for mother

- Advice on avoidance of pregnancy after 35 years of age (Mother Class Programme);
- Advice on avoidance of medications/ X-rays/environmental hazards during early pregnancy (first trimester);
- Detection and treatment of diabetes in women before and during pregnancy
- Rubella vaccine programme for children and women of childbearing age.

e) Micronutrient fortification

Mandatory wheat flour fortification includes iron, zinc, folic acid, and other B vitamins, but the standards need to be revised in accordance with current WHO guidance for maximum public health impact. Based on data of national instant noodle consumption in Indonesia, estimated average flour consumption is 83.5 g/person per day.

The Micronutrient Initiative is working with the Government to achieve the following priority objectives:

- Provide twice-yearly vitamin A supplementation through local health services;
- Create models for the delivery of zinc supplements with oral rehydration salts for the treatment of childhood diarrhoea;
- Provide technical assistance to small-scale salt processors (salt farmers) in the Bima and Lombok districts of West Nusa Tenggara province to iodize salt;
- Help to develop viable models to expand iron and folic acid supplementation within the ANC programme in health centres, private clinics and hospitals to manage supplementation activity, support the education of women on the importance of supplementation, and assist with programme evaluation;
- Carry out a rice fortification pilot project coordinated by Bappenas (National Development Planning Agency) in collaboration with the Indonesia Fortification Coalition.

5. Services for care of people with birth defects

The following services are available within Indonesia.

- Family support programme
- Community-based rehabilitation programmes
- Houses for children with birth defects (e.g. YPAC, YTI)
- Corrective/rehabilitative surgery for spina bifida, cleft palate, atresia ani, etc.
- Special schools for children with disability (SLB)
- For care of thalassemia the cost of blood transfusions, chelation therapy and prenatal diagnosis; and for haemophilia various laboratory investigations, and factor VIII and IX as therapy are paid for by the Government for the poor with the JAMKESMAS (community insurance for poor people) PROGRAMME.

6. Further opportunities for prevention of birth defects

Indonesia has an infant mortality rate of 25/1000 live births, and neonatal mortality of 15/1000 live births, with a reasonable gross domestic product (GDP). Given the consanguinity and large number of births, the frequency of births with congenital malformations, especially heart disease and NTD, thalassemia and other genetic disorders, is high. More data need to be collected on the frequency of malformations and their contribution to morbidity and mortality in the neonatal period and during the first year of life. This can initially be done through neonatal units located in various hospitals. Eventually this should be developed into a National Registry of Birth Defects.

Folic acid and iodine supplementation are widely used in pregnant women. However, the maximum benefit of folic acid and other nutrients to reduce birth defects can only be achieved through fortification of food, to be decided by the Government. The country has accepted food fortification as a policy and this needs to be rigorously enforced.

The newborn screening for hypothyroidism that is being carried out in 11 out of 33 provinces should be extended to other areas, and in due course should be made mandatory.

The Government does extend support for thalassemia treatment (transfusion and oral chelators) and haemophilia (on demand), but a programme of screening prospective couples before marriage may be pursued. In due course hopefully the country may accept prenatal diagnosis (as in the Islamic Republic of Iran and Pakistan), and then pregnant women may be routinely screened for carrier status of thalassemia. Termination of pregnancy is permitted by law, but this has limited indications.

There is a tremendous shortage of geneticists and efforts should be made to train doctors in clinical genetics and genetic counselling. In addition, genetic counselling training can be given to nurses. The Eijkman Institute has excellent laboratory facilities but have limited clinical geneticists.

A parents' organization exists for Down syndrome as well as for thalassemia. Stakeholders involved in improving health services are governmental and nongovernmental agencies, the private sector, and academia for reproductive, maternal, newborn and child health (RMNCH), nutrition, immunization, and NCDs.

MALDIVES

Maldives

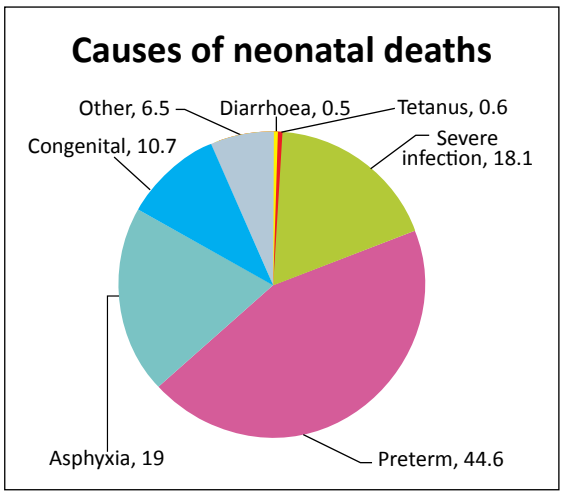
1. Demographic information

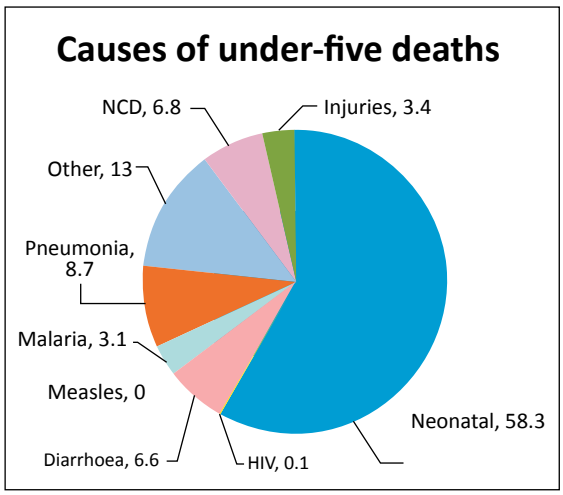
Total population ('000)	316
Estimated annual births ('000)	5
Under-five mortality rate (per 1000 live births)	11
Infant mortality rate (per 1000 live births)	9
Neonatal mortality rate (per 1000 live births)	7
Perinatal mortality rate (per 1000 live births)	18
Number of stillbirths	34
Complete immunization coverage by 12 months (%)	89
Complete immunization coverage at any time between 12–23 months (%)	93
Measles vaccination coverage by 12 months (%)	91
Measles vaccination coverage at any time between 12–23 months (%)	95
Vaccination against rubella:	Yes
Antenatal care coverage (any) (%)	99
First ANC visit before 4 months (%)	90
ANC visits: 1 visit (%)	0.3
ANC visits: 4 or more (%)	85
Iron folic acid supplementation coverage (%)	87
Institutional deliveries (%)	95
Deliveries assisted by skilled birth attendants (%)	95

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *Ministry of Health and Family (MOHF) Maldives and ICF Macro. Maldives Demographic and Health Survey 2009. Calverton, Maryland: MOHF and ICF Macro, 2010.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

Maldives is a small country with only 6000 births per year. The various mortality indicators are very good (under-five mortality 11; infant mortality rate 9; neonatal mortality rate 7), while the GDP is the highest in the SEA Region. The distribution of population over numerous small islands creates logistic problems. However it should be possible to introduce the recording of birth defects along with the registration of births. The excellent surveillance of communicable diseases can also incorporate collection of information on birth defects.

a. Congenital malformations

Remarkable progress has been made in the control of communicable diseases. Malaria has been eliminated from the country and other diseases such as leprosy and filariasis are now at the stage of zero transmission. In this scenario malformations could be important causes of morbidity and mortality. However due to inadequate attention to birth defects and genetic disorders there are hardly any data on these issues. Data on mortality notes that congenital malformations are responsible for 10.7% of neonatal deaths (WHO, 2010c).

b. Perinatal infections

The national laboratory at Indira Gandhi Memorial Hospital (IGMH) in Male recorded 55 patients who were rubella IgM positive from 1999 to 2000, with an epidemic peak in October 2000, but thereafter there have been only 2–3 cases. Rubella IgG antibody screening is conducted at IGMH for pregnant women with a history of miscarriage or stillbirth, and others deemed to be at high risk. Overall, 263 (68%) of 388 specimens from women aged 15–44 years were rubella IgG positive, indicating that 32% remained susceptible. Rubella vaccine was introduced in the immunization schedule in 2007.

Maldives has achieved Universal Child Immunization. The country remains polio free since 1980, with no recorded case of diphtheria or pertussis since 1982. The immunization coverage level with BCG, DTP-3, polio-4, measles-1, hep B-3, MR (introduced in 2006), MMR (introduced in 2007) remains above 90%.

c. Micronutrient deficiency disorders

In 2001, 50% of women in the reproductive age group and 56% of pregnant women were found to be anaemic. However, in 2005 there was not much improvement as anaemia was documented in 49.6% of non-pregnant women of reproductive age, while in preschool children was 81.5%. The proportion of population at risk of inadequate zinc was 13.75 (WHO, 2008).

As Maldives is a country of islands, it has been assumed that iodine deficiency disorders (IDD) are not a public health problem. In 1999 a survey of 30 schools and 2834 children aged 6 to 12 showed a total goitre rate of 23.6% with grade 1 goitre contributing 22.5% of this figure (Pandav et al., 1999). Of the households 44% were using iodized salt and the median of urinary iodine excretion is 115 µg/L *(ICCIDD, 2012).* In the WHO database, it is stated that in 2002 TGR was 25.7% (WH0, 2006b).

Although the country has already achieved five of the eight Millennium Development Goals, including the goal of reducing the number of people suffering from hunger, malnutrition among children is still a major public health problem. Nearly 17% of the children remain underweight and up to 19% suffer from stunting,11% are wasted and 3% are severely undernourished (MOHF Maldives and ICF Macro, 2010). However, this is an improvement over time as in 1997 stunting was reported in 36% and wasting in 45% of

children, while in 2004 stunting was present in 23% and wasting in 20% of children. Contributing factor to this problem is that not many children are exclusively breast-fed. Surveys have revealed that the average duration of exclusive breast feeding is less than two and half months *(Madhok, 2012).*

d. Thalassemia

The high prevalence of thalassemia continues to be a major challenge for the country. One out of every six persons is a thalassemia carrier and about 60 to 70 children are born every year with the disease. Data from the National Thalassemia Centre (2010) show that 716 patients with thalassemia major were registered, 535 were under treatment, and one bone marrow transplant had been done. Needless to say, significant efforts are focused on this disease. Intensive awareness campaigns, rigorous screening and improved treatment have increased the life expectancy of thalassemia sufferers.

Firdous, Gibbons and Modell (2011) reported that screening of 68 986 subjects born between 1960 and 1990 showed a carrier prevalence ranging from 10.1% to 28.2% among different atolls. This was related to the prevalence of falciparum malaria before eradication. There has been a steady fall in average carrier prevalence from 21.3% among those born in 1970 to 16% in those born in 1989. This observed fall was consistent with the predicted effect of malaria eradication, and corresponded to a fall in minimum affected birth prevalence from approximately 12/1000 in 1970 to approximately 6.9/1000 in 2007.

Carrier screening before marriage is encouraged to reduce the number of marriages between carrier couples. In 1999 a fatwa was passed allowing prenatal diagnosis and termination of a foetus affected with a serious disorder.

e. Other genetic disorders

Based on the age distribution of women at delivery (2009), 12% of women were 35 years or above *(MOHF Maldives and ICF Macro, 2010).* This should indicate a high incidence of Down syndrome. There are no published data on other genetic disorders but given that the population is Muslim, and consanguineous marriages are common in this community, a high rate of consanguinity autosomal recessive disorders is expected.

f. Disabilities

The first attempt to include disability questions by the Department of National Planning was by incorporating questions into the 2000 census questionnaire. The results showed that 1.7% of persons had disabilities. The limited questions did not provide sufficient information to formulate policies or programmes for policy-makers and researchers. The Ministry of Health conducted a disability survey in 2003, which showed that 3.4% of persons had a disability (UNESCAP, 2006). The 2006 census had no questions on disability.

A disability testing project (UNESCAP, 2009) examined 1000 persons, 500 from urban and 500 from rural areas. The results showed that impairment of vision was present in 19%, hearing in 8%, mobility in 20%, communication in 4%, cognition in 27%, and learning in 17% of participants.

g. Consanguinity

Consanguinity is expected to be high as the majority of the population is Muslim. However no studies are available to report the precise incidence.

h. Birth defects as per the March of Dimes Report

The March of Dimes Report on Birth Defects estimated that 365 children were born with birth defects annually in Maldives

(Christianson, Howson and Modell, 2006). These comprise 47 children with defects of the cardiovascular system, 12 with NTDs, 38 with haemoglobinopathies, 10 with Down syndrome and 10 with G6PD deficiency.

3. Noncommunicable diseases

Although significant achievements have been made in the control of communicable diseases, NCDs, particularly lifestyle-related diseases, pose a major challenge for health services. A large percentage of patients admitted to hospitals for NCDs suffered from ischemic heart diseases, cerebrovascular and hypertensive diseases. In 2001, 26% of deaths that occurred in IGMH were from diseases of the circulatory system. Physical disability, blindness, deafness and mental illness are the most common causes of disability. Patients seeking medical assistance from cancer are also increasing significantly. The majority of cancers were found to be cervical and breast cancer in women. Oral cancer is also high. In 2001, 14% of deaths in IGMH resulted from various neoplasms or cancers. In addition incidences of renal diseases, diabetes, gastrointestinal diseases and diseases of the nervous system and mental disorders were on the increase, with increasing deaths reported from such diseases. It is estimated that 43% of men and 11% of women use tobacco (WHO, 2012).The import of tobacco has remained high (tobacco leaves 32 827 kg, chewing tobacco 20 447 kg in 2010) and in 2010 about 39.9% of men and 9.6% of women were using tobacco.

4. Country response to birth defects

a. Surveillance programmes

As stated in the introduction, Maldives is a small country with good mortality indicators and a high GDP. Notwithstanding the logistic problems of its numerous small islands, it should be possible to record birth defects along with the registration of births and with surveillance for communicable diseases that has an excellent system.

b. Genetic services

Care of persons with thalassemia is well organized, and a central advanced haematologic and molecular genetics laboratory has been set up for the diagnosis of thalassemia. Prenatal diagnosis is permitted, as is termination of an affected foetus. There are no trained geneticists in the country.

c. Screening programmes

A screening programme for thalassemia is well established. Antenatal screening for anaemia and common infections is in place.

d. Prevention programmes

The immunization programme is highly successful. MMR vaccine has been introduced in the schedule since 2007.

e. Micronutrient fortification programmes

Maldives imports most of its flour and has little or no domestic milling industry. The wheat consumption, however, is high (179.57 g/capita/day). It should be possible to ensure that imported flour is fortified with folic acid and iron.

5. Services for care of people with birth defects

The Government has established facilities for the care of persons with disabilities of different types, varying from physical handicaps, deafness, blindness, mental handicaps, etc. Prominent organizations are the Maldivian Association for the Handicapped (MATH), the Society for the Deaf, Maldives Blind and Visual Impairment Society, and the Society for Health and Education (SHE), the latter working predominantly in the field of thalassemia.

6. Further opportunities for prevention of birth defects

The following issues should be pursued to reduce the incidence and consequences of birth defects in Indonesia.

- Thalassemia is a major problem with a very high frequency of carriers. A good central laboratory for molecular studies has been set up, and efforts should be vigorously pursued to start prenatal diagnosis in the country. A strict pre-marriage screening for thalassemia may also be introduced.
- The country imports the majority of its flour and has little or no domestic milling industry. Methods should be developed to ensure fortification of food with folic acid and other nutrients.
- Newborn screening for hypothyroidism should be made mandatory. Filter papers can be used to collect blood from heel pricks and posted to a central laboratory.
- At least one geneticist should be provided at the Indira Gandhi hospital in the capital. Due to consanguineous marriages and thalassemia this is necessary to provide genetic counselling to needy couples.
- The process of community organization and action has resulted in the establishment of Island Development Committees, Women's Development Committees, and Atoll Development Committees. These community groups have been instrumental in setting up drug cooperatives, raising funds for nutritional activities, and providing finances and labour for construction of health facilities, water tanks, etc. Community-based organizations such as youth clubs have been active in health areas like tobacco control and promotion of exercise at the island level.
- The Ministry of Health has identified the promotion of healthy lifestyles as a priority public health function. This is being implemented through multiple approaches including special School Health Programmes (e.g. Anti-Tobacco School Campaign) and multisectoral interventions (e.g. Sports for All) as well as the use of mass media and community-based interventions.

MYANMAR

Myanmar

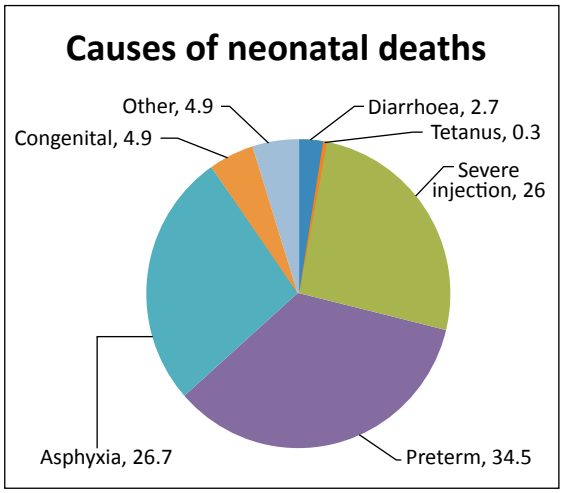
1. Demographic information

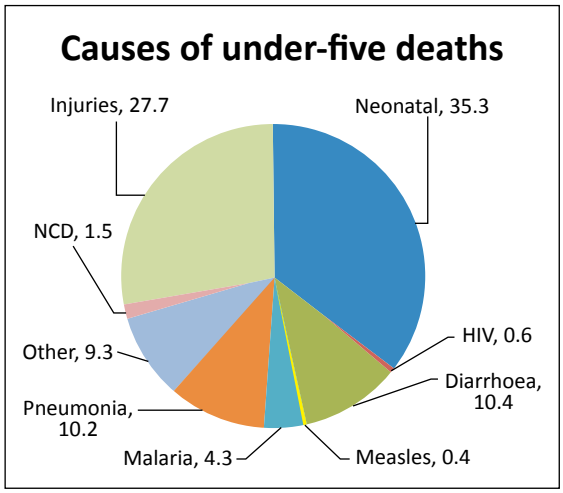
Total population ('000)	47 963
Estimated annual births ('000)	830
Under-five mortality rate (per 1000 live births)	62
Infant mortality rate (per 1000 live births)	48
Neonatal mortality rate (per 1000 live births)	30
Perinatal mortality rate (per 1000 live births)	No data
Number of stillbirths	No data
Complete immunization coverage by 12 months (%)	89
Complete immunization coverage at any time between 12–23 months (%)	97
Measles vaccination coverage by 12 months (%)	91
Measles vaccination coverage at any time between 12–23 months (%)	98
Vaccination against rubella:	No
Antenatal care coverage (any) (%)	93
First ANC visit before 4 months (%)	No data
ANC visits: 1 visit (%)	No data
ANC visits: 4 or more (%)	No data
Iron supplementation coverage (%)	84
Institutional deliveries (%)	36
Deliveries assisted by skilled birth attendants (%)	71

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *Ministry of National Planning and Economic Development and Ministry of Health Myanmar. Myanmar Multiple Indicator Cluster Survey 2009 - 2010 Final Report. Nay Pyi Taw: Ministry of National Planning and Economic Development and Ministry of Health, Myanmar, 2011.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

Registries for birth defects and malformations are present in hospitals like the Central Women's Hospital, Yangon. Diagnostic laboratories for inborn errors of metabolism and chromosomal disorders have not been established.

