
The Global Birth Defects Description and Coding (GBDDC) App

Results of the Basic Version Review

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Introduction

Surveillance systems and pregnancy cohort studies working in low resource environments have difficulty in obtaining accurate information about congenital anomalies (birth defects). The Zika epidemic has further emphasised the need for better birth defects surveillance systems in these settings. In response to this problem, the International Committee for Congenital Anomaly Surveillance Tools (comprising members from Europe, Latin America, Africa, Asia and the USA: <https://globalbirthdefects.tghn.org/about-us/steering-committee/>) developed an App to facilitate the accurate description and coding of birth defects. The App contains diagrams and photos of 92 types of externally visible birth defect, together with ICD10-RCPC code and text description. Special modules on microcephaly and Congenital Zika Syndrome are included.

There are two versions of the App:

Basic Version: Designed for use by persons with an interest in understanding/improving birth defect diagnosis or coding, including for training purposes. This version does not allow data to be recorded.

Surveillance Version: is an extension of the basic version to allow recording of anonymous data for each baby. It is meant to assist a wide range of health professionals contributing to surveillance systems and research in low resource settings.

This report presents feedback from expert review of the Basic Version of the app. The Surveillance Version is currently being field tested in Africa and South America and the results will be published later.

Review Process

At the end of May 2019, invitations were extended to review the Basic Version of the app to the ECLAMC network, the WHO Technical Committee for Birth Defects, and selected others. Those who agreed to review were provided with a feedback questionnaire to guide the review process. Reviewers were given 6 weeks to send in their feedback.

General Description of Reviewers

A total of 12 persons took part in the review of the Basic version of the GBDDC App. The reviewers included medical geneticists (5), epidemiologists/Birth defects researchers (6) and Obstetrician/Gynaecologist (1) From Africa, Europe, North America, South America and Australia.

Functionality of App

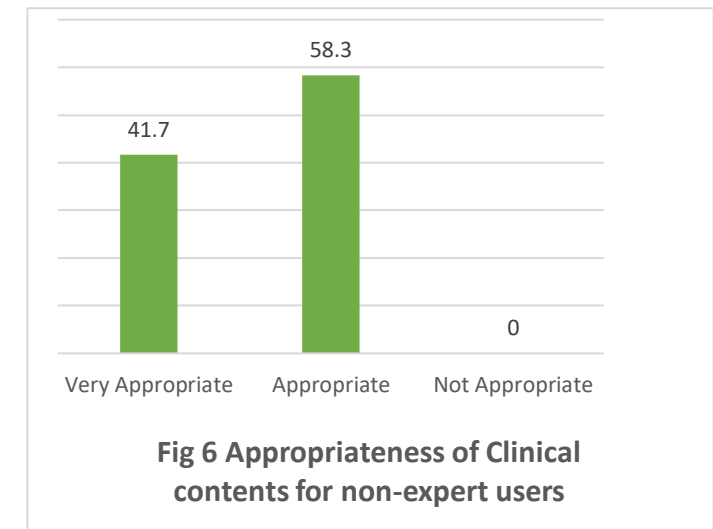