Congenital malformations contribute to 3% of neonatal mortality and 0.6% of morbidity in patients admitted to hospital (total number of admissions 6325 in 846 public hospitals) (Department of Health Planning and Department of Health MoH Myanmar, 2008).

a. Congenital malformations

The most common birth defects are haemoglobinopathies, malformation of circulatory system and GI tract, cleft lip and/or palate, Down syndrome, G6PD deficiency and NTDs including spina bifida.

b. Perinatal infections

Thant et al. (2006) remarked that rubella vaccine was not included in the immunization schedule in Myanmar. Although surveillance for outbreaks of measles and rubella is conducted nationwide, there is no routine surveillance for congenital rubella syndrome (CRS). From 1 December 2000 to 31 December 2002 active surveillance for CRS was conducted among children aged 0–17 months at 13 hospitals and 2 private clinics in Yangon, the capital city. Eighty-one children aged 0–17 months were suspected of having CRS. Of these, 18 had laboratory-confirmed CRS (7 were IgM positive; 7 were reverse transcription polymerase chain reaction (RT-PCR) positive; and 10 were IgG positive at > 6 months of age). One additional child who tested positive by RT-PCR and whose mother had rubella during pregnancy but who had a normal clinical examination was classified as having congenital rubella infection. During 2001–2002 no rubella outbreaks were detected in Yangon Division. In the 31 urban townships of Yangon Division, the annual incidence was found to be 0.1 laboratory-confirmed cases of CRS per 1000 live births.

c. Micronutrient deficiency disorders

Protein energy malnutrition (PEM) and micronutrient deficiencies (iron deficiency anaemia, iodine deficiency disorders, and vitamin A deficiency) have been identified as major nutritional problems. Interventions against these problems have always been targeted at the two most vulnerable age groups, namely pregnant women and children below five years of age. Growth monitoring and promotion (GM/P) for children below 3 years of age is the major PEM control activity taking place through the country. There are also nutrition rehabilitation activities in some selected urban and rural areas. Iron supplementation is the nationwide programme against anaemia during pregnancy, while supplementation for under-five children and adolescent school girls is also implemented in some areas. Universal salt iodization has been adopted for sustained elimination of iodine deficiency disorders while biannual supplementation with high potency vitamin A capsules forms the major intervention against vitamin A deficiency. The coverage of households using iodized salt is high, more than 92%, while the median urinary iodine excretion is 216 µg/L (ICCIDD, 2012).

d. Thalassemia

Harano, Win and Harano (2000) investigated 112 DNAs (deoxyribonucleic acids) from transfusion-dependent thalassemic children in Myanmar. Homozygous Hb E was detected in 66 cases. Homozygotes of CD41/42 (-TCTT) deletion were observed in 19 cases, homozygous IVS

I-1 (G>T) in 4, and homozygous IVS I-5 (G>C) in 4 cases. Sixty-two cases were of compound heterozygotes. The above four mutations along with additional two mutations of CD 17 (A>T) and IVS II-654 (C>T) are common in Myanmar.

Win et al. (2002) examined 158 unrelated Myanmar patients (107 Hb E-β thalassemia and 51 β thalassemia major) for mutations in the β-globin gene mutations. IVSI-1(G>T), codon41/42(-TCTT), IVSI-5(G>C), codon 17(A>T), IVS II-654(C>T), and -28Cap (A>G) were detected in 166/209 (79.4%) alleles. DNA sequencing of 24 alleles from 43ARMS-negative samples (20.6%) identified an additional 12 new mutations, to produce a total of 18 different mutations. Nineteen alleles (9.1%) remained for further characterization. The molecular spectrum of Myanmar β thal is wider and more heterogeneous than previously reported.

Than et al. (2005)analysed samples from 916 members of various ethnic groups from malaria-endemic groups in the southern Shan State for α thalassemia (α thal), β thalassemia (β thal), abnormal haemoglobin variants, and G6PD deficiency. Of these subjects, 530 (57.9%) were found to have at least one of these red cell genetic disorders. The overall frequencies for the various red cell genetic disorders were as follows: α thal, 37.5% (343/916); Hb E, 20.3% (186/916); G6PD-Mahidol, 17.5% (160/916); and β thal, 0.3% (3/916). The Bamar population showed the highest frequencies of α thal (56.9%, 177/311), Hb E (28.3%, 88/311), and G6PD-Mahidol (21.2%, 66/311). Their results showed that race was the dominant factor affecting the frequencies of red cell genetic disorders in malaria-endemic areas of Myanmar.

Win et al. (2005) remarked that the population of Myanmar comprises eight major indigenous races *(Bamar, Chin, Kachin, Kayah, Kayin, Mon, Rakhine and Shan)*. The Bamar reside in the seven central divisions of the country, and the others reside in the seven peripheral states that border neighbouring countries, including China, Laos, and Thailand in the east and Bangladesh and India in the west. Both malaria and Hb E are endemic in Myanmar, although the actual prevalence of the latter in different indigenous races is not yet known. In the four malaria-endemic villages, each having a different predominating indigenous race, the overall prevalence of Hb E was 11.4% (52/456 villagers), ranging from 2–6% in the Kayin-predominant villages to 13.1–24.4% in the Bamar-predominant villages.

Ne-Win et al. (2006) tested 3 common α thal deletions (-α3.7, -α4.2and SEA) in 170 unrelated Myanmar thalassemia patients receiving transfusions. Thalassemia deletions were detected in 27 patients (15.9%) as: (1) α thal-2 (-α3.7/αα) in 12 heterozygous or Hb E-β thal cases; (2) α thal-1 in 7 patients (2-α3.7/-α3.7 and 5 --SEA/αα); and (3) Hb H (-α3.7/--SEA) in 8 patients. The latter 15 α thal-1 and Hb H patients had no β thal mutations and represented 8.8% of the overall patients seeking transfusion for refractory anaemia. This is the first description of α thalassemia in Myanmar from the molecular aspect, and its clinical and racial heterogeneity are described and discussed. The frequency of β thalassemia is about 4.3% in Myanmar (Harano, 2000).

e. Other genetic disorders

No data are available on other genetic disorders.

f. Disabilities

The First Myanmar National Disability Survey was conducted by the Department of Social Welfare (DSW) and The Leprosy Mission International (TLMI) between 2008–2009 (DSW and TLMI, 2010). According to this survey, 2.3% of Myanmar's population has some form of disability. Of these, 68.2%

are persons with physical impairment, 13.3% have visual impairment, 10.4% have impaired hearing and 8.1% have some form of intellectual disability.

Bawi (2012) interviewed disabled persons in three commercial centres of Yangon, Mandalay and Taunggyi and identified the following challenges for their care and rehabilitation: lack of training facilities for independent living, vocational training, supporting organizations, and institutions for the visually and hearing impaired.

g. Consanguinity

No data are available.

h. Birth defects as per the March of Dimes Report

The March of Dimes Report on Birth Defects estimated that 59 436 children were born with birth defects annually in Myanmar (Christianson, Howson and Modell, 2006). These comprise 8026 with defects of the cardiovascular system, 711 with NTDs, 4064 with haemoglobinopathies, 1727 with Down syndrome and 3150 with G6PD deficiency.

3. Noncommunicable diseases

The Ministry of Health has been implementing various NCD projects such as on: accident prevention, cancer control, cardiovascular disease control, diabetes control, drug abuse control, mental health, nutrition promotion, and tobacco control. Project activities consist mainly of public health education through various types of mass media communication such as radio, television, newspaper, magazines, pamphlets, posters, and logos. For tobacco control, the Control of Smoking and Consumption of Tobacco Products Law was adopted in 2006 and came into effect in 2007. This act prohibits smoking in public places and on public transport, the sale of tobacco to and by minors, and all forms of tobacco advertisement.

4. Country response to birth defects

Essential newborn care was established in 2006, and the Integrated Management of Maternal, Newborn and Child initiative in 1998. The Women and Child Health Development scheme is in operation since 2001, and the Infant and Young Child Feeding scheme since 2006. There are national policies on NCDs, reproductive health and adolescent health.

a. Surveillance programmes

Surveillance programmes exist for communicable diseases, but there are none for birth defects, except in large hospitals where register is maintained for newborn morbidity and mortality.

b. Genetic services

There are no medical geneticists, or genetic laboratories. Genetic counselling is carried out by neonatologists and paediatricians.

Currently, prenatal diagnosis of genetic disorders is not carried out. Termination of pregnancy is not permitted by law but is allowed in case life of the mother is in danger because of medical conditions, and at the recommendation of a medical board meeting of administrators, obstetricians, gynaecologists, neonatologists and radiologists.

c. Screening programmes

Antenatal screening programmes exist for anaemia, infectious diseases (especially syphilis), rubella, CMV, and toxoplasmosis. Ultrasound studies are carried out for prenatal diagnosis of malformations. There is no carrier screening for haemoglobinopathies, thyroid disease or Down syndrome. These are however available in private laboratories. Newborn screening programmes are not available.

d. Prevention programmes

Folic acid and iron tablets are given to adolescent girls twice a week in 20 townships. The same tablets are given to pregnant women once daily for 6 months and have 84% coverage over the country.

Genetic counselling is provided by neonatologists and paediatricians. Ultrasound facilities are available in tertiary hospitals as part of ANC. They are used frequently for prenatal diagnosis of malformations/ chromosomal disease, but not for other prenatal diagnostic procedures.

The rubella vaccine programme for children and women of childbearing age is only provided in the private sector.

Advice regarding non-use of alcohol among women during the periconceptional period and pregnancy is given through health education. Harmful use of tobacco(which includes use and exposure during pregnancy) is also emphasized through health education (Figure 7).

Avoidance of medications/x-rays/environmental hazards during early pregnancy is advised.

Detection and treatment of diabetes in women before and during pregnancy is readily available.

Figure 7. Poster emphasizing harmful use of tobacco

e. Micronutrient fortification programmes

Preventive strategies in place are preconception folic acid supplementation, adolescent anaemia control programme, folic acid and iron supplementation to all pregnant women, iodine fortification, and rubella immunization. Several wheat flour millers have expressed interest in food fortification. The country, however, has low wheat flour consumption.

5. Services for care of people with birth defects

The mean prevalence of β thalassemia trait is 4.3% (Aung Than Batu et al., 1968). The cost of blood transfusion and chelation are shared by the Government and the patients. Blood transfusion is carried out in day-care centres (Yangon and Mandalay Children Hospital).

- For haemophilia laboratory investigations, factor VIII and IX as therapy are made available but limited to large cities.
- Family support programmes and networks exist in hospitals.
- Community-based rehabilitation programmes have been established.
- Parent associations exist for cleft lip and palate, autism, and cancer patients.
- Corrective/rehabilitative surgery is available in hospitals.
- A special education and physiotherapy centre, as well as school and day-care centres for disabled children have been established.
- Nongovernment stakeholders include the Myanmar Maternal and Child Welfare Association, Myanmar Medical Association, Médecins Sans Frontières, Malteser, Japanese Organization for International Cooperation in Family Planning, Association Francois-Xavier Bagnoud, Private Sector, MSI, Save the Children, Merlin, World Vision, Joint Initiative on Mothers, Newborn, and Child Health (International Organization for Migration), CDA and United Nations agencies such as WHO, United Nations Children's Fund (UNICEF) and the United Nations Population Fund (UNFPA).

6. Further opportunities for prevention of birth defects

To improve the health situation in Myanmar, perinatal and neonatal databases should be created in all hospitals. These will provide information on the common malformations and birth defects present in the country.

In areas where thalassemia trait has a high frequency, screening of pregnant women should be initiated, with follow-up screening of husbands of women who are diagnosed as carriers. This will reduce burden of thalassemia in the country.

A department of genetics should be set up in the capital with a molecular laboratory to carry out mutation studies in cases of thalassemia. Based on this effort, a control programme for thalassemia can be developed.

Annual gathering of parents of cancer patients and parent associations for children with autism, and cleft lip and palate, could be expanded.

Fortification of flour and rice with micronutrients and folic acid will go a long way in reducing neural tube defects and other folic-acid responsive birth defects.

Legislation in place to prohibit the sale of alcohol to persons under 18 years old, and for harmful use of tobacco, should be further promoted.

The following stakeholders of birth defects prevention should be used as a platform to enhance awareness and action on the issue: Ministry of Health divisions– maternal and child health, women and child health development, nutrition, school health, Central Health Education Bureau , National AIDS program, Expanded program on Immunization, Central Epidemiology unit, National Health Lab, medical care (NCD, tobacco, etc.), clinicians (obstetric gynaecologists, neonatologists, paediatricians, surgeons, radiologists, pathologists, etc.), Department of Health planning (HMIS), Department of medical sciences, Department of medical research, and Ministries of Mines, Social Welfare, Relief and Resettlement and related organizations.

NEPAL

Nepal

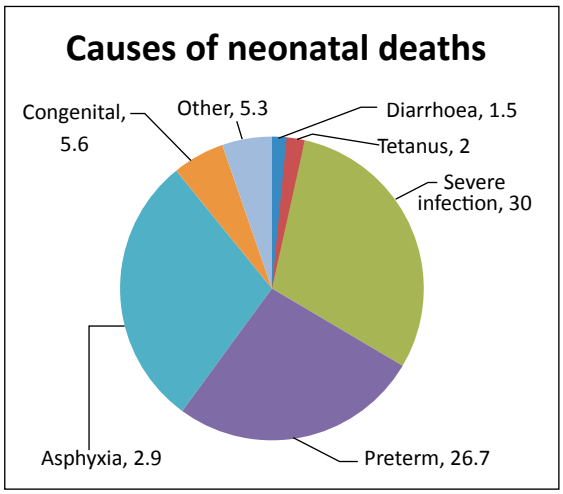
1. Demographic information

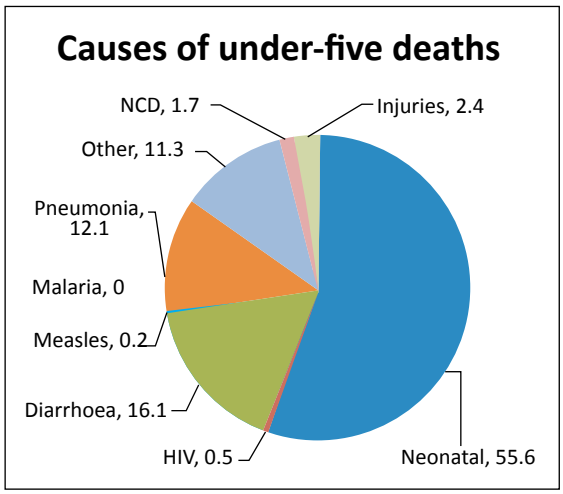
Total population ('000)	29 959
Estimated annual births ('000)	724
Under-five mortality rate (per 1000 live births)	48
Infant mortality rate (per 1000 live births)	39
Neonatal mortality rate (per 1000 live births)	27
Perinatal mortality rate (per 1000 live births)	37
Number of stillbirths	53
Complete immunization coverage by 12 months (%)	80.7
Complete immunization coverage at any time between 12–23 months (%)	87
Measles vaccination coverage by 12 months (%)	82.3
Measles vaccination coverage at any time between 12–23 months (%)	88
Vaccination against rubella:	No
Antenatal care coverage (any) (%)	58.3
First ANC visit before 4 months (%)	49.7
ANC visits: one visit (%)	6.1
ANC visits: four or more (%)	50
Iron folic acid supplementation coverage (%)	79.5
Institutional deliveries (%)	35.3
Deliveries assisted by skilled birth attendants (%)	36

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *Ministry of Health and Population (MOHP) Nepal, New ERA, and ICF International Inc. Nepal Demographic and Health Survey 2011. Kathmandu, Nepal: Ministry of Health and Population, New ERA, and ICF International, Calverton, Maryland, 2012.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

a. Congenital malformations

Peto et al (unpublished, 2012) conducted a community-based birth defects surveillance from 1994-1997 during an antenatal micronutrient supplementation field trial in the Sarlahi, a terai district of Nepal (JiVitA Project, Johns Hopkins University). A total of 15 403 infants born to mothers who participated in the trials were screened by the trained field staff, of which 12% were identified to have abnormalities. Subsequently, 1.1%(n=186) of all liveborn infants were confirmed to have external birth abnormalities by the research physicians. Abnormalities affecting the highest proportions of infants included umbilical hernias, poly/syndactyly, facial clefts and club foot.

Christian et al. (2004) studied a total of 17 767 women, who were pregnant at least once during 3.5 years of the study. Out of this cohort, 30% reported smoking during pregnancy. Cigarette smoking during pregnancy is associated with an increased risk of maternal and infant mortality in rural Nepal. -carotene and to some extent vitamin A may reduce the risk of pregnancy-related mortality, but not infant mortality, among both smokers and non-smokers.

Malla (2007) conducted a preliminary study at the Maternity Hospital. Congenital malformations accounted for 9.7% of perinatal deaths, 11.06% of early neonatal deaths and 7.9% of stillbirths. Of 16 948 total births, 75 (0.36%) had congenital malformations. Of 75 infants with malformations, 30 had central nervous system defects, including 20 cases of spina bifida and anencephaly.

Manandhar et al. (2010) identified births and neonatal deaths in 60 village development committees of Dhanusha district. Families were interviewed at six weeks after delivery, using a structured questionnaire. Cause of death was assigned independently by two paediatricians according to a predefined algorithm; verbal autopsies were available for 601/813 stillbirths and 671/954 neonatal deaths. The perinatal mortality rate was 60 per 1000 births and the neonatal mortality rate 38 per 1000 live births. The three leading causes of neonatal death were birth asphyxia (37%), severe infection (30%) and prematurity or low birth weight (15%). Congenital malformations occurred in 3% of stillbirths and 1% of neonatal deaths.

Pradhan, Poudel and Maharjan (2010) undertook a retrospective review of the medical records of 89 women with singleton pregnancy who had stillbirth at or more than 28 weeks gestation during the period from April 1998 to April 2007 at Nepal Medical College Teaching Hospital. Stillborn infants accounted for more than 50% of perinatal deaths in Nepal. Major malformations were present in 5 (5.6%) of 89 infants including 3 infants with NTDs.

The commonest malformations in the country are cleft lip and palate, NTDs and congenital heart diseases. The South-East Asia Regional Neonatal Perinatal database (SEAR NPD/WHO Report, 2007–2008) reported that congenital malformations accounted for 11.1%of primary causes of neonatal death. Cleft lip/ palate and cardiac defects were found to be the commonest congenital malformations. In stillbirths in whom a cause could be established, malformations were found in 10.9% of cases (second most common cause). In 47.8% cases, the cause could not be established.

b. Perinatal infections

Upreti et al. (2011) analysed cases of rubella reported through measles surveillance during

2004–2009 period. Rubella sero prevalence among women 15–39 years of age in 2008 was evaluated, and children attending a school for the deaf in 2009 were examined for ocular defects associated with CRS. During 2004–2009, there were 3710 confirmed rubella cases and more than 95% of these cases were less than 15 years of age. Of the 2224 women of childbearing age tested for anti-rubella IgG, 2020 (90.8%) were seropositive. Using a catalytic infection model, 1426 infants were born with CRS (192/100 000 live births) in 2008. Among 243 students attending a school for the deaf, 18 (7.4%) met the clinical criteria for CRS. Rubella and CRS were documented as significant public health problems in Nepal. The authors advocated introduction of rubella vaccine in the national programme.

c. Micronutrient deficiency disorders

Micronutrient Initiative estimated that 65% of children below 5 years suffer from iron deficiency anaemia. Around 200,000 children annually were born with intellectual impairment due to iodine deficiency. Thirty three percent of preschool children have sub-clinical vitamin A deficiency (The Micronutrient Initiative and UNICEF, 2004b).

The 2011 Nepal Demographic and Health Survey (NDHS) found 41%, 29% and 11% of children aged less than 5 years are stunted, wasted and underweight, respectively. This represents an improvement over the findings in 2006 (49%, 39% and 13%, respectively). Coverage of households use adequately iodized salt has improved, from 58% in 2005 to 80% in 2011. Iodized salt is considered adequate when it contains of 15 parts of iodine per million part of salt (15 ppm) (MOHP Nepal, New ERA, and ICF International Inc., 2012).

d. Thalassemia

There is high frequency of α thalassemia among some Nepalese populations in the lowland area of Nepal bordering India (Sakai et al., 2000). These areas are endemic for malaria. For example, in Danuwar the frequency of α thalassemia is 79.4% and in Newar 20.5% (Modiano et al., 1999).The Thalassemia International Federation (2003) lists the frequency of β thalassemia carrier status as 4%, and estimates that 327 homozygotes of thalassemia major are born every year. Red Cross blood banks have been providing free blood transfusions to thalassemics in Nepal since 1996, while the Nepalese Thalassemia Society was formed in 2003.

e. Other genetic disorders

Shrestha et al. (2009) carried out chromosomal studies in 30 children selected from patients attending the genetic clinic, Department of Paediatrics, B.P. Koirala Institute of Health Sciences presenting with dysmorphic features, mental retardation, short stature, congenital malformations or ambiguous genitalia, between 0–15 years of age. Chromosomal disorders were identified in 10 (33.34%) children. The most common chromosomal abnormality was Down syndrome (26.67%) followed by Turner syndrome (6.67%).

The percentage of women older than 35 years at the time of delivery was about 7% (MoHP Nepal, New Era, and ICF International Inc., 2012),where there are about 724 000 births per year (UNICEF, 2012).

The top single-gene disorders in the country are thalassemia, haemophilia, and Duchenne muscular dystrophy.

f. Disabilities

A situation analysis on disability conducted in Nepal in 1999 (JICA, 2002) showed that physical disability was present in 28.5% of

a sample survey. Impairment of vision was present in 7.3%, hearing in 13%, intellectual ability in 10%, and psychiatric status in 15.2%.

Organizations dealing with disability are listed in the relevant sections below.

g. Consanguinity

Consanguinity is recorded in about 15% of the population within some ethnic groups. Most of these are marriages between first cousins.

h. Birth defects as per the March of Dimes Report

The March of Dimes Report on Birth Defects estimated that 43 727 children were born with birth defects annually in Nepal *(Christianson, Howson and Modell, 2006).* They comprise of 5767 children with defects of the cardiovascular system, 3431 with NTDs, 146 with haemoglobinopathies, 1533 with Down syndrome, and 2482 with G6PD deficiency.

3. Noncommunicable diseases

NCDs have become increasingly prominent in recent years. In 2011, 82% of outpatient visits and 88% of inpatient cases in the government institutions were due to NCDs (World Bank, 2011). The Government has formulated a plan to tackle these disorders, including the control of thalassemia.

4. Country response to birth defects

a. Surveillance programmes

There is a need to initiate a perinatal and neonatal database in hospitals where deliveries take place. This will provide data on the prevalence of birth defects and their contribution to morbidity and mortality. National birth defects surveillance registry has not been established.

b. Genetic services

- There is only one genetic clinic run in Dharan under the auspices of the Department of Paediatrics, where a register of cases seen is maintained. The paediatrician in charge has received training in genetics. There are no other medical geneticists, or departments of medical genetics.

- It would be useful to set up a department of medical genetics in the capital, because of the large number of patients with genetic disorders who have to go to India for examination and treatment. Currently genetic counselling is provided by neonatologists and paediatricians who identify cases with birth defects.

- Laboratory services. The National Public Health Laboratory is a nodal institute for developing policy, guidelines and an overall framework for capacity-building in the laboratory sector. Attention has been given to strengthening laboratory procedures and communication between national, regional and district levels and strengthening the system to ensure the availability of essential equipment, logistics and human resources. At present there are eight central hospital-based laboratories, three regional hospital-based laboratories, two subregional hospital-based laboratories, 11 zonal hospital-based laboratories, 66 district hospital-based laboratories, and 204 primary health care centre- based laboratories in the country.

c. Screening programmes

No information is available on specific screening programmes.

d. Prevention programmes

The National Anaemia Control Strategy and Iron Intensification Programme was developed in 2003 to increase the coverage and compliance of iron supplementation along with complementary measures such as deworming of pregnant women and dietary diversification, flour fortification (in small scale), and promotion of maternal care practices. With the support from Micronutrient Initiative, UNICEF and WHO, the programme was started in five districts and has expanded to 46 districts by 2007. The government aimed to cover all 75 districts by 2010. To improve access, female community health volunteers were trained to di piloted, whereby school children were mobilized to ensure early identification of pregnant women. Because of these improvements and other complementary measures, anaemia in pregnant women has been reduced from 75% to 42% *(The Micronutrient Initiative, 2009).*

Fortification of flour has been introduced since 2008 and this is likely to lead to reduction in neural tube defects. This was initiated as a World Bank Development project that helped village millers in rural Nepal add essential vitamins and minerals to the cereal flour they produce. The addition of key nutrient is helping to improve the health and productivity of rural Nepalese by decreasing iron deficiency anaemia – a widespread public health problem *(Micronutrient Initiative, 2007).*

Universal Salt Iodization (USI) is in operation since 1998 under a five year Plan of Action for Control of IDD (1998-2003) in collaboration with UNICEF and JICA (MOHP, MI and New Era, 2005). Four in five households in Nepal use adequately iodized salt.

There is no special programme during the preconception period except the folic acid/ iron given to adolescent girls. In hospitals women are advised to take preconceptional folic acid as a common clinical practice.

e. Micronutrient fortification with folic acid

With a view to addressing micronutrient deficiency in Nepali people, the Ministry of Health and Population is producing fortified flour containing vitamin A, iron and folic acid. Each 1 kg of such flour contains 1 mg vitamin A, 60 mg iron and 1.5 mg folic acid. Since flour is widely used in Nepali households, the Government awarded contracts to 22 flour mills in the Tarai region to produce fortified flour. The main objective of the Government is to combat diseases like anaemia, night blindness and vitamin deficiency. Its effect on the prevalence of birth defects should be monitored.

Universal use of iodized salt is recommended.

5. Services for care of people with birth defects

There are several disability-related governmental organizations: National Federation of the Disabled Nepal (NFD); Nepal Disabled Association (NDA) (Kagendra New Life Center); National Disabled Society (NDS); National Association for the Welfare of the Blind (NAWB); National Association of the Blind (NAB); Nepal Netra Jyoti Sangh (NNJS); Nepal National Federation of Deaf and Hard of Hearing (NFDH); National Association of the Deaf and Hard of Hearing (NADH); Nepal Ear Foundation; and Association for the Welfare of the Mentally Retarded (AWMR).

Disability-related nongovernmental organizations (NGOs) comprise His Majesty's Government of Nepal/WHO Nepal Prevention and Control of Blindness/Deafness Project; and Muscular

Dystrophy Association of Nepal. There are many organizations for the deaf (Kathmandu Association of the Deaf; School for the Deaf, Naxal; Nepal National Federation of Parents of Deaf People; Siraha Association of the Deaf; Bhaktapur Deaf Association; Kirtipur Deaf Club; Makwanpur Deaf Association; Pyuthan Deaf Association; and Gorkha Association of the Deaf).

Organizations for people with poor locomotion are: Disability Apang Sarokar Griha; Disabled Newlife Centre; INF Release Project, Green Pastures Hospital; Orthopedica (Special Center for Orthotic and Prosthetics); Nepal Leprosy Relief Association; Nepal Leprosy Trust; Association for IDEA Nepal; and Partnership for Rehabilitation Program RELEASE Project.

Organizations for people with mental retardation are: Navjyoti Center;SUNGAVA (Mentally Disabled Women's Vocational Training Center); Mental Retardation Welfare Center; Manav Bidha Grih; Western Regional Community Mental Health Program; Asha Bal Kendra; Sustha Manasthiti Kalyan Sanstha; Sustha Manasthiti Sewa Kendra; and Shisu Bikas Kendra.

An organization for people with cerebral palsyis the Self-help Group for Cerebral Palsy. There is also a Nepal Autistic Society.

6. Further opportunities for prevention of birth defects

- Maternal Death Review is being undertaken in 16 hospitals. Perinatal and neonatal death reviews could be initiated in these hospitals.
- Perinatal and neonatal databases could be initiated in hospitals where deliveries take place to capture data on birth defects.
- Data on birth defects should be incorporated in health management information systems.
- Prevention of birth defect messages should be incorporated into safe motherhood, neonatal health, adolescent health and NCD programmes.
- Reconstructive surgery camps for birth defects should be integrated within reproductive and child health immunization camps for the benefit of the majority.
- Priority should be given to birth defects and genetic disorders by the Government and a section on birth defects surveillance established.

SRI LANKA

Sri Lanka

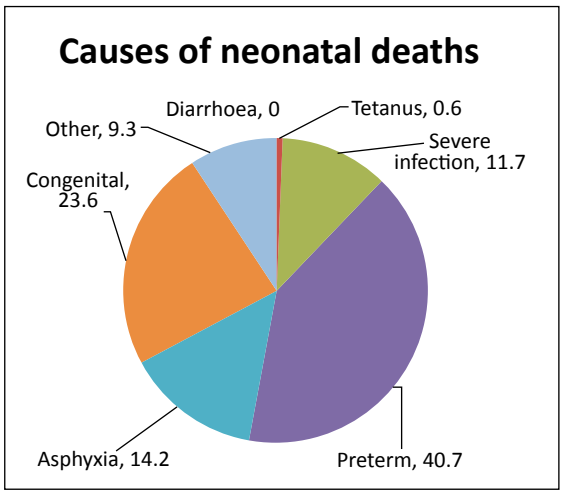
1. Demographic information

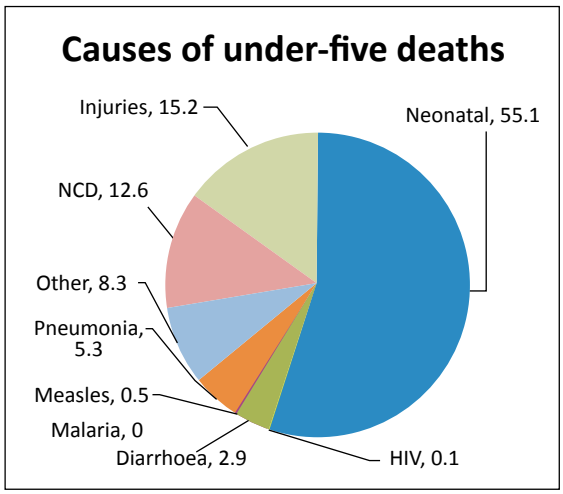
Total population ('000)	20 860
Estimated annual births ('000)	378
Under-five mortality rate (per 1000 live births)	12
Infant mortality rate (per 1000 live births)	11
Neonatal mortality rate (per 1000 live births)	8
Perinatal mortality rate (per 1000 live births)	17
Number of stillbirths	62
Complete immunization coverage by 12 months (%)	94.1
Complete immunization coverage at any time between 12–23 months (%)	97
Measles vaccination coverage by 12 months (%)	94.9
Measles vaccination coverage at any time between 12–23 months (%)	97
Vaccination against rubella:	Yes
Antenatal care coverage (any) (%)	99.4
First ANC visit before 4 months (%)	92.4
ANC visits: one visit (%)	1.3
ANC visits: four or more (%)	92.5
Iron folic acid supplementation coverage (%)	98
Institutional deliveries (%)	98.2
Deliveries assisted by skilled birth attendants (%)	98.6

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *Department of Census and Statistics (DCS) and Ministry of Healthcare and Nutrition (MOH) Sri Lanka. Sri Lanka Demographic and Health Survey 2006-07. Colombo, Sri Lanka: DCS and MOH, 2009.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

a. Congenital malformations

According to 2008 estimates, 1.9% of hospital admissions and 16.6% of hospital deaths are due to birth defects *(Hospital HMIS, 2008).*

Neural tube defects Dissanayake, *Wickramasinghe and Bandara (2010)* studied 14 580 live births at GH Kandy, 20 of whom had NTDs (1.37 per 1000 births). The number of babies with NTDs transferred from other hospitals was 30 (2.05 per 1000 births). They examined the knowledge and use of folic acid periconceptionally in families with affected children vs. a control group comprising 150 mothers with normal babies. Fourteen (28%) of the affected group were able to identify folic acid tablets as a vitamin taken during pregnancy, compared with 87 (58%) of the control group. One (2%) from the affected group and 70 (46%) from the control group knew about the value of preconceptional folic acid. None of the affected mothers had used folic acid preconceptionally whereas 20 (13%) of the control group had used it. Four (8%) of the affected mothers identified preventing birth defects as the primary reason to take folic acid whereas 54 (26%) of the control group mothers believed that it helped in preventing birth defects. Awareness of folic acid usage came mainly from midwives. Knowledge and use of folic acid in relation to pregnancy were grossly inadequate especially among mothers who had affected babies.

b. Perinatal infections

In Sri Lanka, birth prevalence of CRS was 0.9% per 1000 live births in 1994–1995 outbreaks *(Gunasekera and Gunasekera, 1996).* However, since 1994 rubella vaccination has been included in the expanded immunization programme.

c. Micronutrient deficiency disorders

A cross-sectional survey in the Galle district of 248 children aged 3–5 years was performed to determine the prevalence of micronutrient deficiencies *(Hossain, 2010).* The prevalence of anaemia (Hb < 110.0 g L) was 34% in men and 33% in women. In anaemic children, 7% of men and 15% of women were iron deficient (serum ferritin < 15.0 µg L). Folate deficiency (< 3.00 ng mL) was found in 41% and 33% of men and women, respectively, whereas Zn deficiency (< 9.95 µmol L) occurred in 57% and 50% of men and women, respectively. Serum vitamin D deficiency (< 35.0 nmol L) was found in 26% and 25% of men and women, respectively. Only 7.3% of subjects had no micronutrient deficiency, 38.3% were deficient in two micronutrients, 17.7% had three micronutrient deficiencies and 6.0% had four or more micronutrient deficiencies *(Hettiarachchi and Liyanage, 2012).*

The coverage of households using iodized salt is high; it is more than 92%. The median level of urinary iodine excretion is at 153 µg/L *(ICCIDD, 2012).*

d. Thalassemia

Thalassemia is a significant problem in Sri Lanka, especially in the North West province. De Silva et al. (2000) tested 1600 healthy school children in 16 centres throughout the country. There was some variability in the prevalence of both β thalassaemia trait and Hb E trait, the former ranging from 1–5% and the latter from 0–2%. On average, the prevalence was 2.2% (95% CI 1.3–3.0) for β thalassaemia and 0.5% (0.22–0.99) for Hb E, frequencies of 0.11 and 0.0025, respectively. The highest frequency was found in the Kurunegala State and in a small region on the south coast, areas which are, historically, those with the highest frequency of malaria (Simpson, 2007).

From the data on gene frequencies and the anticipated number of new cases born each year, it was predicted that more than 2000 patients will require regular treatment for the disease at any one time and that, based on current figures this will amount to 5% of the current health expenditure budget.

Taking cognizance of this burden, the Health Ministry announced that it will introduce legal provisions to make blood tests mandatory for every young couple before marriage. The Ministry also recommends that carriers should not marry carriers. If blood tests before marriage do not bring the desired results, the Health Ministry plans to bring legislation forbidding marriages between two thalassemia carriers. Such an extreme step is considered necessary as termination of pregnancy is not permitted, even if the foetus has an abnormality (except if it can save the life of the mother). The aim of the Government is to eradicate thalassemia from Sri Lanka by 2015. The government spends around Rs. 1.6 billion to treat thalassemia patients annually, Rs. 350 million of which is spent on drugs. Some 250 000 marriages are performed annually in Sri Lanka and offspring of 1600 parents are born with thalassemia *(Premawardhena et al., 2004).*

e. Other genetic defects

Down syndrome is among the commonest cause of learning handicap in children. Yet the stigma surrounding the condition is immense in Sri Lankan society and the issue is rarely discussed. However, awareness has increased that the condition can be screened for by biochemical tests and ultrasound studies. These tests are now available in the private sector. Autism has been recognized as a major problem, with a frequency of 1 in 93.

f. Disabilities

Currently the prevalence of population with disability is 1.6% (UNESCAP, 2012).During the 2001 census of the population in 18 districts 274 711 people were identified with disabilities. Of these, 25.1% had disability in sight, 26.6% in hearing/speaking, 17.5% of hands, 32.9% of legs, 4.8% had other disabilities and 25.1% had mental disability. Many subjects had multiple disabilities(Department of Census and Statistics Sri Lanka, 2003).

In Sri Lanka, birth prevalence of CRS was 0.9% per 1000 live births in the 1994–1995 outbreaks *(Gunasekera and Gunasekera, 1996).*Fortunately, rubella vaccine is currently in the national immunization schedule.

g. Consanguinity

Exact data on consanguineous marriages are not available for Sri Lanka. It is a cultural practice in certain parts of the country, but is gradually declining.

h. Birth defects as per the March of Dimes Report

The March of Dimes Report on Birth Defects estimated that 22 641 children were born with birth defects annually in Sri Lanka(Christianson, Howson and Modell, 2006). These comprise 2876 children with defects of the cardiovascular system, 728 with NTDs, 218 with haemoglobinopathies, 692 with Down syndrome and 692 with G6PD deficiency.

3. Noncommunicable diseases

NCDs are a priority for the Government, which has drawn up a three-year action plan to be implemented from 2011, through improvements in the primary health care system. An additional allocation of Rs. 900

million for this has been proposed. A sum of Rs. 200 million will be allocated to implement a "Mathata Thitha" initiative aggressively in every single village and township as a national priority. Tax on profits of businesses engaged in the manufacture and distribution of alcohol and cigarettes will increase from 35% to 40%.

4. Country response to birth defects

a) Surveillance programmes

There is no national birth defects surveillance programme at present. A National Steering Committee and focal person (Deputy Director General Primary Health Services) has been appointed to address the problem of birth defects in the country following the Regional Expert Group Meetings on Prevention of Birth Defects in South-East Asia held in New Delhi in December 2011 and in Bangkok in March 2012.

However, Sri Lanka has the second best health indices in SEAR. It has a well-organized health system reaching each household. All services are provided free at the Government hospitals. The health information system is excellent, and is in built in the health infrastructure. Vital statistics on morbidity and mortality are recorded in hospitals and maternal and child health centres, and a newborn information system has been established at health institutions where 98% of deliveries take place. Birth defects are recorded in the Neonatal Examination Form, in the neonatal unit admission note, and the Neonatal Intensive Care and Special Care units admission note. Data are also collected during the perinatal death review that has been established all over the country since 2006.

The neonatal and perinatal database(SEAR NPD/WHO Report, 2007–2008) reported that congenital malformations accounted for 17% of common primary causes of neonatal death in hospitals. Genitourinary, cardiac selling, prenatal diagnosis of genetic disorders and therapeutic abortion. The students responded with greater restraint possibly due to being a younger cohort with less clinical experience. Both groups, however, showed little support for the use of prenatal diagnosis purely to determine foetal sex.

Currently there is only one department of medical genetics, which exists as a unit of the Department of Anatomy in the University of Colombo. This should be upgraded to an independent department. There is a need to set up departments of medical genetics in other medical schools.

Thalassemia is a major problem, and the Government bears all the expenses for the care of thalassemic children. Mounting a control programme is essential, but the absence of a law for termination of pregnancy in the event of an affected fetus makes this difficult. The Government has therefore recommended screening for thalassemia before marriage to prevent union between carriers.

Genetic counselling: This is usually provided by clinicians. Counselling by medical geneticists is available at the Human Genetics Unit, Faculty of Medicine, University of Colombo (established in 1983). Counselling is also available by medical geneticists in the private sector. There are three medical geneticists in service, and five in training.

Genetic laboratories: Two main laboratories offer cytogenetic and molecular genetic testing (one in the university, and the other in the private sector). Tests available are karyotyping and molecular testing for thalassemia, ataxias and Duchenne muscular dystrophy.

c. Screening programmes

Currently there is no nationwide genetic screening programme. Antenatal screening for the following is practised:

- **Anaemia:** HB estimations is done at first antenatal clinic visit and at 28 weeks of pregnancy (coverage – 80%).
- **Haemoglobinopahies:** At first ANC visit by taking comprehensive history and by clinical examination (99%)
- **Syphilis:** At first antenatal clinic visit using venereal disease research laboratory (VDRL) test (98%) at field level.
- **Diabetes:** At first antenatal clinic visit if risk conditions exist, and at 24–28 weeks for all.
- Blood grouping and rhesus screening: in all.
- **HIV testing:** this is not routinely carried out. Information is provided during the first antenatal class and voluntary testing is encouraged.
- **Rubella, CMV, toxoplasmosis:** These tests are not routinely carried out. Facilities are available in major hospitals for women with repeated abortions or stillbirths.
- **Down syndrome and NTDs:** These are not routinely carried out. Facilities are available in the private sector and major hospitals.
- Ultrasound facilities are easily available and used for estimating gestational age and for antenatal diagnosis of birth defects routinely.
- **Thalassemia screening in endemic areas:** Facilities for prenatal diagnosis for thalassemia and other birth defects are available in the private sector and in major hospitals.
- **Newborn screening:** All newborns are examined by a medical officer before discharge from hospital using a newborn examination format to screen for birth defects (coverage – 80%). A newborn screening for hypothyroidism pilot project has been successfully completed in one province and is ready to be mainstreamed.

d. Prevention programmes

The country has a well-planned pre-pregnancy care programme consisting of folic acid supplementation, rubella immunization, and screening for medical diseases. During pregnancy, screening and treatment for syphilis are carried out. There is universal iodization of salt. About 98% of pregnant and lactating women receive iron and folic acid in the second and third trimester and for 6 months in the postnatal period. Coverage in the periconceptional period is not available. A weekly iron and folic acid supplementation programme (for 24 weeks per year) for adolescent girls and boys will be initiated in 2013.

The continuous decline in infant mortality reflects the impact of the health programmes that were implemented with special focus on child care, through the extensive network of medical institutions across the country. Emphasis is laid on preventive as well as curative care, with attention to awareness programmes to educate mothers on environmental sanitation, control of diarrhoeal diseases, healthy feeding habits, monitoring growth and child care. The countrywide immunization programme launched by the health authorities in the past decade to prevent six dangerous childhood diseases has produced the anticipated results: a remarkable 94% of children over one year of age were reported to be fully immunized in 2000. The coverage for BCG, DPT, and polio has almost reached the universal level, while immunization cover for measles stands at 94%. Full immunization cover against these communicable diseases is found to be 86% in the Estate sector and recorded as the lowest. Rubella immunization

is a component of the National Expanded Immunization Programme since 1994; as a result, 98% of women are protected at the time of conception.

Further prevention programmes are as follows.

- Consumption of alcohol and tobacco smoking is rare among women. The health education component of the preconception and maternal care packages include a session on the impact of alcohol during pregnancy. The National Authority on Tobacco and Alcohol conducts awareness programmes for the general public. Smoking in public places is banned in Sri Lanka.
- The impact of consanguinity is discussed in the school curriculum.

The following risk factors are discussed in the pre-pregnancy care package, maternal care package, Family Planning Programme:

- Avoidance of pregnancy after 35 years of age;
- Avoidance of medications, X-rays, environmental hazards, alcohol and tobacco during pregnancy;
- Detection and treatment of diabetes in women before and during pregnancy.

e. Micronutrient and folic acid fortification programme

Universal salt iodization is mandatory.

Two flour milling companies provide at least 90% of the wheat flour consumed in the country and one mill, with a smaller market share, is voluntarily fortifying its flour. Only 10% of wheat flour is fortified with iron and folic acid. The Government is currently promoting consumption of domestically grown rice and has not expressed interest in mandating flour fortification. Rice, which is the staple food, is not fortified with any micronutrient.

5. Services for care of people with birth defects

Minimum assistance for treatment, living, schooling, income-generating activities and housing is provided by the Department of Social Services, NGOs and the presidential fund. Special schools are available for children with visual and hearing impairment. For thalassemia blood transfusions, Chelation is provided by government hospitals, and screening facilities are available for the general public. For haemophilia laboratory investigations, factor VIII and factor IX therapy is available free of charge at government hospitals. Corrective and rehabilitative surgeries are available at both public and private facilities.

Parent organizations exist for Down syndrome, thalassemia, autism spectrum diseases, cerebral palsy, and visual and hearing impairments. A Disability Council is functioning under the guidance of the Department of Social Services.

The various stakeholders for health care are the Ministry of Health, Ministry of Social Services, Ministry of Education, Ministry of Women and Child Development, Ministry of Agriculture, Ministry of Sports and Professional Colleges (Physicians, Paediatrics, Obstetricians, Gynaecologists, Physiotherapists, Surgeons); WHO, UNFPA, UNICEF, World Bank, local NGOs and parent groups.

6. Further opportunities for prevention of birth defects

The Ministry of Health has appointed a nodal officer in the Maternal and Child Morbidity and Mortality Surveillance Unit, Family Health Bureau, to establish a Birth Defects Registry, based on the existing system of data collection. It will also oversee the creation of other facilities for birth defects. This will prove valuable for planning health services for birth defects.

THAILAND

Thailand

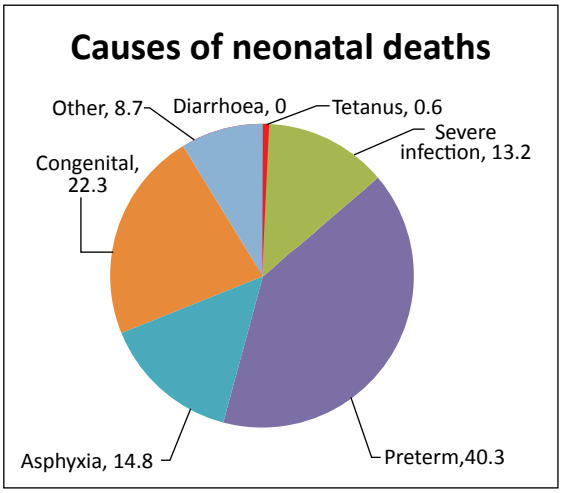
1. Demographic information

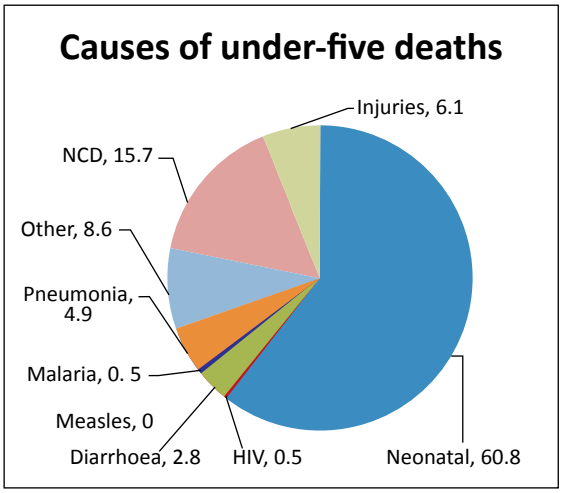
Total population ('000)	69 122
Estimated annual births ('000)	838
Under-five mortality rate (per 1000 live births)	12
Infant mortality rate (per 1000 live births)	11
Neonatal mortality rate (per 1000 live births)	8
Perinatal mortality rate (per 1000 live births)	No data
Number of stillbirths	No data
Complete immunization coverage by 12 months (%)	83.3
Complete immunization coverage at any time between 12–23 months (%)	89.7
Measles vaccination coverage by 12 months (%)	91.4
Measles vaccination coverage at any time between 12–3 months (%)	96.1
Vaccination against Rubella:	Yes
Antenatal care coverage (any) (%)	97.8
First ANC visit before 4 months (%)	No data
ANC visits: one visit (%)	No data
ANC visits: four or more (%)	No data
Iron folic acid supplementation coverage (%)	No data
Institutional deliveries (%)	96.7
Deliveries assisted by skilled birth attendants (%)	97.3

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *Thailand National Statistical Office. Thailand Multiple Indicator Cluster Survey December 2005- February 2006, Final Report. Bangkok, Thailand: National Statistical Office, 2006.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

In 2005–2006, a population survey indicated that almost 98.4% of births and deaths were registered in Thailand (Thailand National Statistical Office, 2007). The number of stillbirths (i.e. death after gestational age of 28 weeks) was about 4000 per year. Neonatal and perinatal deaths registered in the year 2010 were 21.53% and 21.93%, respectively.

The National Health Security Office (2011) reported the following figures for some common birth defects in Thailand: congenital heart defects (29%), ankyloglossia (10.7%), limb defects (8.9%), cleft lip/palate (8.9%), Down syndrome (3.7%) and NTDs (1.9%).

a. Congenital malformations

Table 12 summarizes the data on birth defects observed in various hospitals.

Table 12. Frequency of birth defects in various hospitals in Thailand

Hospital	Year of study	Births	Malformations	/1000 live births
Rachavithee	2501	43 142	400	9.0
Ramathebode	2519	15 187	371	24.0
QSNICH*	2533-2534	45 989	437	10.0
Siriraj	2536	18 958	161	9.0
Phumiphol	2538	8 725	87	10.0
Srinakarind	2540-2541	5 420	120	22.1
Songkla	2531-2542	27 061	337	12.45

**QSNICH, Queen Sirikit National Institute of hild Health.*

Source: Hospital HIS

The contribution of congenital malformations to neonatal deaths is 21.53% (679/3154), while to perinatal deaths it is 21.93% (481/2193). The most common birth defects are congenital heart defects, 23.37%, Ankyloglossia (tongue tie), 30.96%, limb defects, 5.31%, cleft lip/palate, 5.06%, Down syndrome, 2.43%, NTDs, 0.93%, based on analysis of 19 121 birth defects by the National Health Security Office (2011). Ankyloglossia is a minor defect and the numbers here represent the cases that were reimbursed expenditure for the surgical intervention.

All licensed assisted reproductive technology (ART) centres are obliged to submit annual reports on the number of patients, cycles, ART techniques and treatment outcomes to the Reproductive Medicine Subcommittee of the Royal Thai College of Obstetricians and Gynaecologists. Data from all centres were aggregated and analysed retrospectively

(Vutyavanich et al., 2011). The average pregnancy rate for in vitro fertilization (IVF) was 28.9% per retrieval (range, 26.0–32.3%) or 33.8% per transfer (range, 30.7–38.6%).

Multiple pregnancies (of which 89.3% were twins) from all treatment procedures during this period were 11.4% (range, 9.2–14.5%). A congenital abnormality was reported in 0.56% of live births.

Suwatanaviroj and Ratrisawadi (1996) observed that congenital malformations were the leading cause of death in the neonatal and infancy periods. They carried out a case–control study at 17 general hospitals and 2 maternal and child health centres in Thailand's central, northern, north-eastern, and southern regions and sought to identify the risk factors for congenital malformations. All cases seen at these facilities from May 1987 to April 1988 were included; normal infants born before and after the study served as controls. In this 12-month period, congenital malformations occurred in 437 infants (0.95% of total live births). Most common were malformations of the musculoskeletal system, cleft lip or cleft palate, and malformations of the nervous system. Risk factors significantly associated with malformations included maternal age over 35 years, low maternal education, separated/divorced marital status, family history of similar anomalies, an accident during pregnancy, and maternal illness during pregnancy, and maternal hypertension during pregnancy.

Neural tube defects

Wasant and Sathienkijkanchai (2005) carried out a retrospective chart review of patients with NTDs who were born at or referred to Siriraj Hospital during 1990–1999. During the 10-year period, 115 patients with NTDs were treated in the Department of Paediatrics as well as in other departments. The incidence of NTD was calculated as 0.67 per 1000 births. Isolated NTDs accounted for 105 (91%) of cases, and 10 (8.7%) had at least one other structural anomaly such as cleft lip/palate, imperforate anus, amniotic band sequence, or ambiguous genitalia. There was a high rate of mortality. Further studies are warranted to elucidate better the health burden from NTDs in Thailand. In a five-year study (1983–1987) conducted by the Division of Medical Genetics and Department of Paediatrics, Siriraj Hospital, the incidence of NTDs was found to be 0.6 per 1000 births (unpublished). This hospital has 18 000–20 000 deliveries annually. The previous prevalence or incidence reports of NTDs in Thailand from Srinagarind Hospital *(Ratanasiri et al., 1997),* and Maharaj Nakorn Chiang Mai Hospital (Kitisomprayoonkul and Tongsong, 2001) were 0.97 and 0.66 per 1000 births, respectively. One study of anencephaly at Ramathibodi Hospital noted the rate of anencephalic births was 0.62/1000 live births.

Kitisomprayoonkul and Tongsong (2001) analysed NTDs detected on ultrasound during pregnancy. The results showed that the incidence was 0.66/1000 births; however, spina bifida was very rare, found in only 0.06/1000 births, similar to encephalocele. No anencephalic fetus had concurrent spina bifida, and only a few cases had other associated anomalies.

Agthong and Wiwanitkit (2002) analysed 21 cases of meningocele over 10 years in Thailand. The most commonly involved area was the frontoethmoidal region, found in 20 cases.

Sayasathid, Tantiwongkosri and Somboonna (2009) reported that between January and December 2006, 38 055 children were examined by trained nurses and health officers to auscultate the precordium. Those with abnormal heart sounds were re-evaluated by paediatric cardiologists. Of the 278 subjects

with abnormal heart sounds, 43 had proven heart disease, 40 showed congenital heart diseases (1.05 per 1000) and three had rheumatic valvular diseases.

Cleft lip and palate

Cleft lip and palate are common congenital anomalies, and a number of publications have covered this defect. *Chowchuen and Godfrey (2003)* developed a network system for the care of patients with cleft lip and palate in Thailand, and observed that this was a suitable model for care. A community-based model for speech was implemented in Khon Kaen area *(Prathanee and Chowchuen, 2010).*

The South-East Asia Regional Neonatal–Perinatal database (WHO Report, 2007–2008) reported that congenital malformations accounted for 58% of primary causes of neonatal deaths in hospitals. Cardiac, gastrointestinal, genitourinary, hydrocephalus and cleft lip/palate were the commonest types of malformations.

b. Perinatal infections

A study of 49 infants with suspected intrauterine infections found that 56% were rubella-IgM positive *(Cutts et al., 1997).*

c. Micronutrient deficiency disorders

Sirikulchayanonta et al. (2004) determined serum folic acid of 165 normal non-pregnant women. Results showed that 65.5% of the study group had low dietary folate intake, that 18% had low serum folate, and that there was a significant correlation between dietary intake and serum level ($r = 0.68$, $P < 0.001$).

The coverage of households consume iodized salt is low, 47.2% and the median urinary iodine exretion is 144 µg/L (ICCIDD, 2012).

d. Thalassemia

Fuchareon and Winichagoon (2011) commented that haemoglobinopathies are the most common genetic disorders among people living in South-East Asia. Common defects are α thalassemia, β thalassemia, Hb E and Hb Constant Spring (CS). The gene frequencies of α thalassemia reach 30–40% in northern Thailand, and 20–30% elsewhere. The carrier frequency of β thalassemia varies between 1–9%. Hb E is the hallmark of South-East Asia attaining a frequency of 10–60% in Thailand. Hb CS gene frequencies vary between 1–8%. These abnormal genes in different combinations lead to over 60 different thalassemia syndromes, making the SEA Region an area with the most complex genotypes. The four major thalassemic diseases are homozygous α thalassemia 1 (Hb Bart's hydrops fetalis), homozygous β thalassemia, β thalassemia/Hb E and Hb H diseases. It is estimated that there are 600 000–800 000 cases of thalassemia in Thailand.

Wanapirak et al. (2004) analysed 516 pregnant women and observed an overall prevalence of thalassemia trait at 25.4%. The distribution was as follows: α thalassemia-1 (SEA type) trait, 6.6%, β thalassemia trait, 3.7%, Hb E trait, 11.6%, homozygous Hb E, 0.8%, the combination of α thalassemia-1 (SEA type) and β thalassemia trait, 1.2% and the combination of α thalassemia-1 (SEA type) and Hb E trait, 1.5%. They concluded that the prevalence of thalassemia carriers among pregnant women at Maharaj Nakorn Chiang Mai hospital was high, indicating the need for a screening programme aimed at prevention and control of this disease.

Suwanrath-Kengpol et al. (2005) observed non-immune hydrops fetalis in 71 cases in Southern Thailand. The causes of fetal hydrops were identified in 87.3% of the cases. Homozygous α thalassemia-1 dominated

as the cause of non-immune fetal hydrops (28.2%). Taweevisit et al. (2010) showed that autopsies in 78 stillborn hydrops fetalis, no case of immune hydrops were detected. The causes of fetal hydrops were identified in 88.5% of cases, the commonest cause being homozygous α thalassemia (n= 17; 21.8%).

Ratanasiri et al. (2006) screened 1498 women for thalassemia during antenatal period at Srinagarind Hospital in Khon Kaen, of whom 996 (64.2%) were carriers. However, prenatal diagnosis was carried out in only 19 couples where both partners were carriers. They showed that this strategy would be highly effective in reducing the burden of thalassemia in Thailand.

A study on the lifetime cost of homozygous β thalassemia in children was conducted in 2001, and the direct medical cost of treatment was estimated at 6 660 000 Thai baht, or approximately US$ 149 899, over a period of 30 years. This cost estimate was based on expert opinion, covering mainly blood transfusion and iron chelation drugs Fuchareon and Winichagoon (2007)estimated that almost 10 000 infants with various thalassemia syndromes are born in Thailand every year, while there are 434 000 patients living with these syndromes. They calculated that the cost of prevention of 1000 β thalassemia major (or with Hb E) patients) would be 48 280 Baht, while the cost of treatment would be 26 million Baht (US$ 890 000).

e. Other genetic disorders

Down syndrome

A number of studies have been published on the management of Down syndrome, but there is no reported data on the frequency at birth. The percentage of women who are more than 35 years old at the time of delivery is 12.8%, suggesting that Down syndrome frequency should be that observed in developed countries. *Wasant (1994)*estimated that 1000–1200 cases of Down syndrome are born every year in Thailand. She reported that this syndrome is the most common genetic cause of mental retardation and studied the mental development of 100 children with the disease. The mean development quotient (DQ) was 63.78 + 11.25 (range 32–91) with the majority being mild developmental delay. The child and family factors contributing to the DQ outcome were birth place, congenital heart disease, age at the first genetic counselling, regular follow-up in the genetics clinic, age at the first early stimulation programme/speech training programme, parental education/ occupation, and family income (Wasant, 1993). Only family income and age at the first speech-training programme were found to be independently associated with DQ at the age of 3 years (P-value < 0.05). Unachak et al. (2008) reported thyroid functions in children with Down syndrome.

Dr Pornswan Wasant, along with Dr Chaiyan Rajchagool, a sociologist and father of a Down syndrome child, initiated the “Down Syndrome Parents’ Support Group”. They reported that during the 15 years of their experience, no Down syndrome child was abandoned at Siriraj or other hospitals. Children who joined the association received better care and improved quality of life.

Jaruratanasirikul described his experience with 295 Down syndrome children attending the Songklanagarind Hospital stimulation intervention programme (*Jaruratanasirikul et al, 2004).* Congenital heart disease was found in 38.6% of cases, gastrointestinal anomalies in 16.9%, haematological malignancy in 6.1%, and thyroid disorders in 11.4%. The mortality rate of Down syndrome children was 13.2%. Most children (65.6%) received early stimulation, but only 38.9% attended the speech intervention programme within the first two years of life. Of the total 109 Down

syndrome children aged over 5 years that are still being followed, only 74 (67.9%) attended school. The school attendance was correlated with family income, but not with the level of maternal or paternal education.

Pruksanusak et al. (2009) determined the knowledge and attitudes of 714 pregnant Thai women towards Down syndrome screening through self-administered questionnaires. The mean age of respondents was 29.9 +/- 6.4 years. Most pregnant women had inadequate knowledge of Down syndrome and the screening test; 77.6% had a positive attitude to screening; and 92.2% would accept a Down syndrome screening test. Multivariate logistic regression analysis showed that levels of education and Vtypes of health insurance were factors associated with knowledge of Down syndrome screening. Lamlertkittikul and Chandeying (2007) reported on their experience on triple markers serum screening for Down syndrome fetus in Hat Yai Regional Hospital, where there was a 96% uptake of the test.

Wasant and Liammongkolkul (2003) reported their experience of prenatal genetic screening for Down syndrome and open NTD using maternal serum markers in 1793 Thai pregnant women, 60% of whom were older than 35 years.

Prenatal diagnosis

Ratanasiri et al. (2006) screened 1498 women for thalassemia during antenatal period at Srinagarind Hospital in Khon Kaen, 996 (64.2%) of whom were carriers. However prenatal diagnosis was only carried out in 19 couples where both partners were carriers. They showed that this strategy would be highly effective in reducing the burden of thalassemia in Thailand.

Piyamongkol et al. (2006) successfully carried out pre-implantation genetic diagnosis of thalassemia and chromosomal disorders.

Sirichotiyakul et al. (2008) reported results of chorionic villus sampling in Chiang Mai between January 2004 and July 2006 in 185 pregnant women. The mean gestational age was 12.25 + 1.05 weeks (range 10–20 weeks). The indications for prenatal diagnosis included fetal risk for chromosomal abnormalities (110 cases; 59.46%), severe thalassemia syndrome (57 cases; 30.81%), both of them (17 cases; 9.19%) and for human leukocyte antigen (HLA) typing in one case. Rueangchainikhom et al. (2008) reported their experience with chorionic villus sampling in 383 women from 1997 to 2006 in Bangkok, 346 of whom (95.84%) were more than 35 years of age. It was performed for chromosomal analysis in 355 while in 6 it was for abnormal thalassemia screening.

f. Disabilities

Deafness

Kasemsuwan et al. (2010) carried out a retrospective review of 143 ears (140 patients) operated with cochlear implant between 1995 and 2009. Most congenital origin deafness was of unknown etiology and congenital rubella was the most common known cause. From the computerised tomography (CT) scans of cases of congenital deafness, vestibular aqueduct dilatation was the most common finding (29.31% of cases) while Mondini malformation was shown to be 16.37%.

Blindness

The causes of blindness in the country are set out in Table 13. Other genetic disorders observed are skeletal dysplasias, craniosynostosis, neurofibromatosis, haemophilia and Marfan syndrome.

Table 13. Causes of blindness in Thailand

Cause of Thai visual impairment	Percentage
Cataract	51.64
Glaucoma	9.84
Age-related macular degeneration	6.56
Corneal scar	4.92
Congenital anomaly	4.10
Optic atrophy	4.10
Significant pterygium	3.28
Retinitis pigmentosa	3.28
ROP (retinopathy of prematurity)	1.64
Other	10.64

Source: *Thai Visual Project, 2006-2007*

g. Consanguinity

No data are available; estimated 5–10% (Wasant, 1994).

h. Birth defects as per the March of Dimes Report

The March of Dimes Report estimated that 58 522 children were born with birth defects annually in Thailand(Christianson, Howson and Modell, 2006). These comprise 7718 children with defects of the cardiovascular system, 684 with NTDs, 5471 with haemoglobinopathies, 1466 with Down syndrome and 3517 with G6PD deficiency.

3. Noncommunicable diseases

Signalling an epidemiological transition, communicable diseases, with the exception of HIV/AIDS, have been on the decline, while diseases and conditions related to social, environmental and behavioural factors are on the increase. A study in Bangkok revealed that 42% of persons over 15 years of age consumed fast foods and that only about 25% undertook regular exercise. Consumption of alcohol and tobacco are on the increase, with 22.8% of the population aged 15 years and above being regular smokers (1993). Suicide rates have increased in the Bangkok metropolis, as

have sex-related crimes. The Government's response has been to promote healthy lifestyles, especially sports and exercise, together with a series of campaigns launched against tobacco consumption. Therefore, the health plan has identified major strategies addressing issues related to health behaviour, disease prevention and control, and health promotion. Budgetary resources have been made available for health education and promotion, though still inadequate.

Kaufmann et al. (2011) concluded that chronic NCDs currently represent the largest cause of mortality in the Thai population. They suggested that while policy responses to chronic NCDs in the health sector have focused on improving prevention and control of the risk factors, a stronger emphasis on chronic disease treatment and management may be needed in the future.

4. Country response to birth defects

a. Surveillance programmes

Thai National Birth Defect Registry is being established at Queen Sirikit National Institute of Child Health on behalf of the Department of Medical Services (DMS) of the Ministry of Public Health, in cooperation with the Birth Defects Association (Thailand), and university hospitals, funded by DMS. The goal is to establish a National Birth Defects Registry by 2014 as a hospital-based, online 13 digit ID system, ICD 10, by Q mode.

The percentage of births and deaths registered is very high (98.4%). Surveillance programmes for a number of disorders are in place: HIV/ sexually-transmitted diseases, vaccine preventable diseases, and NCDs like cancer and tobacco consumption.

National surveys have been undertaken for mental retardation, disability and deafness by the Ministry of Public Health/Social and Human Development Ministry. There have also been surveys on blindness.

b. Genetic services

Aside from private centres, eight genetic centres in Thailand provide genetic counselling; counselling is also provided at these centres by paediatricians, obstetricians, internists and neurologists. There are 15 trained medical geneticists in the country distributed in various hospitals.

Genetic laboratories

There are a number of genetic laboratories 10 cytogenetic (public and private), 3 biochemical 3 (SI, RA, Chula) and 4 molecular (SI, RA, Chula, Songkla). In the private sector there are also laboratories that perform pre-implantation genetic diagnosis.

The common genetic tests performed in the country are chromosome studies, fragile X syndrome, Prader-Willi syndrome, FISH studies, inborn errors of metabolism, and neurogenetics.

Prenatal diagnosis

This service is available in Thailand in at least eight centres for thalassemia, Down syndrome and other genetic disorders. Termination of pregnancy is permitted up to 24 weeks for maternal mental/physical stress. A decision on termination is taken by a committee.

c. Screening programmes

- Thalassemia screening during pregnancy is carried out in all hospitals with coverage of 68% of all pregnant women.

- Newborn screening for congenital hypothyroid and PKU is carried out in all hospitals with a current coverage of 70.3% of all births. Almost 8 039 131 newborns have been screened. The incidence of congenital hypothyroidism (CH) is 1 in 1923 births, while that of PKU is 1 in 141 037 births.
- The percentage of women at birth who areolder than 35 years at delivery is 12.8%, suggesting that Down syndrome frequency may be that observed in developed countries. A programme on antenatal screening for Down syndrome is available in some government and many private hospitals through maternal serum screening.
- Antenatal screening is available for anaemia, haemoglobinopathies, infectious diseases like syphilis, HIV, rubella, CMV, toxoplasmosis, diabetes mellitus, Down syndrome and NTDs.
- HIV perinatal transmission outcome monitoring system is established in 14 Sentinel Provinces.
- Ultrasound studies during pregnancy are easily available. They are used frequently for prenatal diagnosis of birth defects, at least once during each pregnancy, and more frequently in abnormal pregnancies.

d. Prevention programmes

The Thailand National Plan for Prevention and Care of Birth Defects was initiated in 2009 and receives funding from the Thai Health Promotion Foundation (three-year project 2011–2014). It consists of early detection, diagnosis, treatment, rehabilitation, follow-up and prevention. The latter includes primary prevention through education, food supplement and secondary prevention through a referral team and care plan.

The National Program on Immunization covers more than 90% of all health settings. The rubella vaccine programme for children and women of childbearing age started in 1997. Also, campaigns for the use of alcohol among women during periconception and pregnancy are active in the country. A number of brochures on the effects of alcohol and tobacco consumption are also available at antenatal clinics.

Blindness Prevention and Eye Health Promotion (1999–2019) is in operation. It involves operations for cataracts, screening and proper intervention for diabetic retinopathy, glaucoma and childhood blindness.

e. Micronutrient supplementation and fortification

During pregnancy and lactation, folate, iron and iodine supplements are provided. Folic acid supplementation during the periconceptional period was initiated in all hospitals in 2011.

Nawapun and Phupong (2007) evaluated awareness of the benefits of folic acid and prevalence of the use of folic acid supplements to prevent NTDs among 401 Thai women. Although some pregnant women were aware of the need to take folic acid, the actual impact of the recommendations was negligible. Information to inform patients specifically about the need to take folic acid to prevent NTDs through media and health-care personnel seemed to improve its intake during the protective period.

Flour is centrally produced in eight sophisticated mills. Therefore, incorporating flour fortification in Thailand should be technically straightforward and inexpensive.

It is estimated that 30% of the population consumes 70–75 g/day, which is equivalent to a pack of instant noodles and is sufficient to offer significant protection. Thailand has a long history of close collaboration among government, industry and the NGO sector in initiatives for nutrition and fortification programmes. This has been successful for iodine fortification of salt, and noodles with multiple micronutrients. Greater effort is required to fortify foods with folic acid and achieve universal coverage and thus secure sustainability. Once technology for fortification of rice is developed at low cost this could be introduced throughput the country.

5. Services for care of people with birth defects

Parent organizations exist for thalassemia, lysosomal storage disease, Prader-Willi syndrome, Down syndrome and Marfan syndrome. Community-based rehabilitation programmes are available at the Sirinthorn Centre, community hospitals, as well as community centres. Corrective and rehabilitative surgery is provided at all tertiary care hospitals, especially at Sirithron Centre and Queen Sirikit National Institute of Child Health. A special centre for cleft lip/ palate exists at Chulalongkorn University. Surgery for congenital heart disease and other anomalies is available at many hospitals. For thalassemia, the cost of blood transfusions, chelation, and prenatal diagnosis are covered by the Government, as are laboratory investigations, factor VIII and factor IX as therapy for haemophilia.

6. Further opportunities for prevention of birth defects

Thailand has the best health indices in SEAR. It has an impressive health system extending from the primary to the tertiary level. The newborn screening is worthy of praise. The health department has decided to set up a birth defects surveillance system. What is now required is to set up departments of genetics in the medical schools. The thalassemia programme is a success. Programmes to screen for Down syndrome during pregnancy need to be established. Folic acid fortification of foods should be a priority as it would reduce the number of children born with NTDs.

TIMOR-LESTE

Timor-Leste

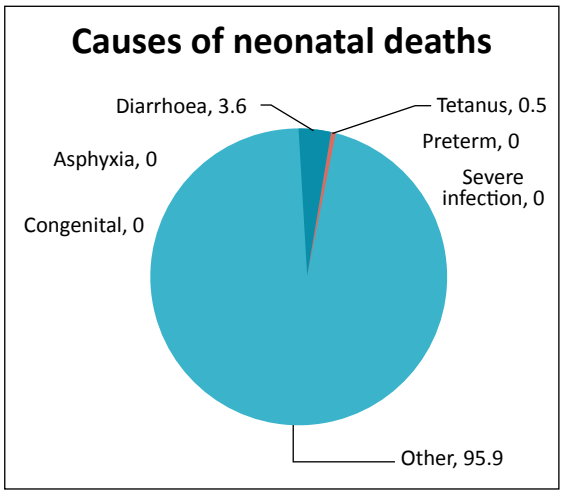
1. Demographic information

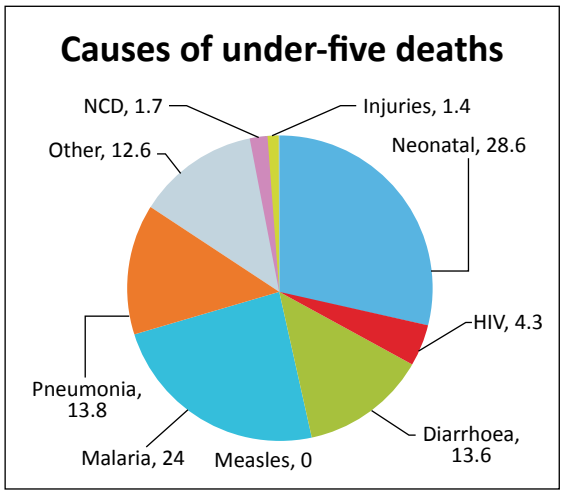
Total population ('000)	1124
Estimated annual births ('000)	44
Under-five mortality rate (per 1000 live births)	54
Infant mortality rate (per 1000 live births)	46
Neonatal mortality rate (per 1000 live births)	24
Perinatal mortality rate (per 1000 live births)	18
Number of stillbirths	22
Complete immunization coverage by 12 months (%)	47
Complete immunization coverage at any time between 12–23 months (%)	53
Measles vaccination coverage by 12 months (%)	60
Measles vaccination coverage at any time between 12–23 months (%)	68
Vaccination against rubella:	No
Antenatal care coverage (any) (%)	86
First ANC visit before 4 months (%)	45.3
ANC visits: one visit (%)	3.2
ANC visits - four or more (%)	55
Iron folic acid supplementation coverage (%)	63
Institutional deliveries (%)	22
Deliveries assisted by skilled birth attendants (%)	30

Sources: 1. *United Nations Children's Fund. State of the world's children 2012: children in an urban world. New York: UNICEF, 2012*

2. *The UN Inter-agency Group for Child Mortality Estimation (IGME). Levels & Trends in Child Mortality: Report 2012. 2012*

3. *National Statistics Directorate (NSD) Timor-Leste, Ministry of Finance Timor-Leste, and ICF Macro. Timor-Leste Demographic and Health Survey 2009-10. Dili, Timor-Leste: NSD Timor-Leste and ICF Macro,2010.*





Source: *World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.*

2. Information related to birth defects

The March of Dimes Report on Birth Defects estimated that 2774 children were born with birth defects annually in Timor-Leste(Christianson, Howson and Modell, 2006). These comprise 363 children with defects of the cardiovascular system, 32 with NTDs, 46 with haemoglobinopathies, 97 with Down syndrome and 483 with G6PD deficiency.

3. Country response to birth defects.

There are currently no services established for birth defects, except those that can be provided through the general health services.

4. Opportunities for prevention of birth defects

- With only 46 000 births per year it should be possible to establish a perinatal and neonatal databases that would provide information on the frequency and types of birth defects. This could be set up in the neonatal units.
- Immunization should be vigorously pursued. Measles and rubella should be added to the immunization schedule if possible.
- Family planning should be aggressively pursued to reduce pregnancies in older women so as to diminish the number of births with Down syndrome. DHS 2009-10 estimated that 23% of births were born from mothers aged 35-49 years (NSD, MoF and ICF Macro, 2010).
- Ultrasonography should be made available in district and tertiary care hospitals for the diagnosis of complications during pregnancy, malformations and markers of Down syndrome.
- Pregnant women should be encouraged to undergo a check-up in the first trimester to measure nuchal translucency and the presence of other congenital anomalies.
- Fortification of flour and rice with folic acid and other micronutrients should be actively considered, as this will combat the high prevalence of anaemia in children as well as in pregnant women. The country imports the majority of its flour and has no or little domestic milling industry. Methods need to be developed to ensure that the imported flour is fortified with folic acid. Once fortification of rice is achieved at low cost, this should be introduced throughout the country.

6. SUMMARY

Analysis of information from countries in SEAR makes it immediately apparent that they are in different phases of development. As health services improve and infant mortality due to communicable diseases is brought under control, the contribution of genetic disorders and birth defects to morbidity and mortality will increase proportionately. WHO and the United Nations have called for developing countries to plan to prevent and manage noncommunicable disorders which are evolving in their countries. Although birth defects should have a place in the different programmes for health services, they receive little attention in almost all SEAR countries at present, although opportunities do exist within current public health initiatives and programmes. One of the main reasons is that countries do not have enough information on the burden that birth defects contribute in terms of mortality and morbidity as well as social costs.

The main objective of commissioning this situation assessment on birth defects in Member States was to develop a better understanding on what information exists in the Region. The information described in previous sections is summarized in the following tables that spotlight important issues.

This information was confirmed by national programme managers during the Regional Programme Managers meeting on Prevention and Control of Birth Defects held in March 2012 in Bangkok, Thailand and organized by SEARO with the support of the United States Centers for Disease Control and Prevention.

1. Demographic information

SEAR comprises 11 countries that vary in terms of population characteristics, from small countries like Maldives with a population of 328 536 to large countries like India with a population of 1.21 billion. Vital registration systems are not well developed in several countries, coverage of births registered varies from 34% in rural areas in Nepal to almost 100% in Bhutan. Registration of deaths is even less developed except in India (63%), Sri Lanka (94%) and Thailand (98.4%). Data on the frequency of stillbirths are not available since only Nepal (3831 cases per year) and Thailand (4000 cases per year) were able to provide the numbers. The proportion of deliveries to women aged older than 35 years, who are at higher risk of producing babies with genetic disorders like Down syndrome, ranges from 4.5% in Bhutan to 15.5% in Sri Lanka.

Table 14. Selected demographic Information

Country	Births registered (%)	Deaths registered (%)	Stillbirths registered	Women older than 35 years at delivery (%)	Population who marry consanguineously
Bangladesh	Project in progress	NA	NA	NA	1 district: 10%
Bhutan	99.9	NA	NA	4.5 (single facility data)	Exist, Data NA

India	National: 68 7 states: 100	63	NA	Negligible	South India: 20% Muslims: 20%
Indonesia	NA	NA	NA	NA	NA
Maldives	99	Yes	Yes	11	Uncommon
Myanmar	65	NA	NA	Yes	NA
Nepal	Urban: 42 Rural: 34	NA	3831 cases/ year	7.8	7%
Sri Lanka	97.2	94	Limited	15.5	Uncommon
Thailand	98.4	98.4	4000 cases/	12.8 year	NA

NA, not available.

2. Information related to birth defects

Table 16 summarizes information on birth defects for nine SEAR countries as there are little data available from DPR Korea and Timor-Leste. In six countries (Bangladesh, Bhutan, Indonesia, Maldives, Sri Lanka, and Thailand), congenital heart diseases are the most common birth defects. In India, NTDs are the most common; while in Myanmar and Nepal it is thalassemia and cleft disorder, respectively. Thalassemia is significant in Bangladesh, India, Indonesia and Maldives. In India, Down syndrome has a high occurrence. Ankyloglossia is a minor birth defect and often subjected to unnecessary surgery. It has been reported from Thailand based on the reimbursement records.

The source of information on birth defects varies across countries (Table 16). However, the data are mainly collected from hospital-based surveys (except in Bangladesh and Indonesia) and/or published studies. In Bangladesh, there is no registry, survey or surveillance for birth defects. A national registry on birth defects has been initiated in Thailand, while in India such a registry operates in some states. A population-based survey on birth defects has been conducted in Sri Lanka. Surveys for disability have been conducted in India, Indonesia, Myanmar, Sri Lanka and Thailand, while in Bhutan a survey on mental disorders has been piloted. Surveillance systems exist for communicable diseases in all Member States; registries for diseases like cancer and diabetes exist in only a few countries.

Table 15. Common birth defects in countries of the South-East Asia Region

Rank	Bangladesh	Bhutan	India	Indonesia	Maldives	Myanmar	Nepal	Sri Lanka	Thailand
1	CHD	CHD	NTD	--	CHD	Thal	Cleft	CHD	CHD
2	NTD	NTD	Down	--	NTD	CHD	CHD	Chrom	Ank*
3	Thal	Cleft	MD	--	Thal	Cleft	NTD	NTD	Limb defects
4	Down	Chrom	Thal	--	Down	CGI	CRS		Clefts
5	G6PD		CH	--	G6PD	Down			Down
6			DMD			G6PD			NTD

* Based on payment reimbursement records.

Ank, ankyloglossia; CGI, congenital gastrointestinal conditions; CH, congenital hypothyroidism; CHD, congenital heart defects, includes cardiovascular system defects; Chrom, chromosomal abnormalities, including Down syndrome; Cleft, cleft lip or palate; CRS: congenital rubella syndrome; DMD, Duchene muscular dystrophy; Down, Down syndrome; G6PD, glucose-6-phosphate dehydrogenase deficiency; MD, metabolic disorders; NTD, neural tube defects, including spina bifida; Thal, thalassemia and pathological haemoglobin disorders.

Table 16. Source of data/information on birth defects available in the country

Country	Registry: national or subnational	Hospital-based health information survey	Population-based health information survey	Special surveys	Published studies
Bangladesh	No	No	No	No	Yes
Bhutan	No	Yes	No	Pilot survey on mental disorders	Yes
India	Subnational	Yes	No	Disability	Yes
Indonesia	No	Yes	No	Disability	
Maldives	No	Yes	No	No	Yes
Myanmar	Some hospitals	Some have registries	No	Disability (2009)	Yes
Nepal	No	Yes	No	No	Yes
Sri Lanka	Pilot	Yes	Yes	No	Yes
Thailand	Yes	Yes	No	Yes	Yes

** Based on March of Dimes (2006).*

3. Country responses to birth defects

a. Surveillance programmes

Member States have limited surveillance activities for birth defects at present (Table 17). A hospital-based health information system for birth defects exists in almost all countries, except Bangladesh. It is unclear if a standardized coding system is used in these countries. A birth defect registry exists in parts of India and has recently been initiated in Thailand. Surveillance for communicable diseases like HIV/STI exists in Bangladesh, Maldives, Myanmar, Sri Lanka and Thailand; surveillance for NCDs like cancer and tobacco consumption is established in Bhutan and Thailand. A comprehensive school health screening programme is available in Bhutan. Newborn screening is available on a small scale in some countries. Clearly, there are opportunities to use the existing activities and capacity for birth defects surveillance.

Table 17. Surveillance programmes on response to birth defects

Country	Surveillance system	Hospital-based HIS	Birth Defect Registry	School health screening
Bangladesh	- Nutritional surveillance with Helen Keller International - Surveillance for communicable diseases (HIV/STI) - Surveillance for acute flaccid paralysis and poliomyelitis	N/A	N/A	N/A
Bhutan	Cancer registry being developed	Yes	Birth Registry mentions congenital anomalies	Yes
India	N/A	Yes	Yes, in Chennai	N/A
Indonesia	N/A	Yes	N/A	N/A
Maldives	Surveillance for communicable diseases	N/A	Yes	N/A
Myanmar	Surveillance for communicable diseases	Yes	Some	N/A
Nepal	N/A	To be initiated	N/A	N/A
Sri Lanka	Perinatal death reviews, CD	Yes	Pilot	N/A
Thailand	Yes: HIV/STI, vaccine preventable diseases, NCDs (cancer, tobacco use).	Yes	Being established	N/A

NA, Not available

b. Genetic services

Limited genetic services are available only in a few SEAR countries (Table 18). In India, Sri Lanka and Thailand, genetic services such as in screening, counselling, laboratories and genetic tests are available at selected centres. In Bangladesh and Indonesia, a few genetic laboratories are available. Genetic services for screening counselling, and laboratories in Maldives are limited to thalassemia at present.

Table 18. Genetic services

Country	Genetic screening	Genetic counselling	Genetic laboratories	List of available genetic tests
Bangladesh	No	No	Few	No
Bhutan	No	No	No	No
India	Yes: Support from Indian Council of Medical Research (ICMR)	Yes: 45 ICMR facilities	26 biochemical, 28 molecular labs	Yes
Indonesia	No	No	Few	No
Maldives	Thalassemia	Yes: for thalassemia	Yes: for Thalassemia	No
Myanmar	No	Limited	Limited	No
Nepal	No	No	No	No
Sri Lanka	Yes, for Thalassemia	Yes	No	Yes
Thailand	Yes	Yes	Yes	Yes

c. Screening programmes

Screening programmes on birth defects vary across countries (Table 19). Screening during the antenatal period is available in almost all countries and includes screening for anaemia, haemoglobinopathies, and infectious diseases like syphilis. Using ultrasonography for screening on birth defects is practised in Bangladesh, India, Maldives, Myanmar, Sri Lanka and Thailand at selected centres only. Newborn screening, mainly for hypothyroidism, is available in limited areas in Bangladesh, India, Indonesia, Maldives, Sri Lanka, and Thailand. Population screening for thalassemia is available in India, Maldives, Sri Lanka, and Thailand. Limited facilities for prenatal diagnosis are available in Bangladesh, India, Sri Lanka, and Thailand. Medical termination of pregnancy of foetus with birth defects is legally allowed in Bangladesh, Bhutan, and India only.

Table 19. Screening programmes

Country	Antenatal screening	Ultrasound screening	Newborn screening	Population screening for thalassemia	Prenatal diagnosis	Medical termination of pregnancy
Bangladesh	Yes, only for high-risk groups	Yes	Hypothyroidism	No	Sporadic	Yes
Bhutan	National: NTDs All levels: anaemia	Only for confirming pregnancy	No	No	No	Yes
India	Yes (some states for Hbpathies)	Yes	Yes, in some states	Yes	Yes	Yes
Indonesia	Yes, for anaemia, Hbpathies, infectious diseases	No	Yes, in some provinces for Hypothyroidism	No	No	No
Maldives	Yes	Yes	Selectively	Yes	Abroad	Yes, up to 120 days
Myanmar	Yes, for anaemia, infectious diseases)	Yes: tertiary care and private	No	No	No	No No
Nepal	No	No	No	No	No	No
Sri Lanka	Yes	Yes	Yes	Yes	Yes	Not legal
Thailand	Hbpathies, Down syndrome, anaemia, infectious diseases	Yes	Yes: PKU, hypothyroidism	Yes	Yes	Yes, up to 12 weeks

Hbpathies, haemoglobinopathies; PKU, phenylketonuria.

d. Pre-pregnancy and pregnancy prevention programmes

Prevention interventions for birth defects during pre-pregnancy and pregnancy period are available under several health and nutrition programmes in SEAR Member States (Table 20).

Table 20. Pre-pregnancy and pregnancy care

Country	Rubella vaccine	Use of alcohol among women	Use of tobacco and exposure during pregnancy	Education and awareness programmes		
				Avoid pregnancy > 35 years	Avoid teratogens	Detection of type 2 diabetes
Bangladesh	Yes: 2012	Banned	General	Sporadic	Yes	Urine test
Bhutan	Yes: since 2006 (women & children)	No programmes but awareness programmes, high taxation	Strict legislation against tobacco	Maternal & child health handbook, posters	Yes	Yes
India	Yes: some states	No: national ban in one state	Yes	No	No	Yes: secondary & tertiary care
Indonesia	Yes	Yes: ANC	Yes: ANC	Yes: ANC	Yes: ANC	Yes: ANC
Maldives	Yes: since 2007	General national campaign	General national campaign	Yes	Yes	Yes
Myanmar	Private sector	Health education	Health education	No	Yes	Yes: for DM during ANC
Nepal	Yes: 2012	Yes	Yes	No	Yes	Yes
Sri Lanka	Yes: Since 1994	General national campaign	General national campaign	Yes	Yes	Yes
Thailand	Yes	Yes	Yes	Yes	Yes	Yes

ANC, antenatal care; DM, diabetes mellitus.

e. Micronutrient fortification with folic acid

Folic acid fortification and supplementation programmes are being implemented at varying scales in Member States (Table 21) that may offer the potential for prevention of NTD. Another micronutrient fortification programme is salt iodization.

Table 21. Folic acid and micronutrient supplementation and fortification

Folic acid supplementation and fortification					Other micronutrients
Country	**Adolescents**	**Pregnancy and lactation**	**Peri-conceptional**	**Composition of tablets**	
Bangladesh	Yes	Yes	No	NA	Law exists for universal salt iodization
Bhutan	Yes, school children (WIFS)	Yes: same dose daily	Yes: up to 3 months	Iron: 60 mg Folic acid: 0.5 mg	Yes, iodized salt
India	Yes (WIFS)	65% coverage	No	Iron: 100 mg Folic acid: 0.5 mg	Flour fortification with iron in some states
Indonesia	No	Yes	No	Iron: 60 mg Folic acid: 0.25 mg	Yes: (Biscuits with folic acid and iron in 89 districts) (Iodized salt)
Maldives	No	Yes	No	Iron: 60 mg Folic acid: 0.4 mg	No
Myanmar	Yes: twice a day, partial coverage	Yes: daily	No	Iron: 200 mg Folic acid: 0.4 mg	Yes, iodine fortification
Nepal	Yes	Yes	Private clinics	NA	Yes, iodized salt, flour fortification containing vitamin A, iron and folic acid
Sri Lanka	Starting in 2013	Yes	No	Iron: 60 mg Folic acid: 1 mg	Iodized salt
Thailand	No	Yes	Yes	Triferdine: iodine, iron & folic acid	Yes, iodized salt

NA, not available.

4. Services for care of people with birth defects

Limited services are in place for care of people with birth defects in Member States, through family support programmes, corrective surgeries and rehabilitation (Table 22).

Table 22. Services for care of people with birth defects

Country	Family support programmes	Corrective surgeries and/or rehabilitation programmes	Community rehabilitation programmes	Parent organizations	List of stakeholders
Bangladesh	Yes	Yes	No	Yes	Yes
Bhutan	None	Yes: "camps" for cleft lip and palate 2x/year	Yes	Disabled persons association for visually impaired	NA
India	Some	Medical colleges and tertiary care	NGOs/civil society	Yes	Multiple
Indonesia	Yes	Yes	Yes	Yes	Yes
Maldives	Yes	Yes	Yes	Yes	Yes
Myanmar	Yes	Yes	Yes	Yes: autism, cleft lip and cancer	Multiple
Nepal	Yes	Yes	Yes	Yes	Yes
Sri Lanka	Yes	Yes	Yes	Yes	Yes
Thailand	Yes	Yes	Yes	Yes	Multiple

NA, not available; NGO, nongovernmental organizations.

5. Further opportunities for prevention of birth defects

The situation analysis reveals that there are high potential opportunities in the Member States to mount prevention and control programmes for birth defects. Prevention interventions can be integrated in the health and nutrition programmes already being implemented in the countries. RMNCAH services are being implemented in all Member States with significant success. Several birth defect prevention interventions can be integrated in other programmes like immunization and NCD. Activities of birth defects surveillance can also be integrated into existing information systems like vital registration system, perinatal and stillbirth surveillance, national household surveys and facility-based health management. Some countries already have experience with registries such as those for cancer.

6. Challenges

Common challenges reported by countries are summarized in the box below.

Challenges faced by Member States of the South-East Asia Region

- Competing priorities like communicable diseases: need strong advocacy on the significance of birth defects as a cause of mortality, morbidity and disability.
- Absence of a focal department/unit and intersectoral collaboration for the prevention of birth defects.
- Gaps in the health information system: weak VRS, poor data on stillbirths/abortions, incomplete HMIS, and no dedicated birth defect registry.
- Limited resources: for screening services, genetic services; pathological postmortem, human resources (laboratory, genetics diagnosis, counselling etc.), infrastructure, and funding.
- Improvement of the Laboratory capacity and Information System (LIS).
- Expansion of food fortification with folic acid and other micronutrients.
- Limited facilities for corrective and rehabilitative facilities.
- Legal framework for mandated surveillance, termination of pregnancy.
- Ethical issues and stigma related to birth defects.

6.7 Main areas in which support is required

Countries have conveyed the following areas in which they would need support for initiating birth defects prevention and control programmes.

Required support by South-East Asia countries for birth defects programmes

- Support for national advocacy and communication strategy.
- Support for development of national strategy: implementation plans and guidelines.
- Development of integrated surveillance systems: strengthening VRS, HMIS, national surveys and birth defect registries.
- Integrating birth defects prevention into safe motherhood, family planning, neonatal child and adolescent health programmes.
- Capacity development for screening programmes, diagnostics, and genetics services.
- Capacity-building of health workers/care givers at various levels and in various disciplines.
- Care and support for children with birth defects, and special needs (educational centres) for these children.
- Large-scale food fortification (folic acid and other micronutrients).
- Capacity for monitoring, supervision, research and evaluation.
- Development of networks of partners and institutions.
- Funding.

REFERENCES

Agarwal KN, Agarwal DK. Prevalence of anaemia in pregnant and lactating women in India. Indian J Med Res 2006;124:173-84.

Agthong S, Wiwanitkit V. Encephalomeningocele cases over 10 years in Thailand: a case series. BMC Neurol. 2002 May 13;2:3.

Aguayo VM, Paintal K, Singh G. The Adolescent Girls' Anaemia Control Programme: a decade of programming experience to break the inter-generational cycle of malnutrition in India. Public Health Nutr.2013 Jan 24:1-10.

Akhter N, Haselow N. Using data from a nationally representative nutrition surveillance system to assess trends and influence nutrition programs and policy. Field Actions Science Reports. 2010;4:1-7.

Ambrus A, Field E, Torero M. Muslim family law, prenuptial agreements and the emergence of dowry in Bangladesh. Quarterly J Economics. 2010;125(3):1349–97,

Appaji Rao N, Radha Rama Devi A, Savithri HS, Venkat Rao S, Bittles AH. Neonatal screening for amino acidemias in Karnataka, South India. Clin Genet. 1988;34:60–63.

Aung-Than-Batu, U-Hla-Pe, Khin-Kyi-Nyunt, Tin-U. The thalassemia in Burma. Union of Burma Journal of Life Sciences. 1968;1:241–247.

Bale JR, Stoll BJ, Lucas AO. Reducing birth defects:meeting challenges of the developing world. Washington DC: National Academies Press, 2003.

Bangladesh NCD Network. Tale of endurance. July-Sept 2011. Available at http://www.bdncdnet.com/- accessed 19 Feb 2013.

Bawi SV. Understanding the challenges of disability in Myanmar.Internships Asia and Hussman Foundation, 2012.

Begum NL, Azad K, Akhter S, Nahar N, Hasan A, Abdullah M. Assessment of perinatal mortality in a tertiary care hospital by using Wigglesworth classification. Bangladesh J. Child Health. 2007;31(1-3):32-39.

Bhandari, P. Incidence of Congenital heart diseases in Mongar Hospital, 2010 (Unpublished)

Bharucha BA, Kher AS, Naik GG, et al.Spectrum of inherited metabolic disease in India:a 15 year experience.In: Chaudhry P, Sachdev HPS, Puri RK, et al, editors.Abstracts of the 8th Asian Congress of Pediatrics. New Delhi: Jaypee Publishers, 1994.p 98.

Bhasin MK. Genetics of Castes and Tribes of India: Glucose-6-Phosphate Dehydrogenase Deficiency and Abnormal Haemoglobins (HbS and HbE). Int J Hum Genet. 2006;6(1):49-72.

Bhattacharjee A, Chakraborty A, Purkaystha P. Frontoethmoidal encephalo-meningocoele with colpocephaly: case report and clinical review. J Laryngol Otol. 2008 Mar;122(3): 321-3.

Birth defects registry of India - a 'Saving Babies' project. MedIndia.Available at http://www.medindia.net/news/healthwatch/Birth-Defects-Registry-of-India-A-Saving-Babies-Project-78389-1.htm - accessed 19 February 2013.

Birth defects registry of India. Newsletter volume 11, Jan – June 2011.Available at http://www.fcrf.org.in/bdri_news.asp - accessed 19 February 2013.

Bittles AH.A background summary of consanguineous marriage. Perth: Centre for Human Genetics, 2001. Available at http://consang.net/images/d/dd/01AHBWeb3.pdf - accessed 19 February 2013.

Bittles AH. The impact of consanguinity on the Indian population.Indian J Hum Genet.2002; 8:45-51.

Black ML, Sinha S, Agarwal S, Colah R, Das R, Bellgard M, Bittles AH. A descriptive profile of β-thalassaemia mutations in India, Pakistan and Sri Lanka.J Community Genet. 2010; 1:149–157

Bundhamcharoen K, Odton P, Phulkerd S, Tangcharoensathien V. Burden of disease in Thailand: changes in health gap between 1999 and 2004.BMC Public Health. 2011; 11:53.

Central Intelligence Agency (CIA). The world factbook. Available at https://www.cia.gov/library/publications/the-world-factbook/geos/bg.html - accessed 19 February 2013.

Central Bureau of Statistics (CBS), Democratic People's Republic of Korea. Democratic People's Republic of Korea Multiple Indicator Cluster Survey 2009, Final Report. Pyongyang: CBS, 2010.

Chowchuen B, Godfrey K. Development of a network system for the care of patients with cleft lip and palate in Thailand. Scand J Plast Reconstr Surg Hand Surg. 2003; 37 (6):325-31.

Christian P, West KP Jr, Katz J, Kimbrough-Pradhan E, LeClerq S C, Khatry SK, Shrestha S R. Cigarette smoking during pregnancy in rural Nepal. Risk factors and effects of beta carotene and vitamin A supplementation.Eur J Clin Nutr. 2004; 58: 204–211.

Christianson A, Howson CP, Modell B, editors. March of dimes global report on birth defects: the hidden toll of dying and disabled children. New York: March of Dimes Birth Defects Foundation, White Plains, 2006.

Colah R, Gorakshakar A, Phanasgaonkar S, D'Souza E, Nadkarni A, Surve R, et al. Epidemiology of β-thalassaemia in Western India: mapping the frequencies and mutations in sub-regions of Maharashtra and Gujarat. British Journal of Haematology. 2010;149 (5): 739–47.

Cutts FT, Robertson SE, Diaz-Ortega JL, Samuel R. Control of rubella and congenital rubella syndrome (CRS) in developing countries, part 1: burden of diseases from CRS. Bull World Health Organ. 1997;75 (1): 55-68.

Czeizel AE. The primary prevention of birth defects: Multivitamins or folic acid? Int J Med Sci. 2004; 1:50-61.

Deka D. Congenital intrauterine infections. New Delhi: Jay Pee Medical Publishers, 2010.

Department of Census and Statistics, Government of Sri Lanka. Census of population and housing – 2001:information on disabled persons. Colombo: Department of Census and Statistics, 2003. http://statistics.sltidc.lk/index.php/catalog/939/reports - accessed 19 February 2013.

Department of Census and Statistics (DCS) and Ministry of Healthcare and Nutrition (MOH) Sri Lanka. Sri Lanka Demographic and Health Survey 2006-07. Colombo, Sri Lanka: DCS and MOH, 2009.

Department of Health Planning and Department of Health, Ministry of Health, the Government of the Union of Myanmar. Annual hospital statistics report 2008. Nay Pyi Taw: Department of Health Planning and Department of Health, 2010.

Department of Social Welfare (DSW) and the Leprosy Mission International (TLMI). Myanmar national disability survey. 2010

Dey AC, Shahidullah M, Mannan MA, Noor MK, Saha L, Rahman SA. Maternal and

neonatal serum zinc level and its relationship with neural tube defects. J Health Popul Nutr. 2010 Aug; 28 (4): 343-350.

De Silva S, Fisher CA, Premawardhena A, Lamabadusuriya SP, Peto T, Perera G, Old JM, Clegg J, Olivieri NF, Weatherall DJ.Thalassaemia in Sri Lanka: implications for the future health burden of Asian populations.Lancet. 2000; 355: 786–791.

Dineen BP, Bourne RR, Ali SM, Huq DM, Johnson GJ.Prevalence and causes of blindness and visual impairment in Bangladeshi adults: results of the national blindness and low vision survey of Bangladesh. Br J Ophthalmol.2003 Jul;87(7): 820-8.

Disease Control Priority Project. Controlling birth defects: reducing the hidden toll of dying and disabled children in low income countries. 2008

Dissanayake NN, Wickramasinghe SC, Bandara DMWH. Study on neural tube defects of neonates and folic acid awareness at a Teaching Hospital in Sri Lanka. Sri Lanka J Child Health. 2010;39:11-16

Durkin MS, Khan NZ, Davidson LL, Huq S, Munir S, Rasul I, Zaman SS.Prenatal and postnatal risk factors for mental retardation among children in Bangladesh. Amer J Epidemiol. 2000; 152:1024-32.

Elango R, Verma IC. Fragile X syndrome among children with mental retardation.Indian J Pediatr. 1996; 63:533–538.

Ellis M, Azad K, Banerjee B, Shaha SK, Prost A, Rego RA, et al. Intrapartum-related stillbirths and neonatal deaths in rural Bangladesh: prospective, community-based cohort study iIntrapartum-related stillbirths and neonatal deaths in rural Bangladesh. Pediatrics. 2011;127:1182.

Ellis M, Manandhar DS, Manandhar N, Wyatt J, Bolam AJ, Costello AM. Stillbirths and neonatal encephalopathy in Kathmandu, Nepal: an estimate of the contribution of birth asphyxia to perinatal mortality in a low-income urban population. Paediatr Perinat Epidemiol. 2000 Jan; 14 (1):39-52.

Emma – Lindstro M, Mohammad, Hossain B, Nnerdal NL, Raqib R, El Arifeen S, EKSTRO E. Prevalence of anemia and micronutrient deficiencies in early pregnancy in rural Bangladesh, the MINIMat trial. Acta Obstetricia et Gynecologica Scandinavica. 2011;90:47–56.

Firdous N, Gibbons S, Modell B. Falling prevalence of beta-thalassaemia and eradication of malaria in the Maldives. J Community Genet.2011 Sep; 2(3):173–89.

Fucharoen S, Winichagoon P. Prevention and control of thalassemia in Asia. Asian Biomedicine. 2007;1:1-6.

FucharoenS, Winichagoon P. Haemoglobinopathies in Southeast Asia. Indian J Med Res. 2011 Oct; 134:498-506.

Gamble M V, Ahsan H, Liu X, Litvak P F, Ilievski V, Slavkovich V, Parvez F, Graziano JH. Folate and cobalamin deficiencies and hyperhomocysteinemia in Bangladesh. Am J Clin Nutr 2005; Jun; 81(6):1372-7.

Geetha T, Chenoy R, Stevens D, Johanson RB. A multicentre study of perinatal mortality in Nepal. Paediatr Perinatal Epidemiol. 1995; 9: 74-89.

Glasgow BG, Goodwin MJ, Jackson F, Kopeć AC, Lehmann H, Mourant AE, et al.The Blood Groups, Serum Groups and Haemoglobins of the Inhabitants of Lunana and Thimbu, Bhutan, Vox Sanguinis. 1968;14:31–42.

Glinka J. Consanguineous marriage. In:Gesch PF, editor. Culture, gospel and church.Madang: Divine Word Mission. 1994. p. 92 99.

Godbole KU, Yajnik C. Nutri-genetic determinants of neural tube defects in India. Indian Pediatr. 2009:46(6):467-75

Government of India, Ministry of Home Affairs. Provisional population totals: India: census 2011. Available at http://censusindia.gov.in/2011-prov-results/indiaatglance.html - accessed 19 February 2013

Green TJ, Skeaff CM, Venn BJ, Rockell JE, Todd JM, Khor GL, Loh SP, Duraisamy G, Muslimatun S, Agustina R, Ling X, Xing X. Red cell folate and predicted neural tube defect rate in three Asian cities. Asia Pac J Clin Nutr. 2007;16(2):269-73.

Gulati R. Consanguineous marriages still high in Puducherry. The Hindu 2011, December 4.

Gunasekera DP, Gunasekera PC. Rubella immunisation—learning from developed countries. Lancet. 1996;347:1694–1695.

Habib S, Rahman AJEN, Choudhury T, Kabir AKMN. Clinical features and cytogenetic pattern of Down syndrome.Bangladesh Med Res Counc Bull.2007; 33: 108-10.

Han A M, Han KE, Myint TT. Thalassemia in the outpatient department of the Yangon Children's Hospital in Myanmar: cost analysis of the day-care-room services for thalassemia. Southeast Asian J Trop Med Public Health. 1992;23(2): 273-7.

Harano K, Win N , Harano T. Molecular Aspects of Transfusion Dependent Thalassemic Children in Myanmar: Analysis of Common .BETA.-Thalassemia in Myanmar by Amplification Refractory Mutation System (ARMS). Kawasaki Med J.2000;26(4):161-164.

Harano T. An insight into heterozygous β-thalassemia, Myanmar.Kawasaki Med J. 2000; 26: 127-131.

Helen Keller International.Indonesia. 2012. Available at http://www.hki.org/working-worldwide/asia-pacific/indonesia/ - accessed 19 February 2013.

Hettiarachchi M, Liyanage C. Coexisting micronutrient deficiencies among Sri Lankan pre-school children: a community-based study. Matern Child Nutr. 2012 Apr;8(2):259-66.

Ho NK. Perinatal infections, problems in developing countries. Singapore Med J. 1998 Jun;39(6):266-70.

Hossain J. Congenital heart defects. The Independent, Dhaka 16/6/2010.

Howell SR, Barnett AG, Underwood MR. The use of pre-conceptional folic acid as an indicator of uptake of a health message amongst white and Bangladeshi women in tower hamlets east London. Oxford: Oxford University Press, 2001.

International Council for the Control of Iodine Deficiency Disorders (ICCIDD).Global Iodine Nutrition Scorecard for 2012. Available at http://www.iccidd.org/ - accessed 10 April 2013.

International Institute for Population Sciences (IIPS) and Macro International. National Family Health Survey (NFHS-3), 2005–06: India: Volume I.Mumbai: IIPS, 2007.

Jain U, Verma IC, Kapoor AK: Prevalence of fragile X(A) syndrome in mentally retarded children at a genetics referral centre in Delhi, India. Indian J Med Res. 1998;108:12–16.

Japan International Cooperation Agency Planning and Evaluation Department.JICA country profile on disability, Kingdom of Nepal. 2002

Jaruratanasirikul S, Soponthammarak S, Chanvitan P, Limprasert P, Sriplung H, Leelasamran W, et al. Clinical abnormalities, intervention program, and school attendance of Down syndrome children

in southern Thailand. J Med Assoc Thai.2004; 87: 1199-204.

Jayasekara RW. The attitudes of doctors and students towards a genetic service in an Asian country: Sri Lanka.Asia-Oceania J Obstet Gynaecol. 1989;15:267–270.

Joint Report by Nutrition Section-Directorate of Health Services-Royal Government of Bhutan, International Council for Control of Iodine Deficiency Disorders (ICCIOD), Clinical Epidemiology Unit-AIIMS; UNICEF Thimphu, WHO Thimphu, WHO-SEARO and the Micronutrient Initiative (MI). Tracking progress towards sustainable elimination of iodine deficiency disorders in Bhutan.Ottawa, 1996.

Jung SC, Kim SS, Yoon KS, Lee JS. Prevalence of congenital malformations and genetic diseases in Korea. J Hum Genet. 1999;44(1):30-4.

Kapil U. Successful efforts toward elimination iodine deficiency disorders in India. Indian J Community Med. 2010 Oct;35(4):455-68.

Kapil U, Sareen N. Combating iodine deficiency disorders to achieve millennium development goal 4 in India: Reduction in infant mortality rate. Journal of Trace Elements in Medicine and Biology 2012; 26: 145– 148.

Kapoor S, Kabra M. Newborn screening in India: current perspectives. Indian Pediatr. 2010 Mar;47(3):219-24.

Kasemsuwan L, Cheewaruangroj W, Tungkeeratichai J, Bhongmakapat T, Thawin C, Lertsukprasert K, et al. Fourteenyears experience with cochlear implantation in Ramathibodi Hospital. J Med Assoc Thai.2010 Dec;93(12):1399-405.

Kaufman ND Chasombat S, Tanomsingh S, Rajataramya B, Potempa K. Public health inThailand: Emerging focus on non-communicable diseases. Intl J Health Planning Management. 2011;26(3):e197–e212.

Kaur M, Das GP, Verma IC: Inborn errors of amino acid metabolism in North India. J Inherit Metab Dis. 1994;17:230–233.

Khambalia A, O'Connor DL, Zlotkin S. Periconceptional Iron and Folate Status Is Inadequate among Married, Nulliparous Women in Rural Bangladesh. J Nutr. 2009: 139: 1179–1184

Khan N. Effects of parental consanguinity on offspring mortality in Bangladesh. Dhaka: International Centre for Diarrhoeal Diseas Research, 1997 (unpulished paper)

Khan WA. Banu B, Amin SK. Prevalence of β-thalassaemia trait & Hb E trait in Bangladesh school children and health burden of thalassemia in our population. Dhaka Shishu (Child) Hosp J.2005; 21(11):1-7.

Khanum S, Noor K, Kawser CA. Studies on congenital abnormalities and related risk factors. Mymensingh Med J. 2004 Jul;13(2):177-80.

Kitisomprayoonkul N, Tongsong T. Neural tube defects: a different pattern in Northern Thai population. J Med Assoc Thai. 2001;84:483-8.

Kulkarni ML ,Mathew MA , Ramachandran B. High incidence of neural tube defects in South India. The Lancet.1987; 1260.

Kulkarni ML, Mathew MA, Reddy V. The range of neural tube defects in southern India. Arch Dis Child. 1989;64:201-204

Kumta NB. Inborn errors of metabolism (IEM): an Indian perspective. Indian J Paediatr. 2005;72:325-332.

Labrique AB, Massie A, Rashid M et al.Community–based estimation of prevalence of birth defects in rural Bangladesh. 8th CCPDM. 2007

Lamlertkittikul S, Chandeying V. Experience on triple markers serum screening for Down's syndrome fetus in Hat Yai, Regional Hospital. J Med Assoc Thai. 2007 Oct;90(10):1970-6.

Lindström E, Persson LA, Raqib R, El Arifeen S, Basu S, Ekström EC. Prevalence of anemia and micronutrient deficiencies in early pregnancy in rural Bangladesh, the MINIMat trial.

Proc Natl Acad Sci USA. 2010 Dec 21;0107(51):22360.

Lokare PO, Karanjekar VD, Gattani PL, Kulkarni AP. A study of prevalence of anemia and sociodemographic factors associated with anemia among pregnant women in Aurangabad city, India. Ann Nigerian Med [serial online] 2012 [cited 2013 Feb 8];6:30-4.

Madan N, Sharma S, Sood SK, Colah R, Bhatia HM. Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India. Indian J Hum Genet. 2010; 16:16-25

Madhok R. In Maldives, UNICEF supports efforts by the government to fight malnutrition. 2012. Available at http://www.unicef.org/infobycountry/maldives_62471.html - accessed 21 February 2013.

Maheshwari M, Arora S, Kabra M, Menon PS. Carrier screening and prenatal diagnosis of beta-thalassemia. Indian Pediatr. 1999 Nov;36(11):1119-25.

Malla BK One year review study of congenital anatomical malformation at birth in Maternity Hospital (Prasutigriha), Thapathali, Kathmandu. Kathmandu University Medical Journal. 2007;5(4):557-60.

Manandhar SR1, Ojha A1, Manandhar DS2, Shrestha B2, Shrestha D2, Saville N3, Costello AM3, Osrin D3 Causes of stillbirths and neonatal deaths in Dhanusha district, Nepal: A verbal autopsy study. Kathmandu University Medical Journal. 2010;8(1):62-72.

Marzuki S. Thalassemia research network in Indonesia. Eijkman Institute for Molecular Biology, Jl. Diponegoro 69, Jakarta.PHTDI - Perhimpunan Hematolgi dan Transfusi Darah, Indonesia.

Masloman N, Mustadjab I, Munir M. Congenital malformation at Gunung Wenang Hospital Manado: a five-year spectrum. Paediatr Indones. 1991 Nov-Dec;31(11-12):294-302.

Ministry of Health and Family Welfare, Bangladesh. Health information system assessment: Bangladesh country report. Dhaka: Health Metrics Network Secretariat, 2009.

Ministry of Public Health, Democratic People's Republic of Korea. Annual report of the

health state, DPR Korea, 2007. Pyongyang, 2007.

Ministry of Health,Government of India. Report of Working group on disease burden for 12th Five year plan. WG3-non-communciable diseases. 2011

Ministry of Health, Republic of Indonesia. Indonesia health profile 2008. Jakarta: Ministry of Health, 2010.

Ministry of Health, Republic of Indonesia. Riskesdas,National basic health research, 2007. Jakarta: Ministry of Health, 2008.

Ministry of Health and Family (MOHF)[Maldives and ICF Macro. Maldives demographic and health survey 2009. Calverton, Maryland: MOHF and ICF Macro, 2010.

Ministry of National Planning and Economic Development and Ministry of Health Myanmar. Myanmar Multiple Indicator Cluster Survey 2009 - 2010 Final Report. Nay Pyi Taw: Ministry of National Planning and Economic Development and Ministry of Health, Myanmar, 2011.

Ministry of Health and Population (MOHP) Nepal, New ERA, and ICF International Inc. Nepal Demographic and Health Survey 2011.

Kathmandu, Nepal: Ministry of Health and Population, New ERA, and ICF International, Calverton, Maryland, 2012.

Ministry of Health and Population (MOHP) Nepal, New ERA, and Macro International Inc. Nepal demographic and health survey 2006. Kathmandu: Ministry of Health and Population, New ERA, and Macro International Inc, 2007.

Modell B, Kuliev AM. The impact of public health on human genetics. Clin Genet. 1989; 36:286-289.

Modell B, Kuliev AM, Wagner M. Community genetics services in Europe. WHO Regional Publication. European Series No. 38.WHO European Regional Office. Copenhagen: World Health Organization, Regional Office for Europe, 1992.

Modiano G, Morpurgo G, Terrenato L et al. Protection against malaria morbidity: near-fixation of the alphathalassemiagene in a Nepalese population. Am J Hum Genet. 1991.48: 390-397.

Modiano D, Chiucchiuini A, Petrarca V, et al. Humoral response to Plasmodium falcifarum Pf155/ring-infected erythricyte surface antigen and Pf332 in three symphatric ethnic groups of Burkino Faso. Am J Trop Med Hyg. 1999.58,220-224.

Mohanty D, Colah R, Gorakshakar Jai Vigyan S & T Mission Project on community control of thalassemia syndromes - Awareness, screening, genetic counseling and prevention. New Delhi: Indian Council of Medical Research; 2008.

Moosa MM, Ayub MI, Bashar AE, Sarwardi G, Khan W, Khan H, Sabina Yeasmin S, Combination of two rare mutations causes thalassaemia in a Bangladeshi patient Genetics and Molecular Biology (Online Ahead of Print).

National Institute of Population Research and Training (NIPORT), Mitra and Associates, and Macro International. Bangladesh Demographic and Health Survey 2007. Dhaka, Bangladesh and Calverton, Maryland, USA: National Institute of Population Research and Training, Mitra and Associates, and Macro International, 2009.

National Statistics Bureau, Royal Government of Bhutan. Bhutan Multiple Indicator Survey 2010. Thimphu, Bhutan : National Statistics Bureau, 2011.

National Statistics Directorate (NSD) Timor-Leste, Ministry of Finance Timor-Leste, and ICF Macro. Timor-Leste Demographic and Health Survey 2009-10. Dili, Timor-Leste: NSD Timor-Leste and ICF Macro,2010.

National Neonatology Forum. National neonatal-perinatal database. Report for the year 1995. New Delhi: Department of Pediatrics, All India Institute of medical Sciences, 1996.

National Planning Commission Secretariat, UNICEF, New ERA Ltd. Nepal - a situation analysis on disability Volume–I. New Delhi: National Planning Commission Secretariat, 1999.

Nawapun K, Phupong V. Awareness of the benefits of folic acid and prevalence of the use of folic acid supplements to prevent neural tube defects among Thai women. Arch Gynecol Obstet. 2007 Jul;276(1):53-7.

Ne-Win,Harano K, Harano T, Myint T. T , Mra R, Myint A. A, Shimono K , Okada S. Common α-Thalassemia deletions in transfusion-dependent thalassemia patients in the Southeast Asia Region of Myanmar. Lab Hematol. 2006;12:139-142.

Office of the Census Commissioner. Population and housing census. Thimphu: Royal Government of Bhutan, 2005.

Pandav CS, Rasheed M, Solih I, Saeed M, Shaheed M, Awal A, Anand K, Shreshta R. Iodine deficiency disorders in the Maldives: A public health problem. Asia Pacific Journal of Clinical Nutrition; 1999: 8: 9 – 12.

Patel ZM, Adhia RA. Birth defects surveillance study. Indian J Pediatr. 2005;72:489-91.

Penchaszadeh VB. Preventing congenital anomalies in developing countries. Community Genetics. 2002;5:61-69.

Perera DMD, Gunasekara, Wijekoon A, Petrou M. Thalassaemia control in developing countries - the Sri Lankan perspective. Ceylon Medical J.2000;45:12-16.

Peto H, Labrique A, Khatry SK, Shahabuddin M, Christian P, Ali H, Klemm R, Shaikh S, Shamim AA, LeClerq SC, Mehra S, West KP Jr. Implementation of community birth defects surveillance systems in rural Nepal (1994-1997) and Bangladesh (2001-2007), 2012 (unpublished paper).

Piyamongkol W , Vutyavanich T, Piyamongkol S, Wells D, Kunaviktikul C, Tongsong T, Chaovisitsaree S, Saetung R, Sanguansermsri T. A successful strategy for preimplantation genetic diagnosis of beta-thalassemia and simultaneous detection of Down's syndrome using multiplex fluorescent PCR. J Med Assoc Thai. 2006; 89(7): 918-27.

Pradhan P, Poudel S, Maharjan A. Still-birth - a tragic journey: a critical analysis. Nepal Med Coll J. 2010 Dec; 12(4):239-43.

Prathanee B, Chowchuen B. Community-based network system and interdisciplinary management for children with cleft-lip/palate. J Med Assoc Thai. 2010 Oct;93(Suppl 4):S63-70

Premawardhena A, de Silva S, Arambepola M, Olivieri N, Merson L, Julia Muraco J,Allen A,Fisher C,Peto T, Vichinsky E and Weatherall D. Thalassemia in Sri Lanka: a progress report. Human Molecular Genetics. 2004;13(2): R203–R206.

Pruksanusak N, Suwanrath C, Kor-anantakul O, Prasartwanakit V, et al. A survey of the knowledge and attitudes of pregnant Thai women towards Down syndrome screening. Journal of Obstetrics and Gynaecology Research. 2009;35(5):876–881.

Puri RD, Verma IC. Genetic services in India: model for developing countries. In: Kumar D, editor. Genomics and Health in the Developing World. Oxford: Oxford University Press. 2012. p 927-935.

Rahman, Omar, Jane Menken, Andrew Foster, and Paul Gertler.Matlab [Bangladesh] Health and Socioeconomic Survey (MHSS), 1996. ICPSR02705-v5. Ann Arbor, MI: Inter-university Consortium for Political and Social Research [distributor], 2001.

Rahman MM, Khan AM, Hafiz MM, Ronny FM, Ara S, Chowdhury SK, Nazir SS, Khan WI. Congenital hearing impairment associated with rubella: lessons from Bangladesh. Southeast Asian J Trop Med Public Health. 2002 Dec;33(4):811-7.

Rashid SQ. A study of fetal anomalies detected by ultrasound in Bangladesh and their relative frequencies. J R Soc Promot Health. 2002 Mar;122(1):55-7.

Rasnayaka M, Mudiyanse RM. Thalassemia treatment and prevention in Uva Province, Sri Lanka: apublic opinion survey hemoglobin. 2006;30(2):275-89.

Ratanasiri T, Charoenthong C, Komwilaisak R, Changtrakul Y MSc,Fucharoen S, Wongkham J, Kleebkaow P, Seejorn K. Prenatal prevention for severe thalassemia disease at Srinagarind Hospital. J Med Assoc Thai. 2006;89 (suppl4): S87-93.

Ratanasiri T, Piensriwatchara E, Prasertcharoensuk W. Birth prevalence of neural tube defects at Srinagarind Hospital, 1988-1996. Srinagarind Med J. 1997;12:139-43.

Ravikumara M, Bhat BV, Oumachigui A. Perinatal mortality trends in a referral hospital. Indian J Pediatr. 1996;63:357-61.

Retrospective rubella review: Maldives. Weekly Epidemiological Record. 2005; 80(10):88-92

Riewpaiboon A, Nuchprayoon I, Torcharus K, Indaratna K, Thavorncharoensap M, Ubol BO. Economic burden of beta-thalassemia/Hb E and beta-thalassemia major in Thai children. BMC Res Notes. 2010 Jan 30;3:29.

Royal Government of Bhutan Nutrition Section, Directorate of Health Services, ICCIDD, All India Institute of Medical Sciences, UNICEF, WHO, MI. Tracking progress towards sustainable elimination of iodine deficiency disorders in Bhutan. New Delhi, 1996.

Rueangchainikhom W, Sarapak S, Orungrote N. Chorionic villus sampling for early prenatal diagnosis at Bhumibol Adulyadej Hospital. J Med Assoc Thai. 2008;91(1): 1-6.

Rustama DS, Fadil MR, Harahap ER, Primadi A. Newborn screening in Indonesia. Southeast Asian J Trop Med Public Health. 2003;34 (Suppl 3):76-9.

Sachdev HP, Gera T. Preventing childhood anemia in India: iron supplementation and beyond. Eur J Clin Nutr. 2013 Feb 6. doi: 10.1038/ejcn. 2012.212.

Sakai Y, Kobayashi S, Shibata H et al. Molecular analysis of alpha-thalassemia in Nepal: correlation with malaria endemicity. J Hum Genet.2000; 45: 127-132.

Sayasathid J, Tantiwongkosri K, Somboonna N. Unrecognized congenital heart disease among Thai children. Cardiac Center, Naresuan University Hospital, Phitsanulok, Thailand. J Med Assoc Thai. 2009 Mar; 92(3):356-9.

Senanayake H, Goonewardene M, Ranatunga A, Hattotuwa R, Amarasekera S, Amarasinghe I. Achieving Millennium Development Goals 4 and 5 in SriLanka. BJOG. 2011;18(Suppl 2):78-87.

Sever LE, editor. Guidelines for surveillance of birth defects. National Birth Defects Prevention Network.

Sharma D, Gupta M, Thelma BK. Expansion mutation frequency and CGG/GCC repeat polymorphism in FMR1 and FMR2 genes in an Indian population. Genet Epidemiol. 2001;20:129-44.

Shrestha U, Bhattacharya S, Bhatta NK, Jha CB. Cytogenetic analysis of children with suspected genetic disorder. Kathmandu Univ Med J (KUMJ). 2009 Jan- Mar;7(25):40-3.

Simpson B. Negotiating the therapeutic gap: prenatal diagnostics and termination of pregnancy in Sri Lanka. Bioethical Enquiry. 2007;9:207-215.

Sirichotiyakul S MD, Piyamongkol W MD, Tongprasert F MD, Srisupandit K MD,Luewan S MD Transabdominal Chorionic Villus Sampling: Experience at Maharaj Nakorn Chiang Mai Hospital J Med Assoc Thai. 2008;91(6):813-7

Sirikulchayanonta C, Madjupa K, Chongsuwat R, Pandii W. Do Thai women of child bearing age need pre-conceptional supplementation of dietary folate? Asia Pac J Clin Nutr. 2004;13 (1): 69-73.

Sithey G. 2006. Progress towards sustainable elimination of IDD in Bhutan—lessons to be shared. IDD News Vol. IV, Issue 1, pp. 10–12.

South East Asia Regional Neonatal Perinatal Database (SEAR-NPD). Report, 2007-08.

SRS (Sample Registration System) Bulletin. 2011 Dec; 46(1): 1-6.

Statistics Indonesia (Badan Pusat Statistik—BPS) and Macro International. Indonesia Demographic and Health Survey 2007. Calverton, Maryland, USA: BPS and Macro International, 2008

Stoll C, Alembik Y, Dott B, Feingold J. Parental consanguinity as a cause of increased incidence of birth defects in a study of 131, 760 consecutive births. Amer J Med Genet. 1994;49:114-117.

Suresh S, Thangavel G, Sujatha J, Indrani S. Methodological issues in setting up a surveillance system for birth defects in India. Natl Med J India. 2005;18(5):259-62 .

Suryantoro P. Glucose-6-phosphate dehydrogenase (G6PD) deficiency in Yogyakarta and its surrounding areas. Southeast Asian J Trop Med Public Health. 2003;34 (Suppl 3):138-9.

Suwanrath-Kengpol C, Kor-anantakul O, Suntharasaj T, Leetanaporn R. Etiology and outcome of non-immune hydrops fetalis in southern Thailand. . Gynecol Obstet Invest. 2005;59(3):134-7.

Suwatanaviroj A, Ratrisawadi V. Factors associated with congenital malformations in Thailand. J Med Assoc Thai. 1996 Sep; 79(9): 545-9.

Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformations at birth in Central India: A rural medical college hospital based data. Indian J Hum Genet. 2010; 16:159-63.

Taweevisit M, Thorner PS. Hydrops fetalis in the stillborn: a series from the central region of Thailand. Pediatr Dev Pathol. 2010 Sep-Oct;13(5):369-74.

Thailand National Statistical Office. Report on 2005-2006 survey of population change. Bangkok: Statistical Forecasting Bureau, 2007.

Thailand National Statistical Office. Thailand Multiple Indicator Cluster Survey December 2005- February 2006, Final Report. Bangkok, Thailand: National Statistical Office, 2006.

Thallasaemia International Federation (TIF). Prevention of Thallasaemias and other haemoglobin disorders. Nicosia, 2003.

Than AM, Harano T, Harano K, Myint A. A, Ogino T and Okadaa S. High Incidence of α-Thalassemia, Hemoglobin E, and Glucose-6-Phosphate Dehydrogenase Deficiency in Populations of Malaria-Endemic Southern Shan State, Myanmar. Internat J Hematol. 2005;82:119-123.

Thant KZ, Oo WM, Myint TT, Shwe TN, Han AM, Aye KM, Aye KT, Moe K, Thein S,Robertson SE. Active surveillance for congenital rubella syndrome in Yangon, Myanmar. Bull World Health Org. 2006;84(1):12-20.

The National Institute of Health Research and Development. Indonesia report on result of national basic health research. Jakarta: Ministry of Health, Republic of Indonesia, 2008.

The Micronutrient Initiative and UNICEF. Vitamin and Mineral Deficiency: A damage assessment report for Bangladesh. Ottawa, Ontario, 2004a.

The Micronutrient Initiative and UNICEF. Vitamin and mineral deficiency: aglobal progress report. Ottawa, Ontario, 2004b, p. 20.

The Micronutrient Initiative. Investing in the future: A united call to action on vitamin and mineral deficiencies: Global report 2009. Ottawa, New York, 2009.

The Micronutrient Initiative and UNICEF. Vitamin & mineral deficiency: a damage assessment report for Bangladesh. Ottawa, New York, 2009.

Titumir RAM, Hossain J. Disability in Bangladesh-prevalence, knowledge, attitudes, and Practices. Unnayan Onneshan. 2005. http://www.unnayan.org/report - accessed 19 February 2013.

Toteja GS, Singh P, Dhillon BS et al. Prevalence of anemia among pregnant women and adolescent girls in 16 districts of India. Food and Nutrition Bulletin 2006; 27(4):311-315.

Turnpenny PD, Ellard S. Emery's Elements of Medical Genetics. Elsevier Churchill Livingstone, 2005.

Unachak K, Tanpaiboon P, Pongprot Y, Sittivangkul R, Silvilairat S, Dejkhamron P, Sudasna J. Thyroid functions in children with Down's syndrome. J Med Assoc Thai. 2008 Jan;91(1):56-61.

University of Sydney, Initiative for Cardiovascular Health Research in the Developing Countries, Mailman School of Public Health, the Earth Institute at Columbia University. A Race Against Time: The Challenge of Cardiovascular Disease in Developing Economies. New York: Trustees of Columbia University; 2004.

United Nations Economic and Social Commission for Asia and Pacific (UNESCAP). Disability at a glance: a profile of 28 countries and areas in Asia and the Pacific. Bangkok, 2006.

United Nations Economic and Social Commision for Asia and Pacific (UNESCAP) Project on Improvement of Disability Measurement and Statistics. Pilot test. Maldives . 2009.

United Nations Economic and Social Commission for Asia and Pacific (UNESCAP). Disability at glance 2012: strengthening the Evidence Base in Asia and Pacific. Bangkok, 2012.

United Nations Children's Fund (UNICEF). Maternal, newborn & child survival: country profile. New York: UNICEF, 2010.

United Nations Children's Fund (UNICEF). State of the world's children 2012: children in an urban world. New York: UNICEF, 2012.

United Nations Inter Agency Group for Child Mortality Estimation- http:// www.childinfo.org/ index.html.accessed Sept 17, 2012

Upazila M. Report on Seventeen 30-Cluster EPI Coverage Evaluation Survey in the CHT. UNDP, 2010.

Upreti SR, Thapa K, Pradhan YV, Shakya G, Sapkota YD, Anand A, Taylor T, Mach O,Reef S, Pattamadilok S, Liyanage J, O'Connor P, Sedai T, Bhandary SR, Partridge J, Schluter W. Developing rubella vaccination policy in Nepal - results from rubella surveillance and seroprevalence and congenital rubella syndrome studies. J Infect Dis. 2011 Jul; 204 (Suppl 1):S433-8.

United States Agency for International Development (USAID)/Indonesia Nutrition Assessment for 2010. New Project Design

Verma IC, Bijarnia S. The burden of genetic disorders in India, and a framework for community control. Community Genet. 2002;5:192-196.

Verma IC: Burden of genetic disorders in India. Indian J Pediatr. 2000;67:893–898.

Verma IC. Genetic causes of mental retardation. In: Niermeijer M, Hicks E, editors. Mental retardation, genetics and ethical considerations. Amsterdam, 1988. pp 99–106.

Verma IC. High frequency of neural tube defects in India. Lancet. 1978a;i:879–880.

Verma IC. Neural tube defects in India. In: Verma IC, editor. Medical genetics in India. Pondicherry, Auroma Enterprises. 1978b; 1:33–40.

Verma IC, Anand NK, Modi UJ, Bharucha BA: Study of malformations and Down syndrome. in India – A multi-centric study. Mumbai, Department of Atomic Energy, and Trombay, Bhabha Atomic Research Center, 1998.

Verma IC, Kumar D. Epidemiology of genetic diseases in the Indian subcontinent. In: Kumar D, editor. Genomics and health in the

developing world. Oxford University Press. 2012. Pp 923-926.

Verma IC, Saxena R, Kohli S. Past, present & future scenario of thalassaemic care & control in India. Indian J Med Res. 2011 Oct; 134; 507-521.

Viveki RG, Halappanavar AB, Viveki PR, Halki SB, Maled VS. Deshpande PS. Prevalence of anaemia and its epidemiological determinants in pregnant women. Al Ameen Journal of Medical Sciences. 2012; 5 (3):216-223.

Vutyavanich T, Piromlertamorn W, Ellis J. Assisted reproductive technologies in Thailand: 2001-2007 results generated from the ART Registry, Royal Thai College of Obstetricians and Gynaecologists. J Obstet Gynaecol Res. 2011 Mar;37(3):236-44

Wahidayat PA, Gatot D, Tjitrasari T, Ringoringo HP, Marzuki NS, Taufani RA, Setianingsih I, HArahap A. Phenotypic diversity in beta-HbE thalassemia patients. Paediatrica Indonesiana, Vol. 46, No. 3-4 • March - April 2006

Walia GK. Disability in India. Version 4. South Asia Network for Chronic Diseases.

Wanapirak C MD, Muninthorn W MD, Sanguansermsri T MD, Dhananjayanonda P MSc, Tongsong T MD. Prevalence of thalassemia in pregnant women at Maharaj Nakorn Chiang Mai Hospital. J Med Assoc Thai. 2004;87(12):1415-8.

Wasant P. Approach, counseling and care of down syndrome children at Siriraj Hospital. Thai J Pediatr. 1993;32:77-82.

Wasant P. Children with delayed development. Thai J Pediatr. 1994;33:243-7.

Wasant P, Liammongkolkul S. Prenatal genetic screening for Down syndrome and open neural tube defects using maternal serum markers in Thai pregnant women. Southeast Asian J Trop Med Public Health. 2003;34 Suppl 3:244-8.

Wasant P, Sathienkijkanchai A. Neural tube defects at Siriraj Hospital, Bangkok, Thailand - 10 years review (1990-1999). J Med Assoc Thai. 2005;88 (Suppl 8):S92-9

Wasant P. Wiwatkunupakarn J. The ten-year study of Down syndrome at Siriraj Hospital (1977-1986). Siriraj Hosp Gaz 1994; 46: 696-705.

Win N, Harano T, Harano K, Myint TT, Mra R, Okada S, Shimono K, Myint AA.A wider molecular spectrum of beta-thalassaemia in Myanmar. Br J Hematol. 2002;117(4):988-92.

Win N, Lwin AA, Aye KS, Soe S, Okada S. Hemoglobin E prevalence in malaria-endemic villages in Myanmar. Acta Medica Okayama. 2005;9(2).

World Bank. World development report 1993: investing in health. New York: Oxford University Press, 1993.

World Bank. NCDs Policy Brief: Nepal. 2011. Available at <siteresources.worldbank.org/SOUTHASIAEXT/ rces/223546-129668007256/7707437-1296680114157/NCD_NP_Policy_Feb_2011.pdf> Accessed 9 April 2013.

World Health Organization. Community approaches to the control of hereditary diseases: report of the advisory group. Geneva, 1985.

World Health Organization. Control of hereditary diseases. WHO Technical Report Series 865. Geneva, 1996.

World Health Organization. Primary health care approaches for prevention and control of congenital disorders: report of a WHO meeting, Cairo, Egypt, 6-8 December. Geneva: WHO, 1999a

World Health Organization. Services for the prevention and management of genetic disorders and birth defects in developing countries: report of a Joint WHO/WAOPBD, The Hague, January 1999b

World Health Organization, Regional Office for the Eastern Mediterranean. Community control of genetic and congenital disorders. Cairo, 2000.

World Health Organization. Preventing chronic diseases: a vital investment. Geneva: WHO, 2005.

World Health Organization. Management of birth defects and haemoglobin disorders: report of a Joint WHO-March of Dimes Meeting. Geneva, 2006a.

World Health Organization. WHO Global Database on Iodine Deficiency: Maldives. 2006b. Available at http://who.int/vmnis/iodine/data/database/countries/mdv_idd.pdf.accessed 21 February 2013.

World Health Organization. Worldwide prevalence of anemia 1993–2005. Country estimates of anaemia in non-pregnant women of reproductive age WHO global database on anaemia. Geneva: WHO, 2008.

World Health Organization. Global prevalence of vitamin A deficiency in populations at risk 1995-2005: WHO global database on vitamin A deficiency. Geneva: WHO, 2009.

World Health Organization -Birth Defects, EB126 R6, notes. 2010a

World Health Organization. Sixty –Third World Health Assembly. A63/10, April 2010b, Report by Secretariat.

World Health Organization. Child Health Epidemiology Reference Group (CHERG). Geneva: WHO, 2010c.

World Health Organization. World health statistics 2011. Geneva: WHO, 2011.

World Health Organization, Country Office for Bangladesh. Non-communicable disease risk factor survey Bangladesh 2010. Dhaka, 2011.

World Health Organization. World health statistics 2012. Geneva: WHO, 2012.

World Health Organization. Country profile: Bhutan. Geneva. Available at http://www.who.int/countries/btn/en/ - accessed 19 February 2013.

World Health Organization, Country Office for DPR Korea. Country profile of maternal, newborn and child health fact file.

World Health Organization, Country Office for DPR Korea. Country Cooperation Strategy at a glance. 2011. Available at http://www.who.int/countryfocus/cooperation.../ccsbrief_krd_en. pdf Accessed 9 April 2013.

World Health Organization, Country Office for Indonesia. Indonesia, Non-communicable diseases. http://www.ino.searo.who.int/en/Section3_30.htm - accessed 19 February 2013.

World Health Organization. Global Health Observatory.Available at http://www.who.int/gho/child_health/en/index.html - accessed November 2012.

World Health Organization, Regional Office for South-East Asia. WHO country cooperation strategy: Democratic People's Republic of Korea: 2009-2013. New Delhi, 2010.

Yang JH, Kim YJ, Chung JH, Kim MY, Ryu HM, Ahn HK, Han JY, Yang SH, Kim A, Kim HS, Lee PJ, Kim SS, Kim YJ, Koh KS, Shin JC, Cho YK, Yoon BH. A multi-center study for birth defect monitoring systems in Korea. J Korean Med Sci. 2004:19(4):509-13.

The World Health Assembly has expressed concern about the high number of stillbirths and neonatal deaths occurring worldwide, and the large contribution of neonatal mortality to under-five mortality. The Assembly recognized the importance of birth defects as a cause of stillbirths and neonatal mortality, and that the attainment of MDG 4 on reduction of child mortality will require accelerated progress in reducing neonatal mortality, including prevention and management of birth defects. Resolution WHA 63.17, adopted in May 2010, forms the basis of initiatives on the prevention of birth defects in the WHO South-East Asia Region. Epidemiological information is required to design effective preventive strategies for birth defects and evaluate them upon implementation. It has been observed that, at present, none of the Member States has nationally representative data and information related to birth defects.

In view of this, the WHO Regional Office for South-East Asia has developed situation analysis based on the published literature and information obtained from experts and national programme managers from the Member States. The information from different countries varies in extent but provides a reasonable insight on the burden and common types of birth defects, and surveillance and preventive activities for birth defects that may be available in the existing programmes in the Member States. Further opportunities for preventive services, challenges and support required to develop surveillance and preventive actions for birth defects in the countries are also described. The Regional situation analysis would be useful to develop prevention programmes for birth defects in the Member States of the Region.

World Health Organization
Regional Office for South-East Asia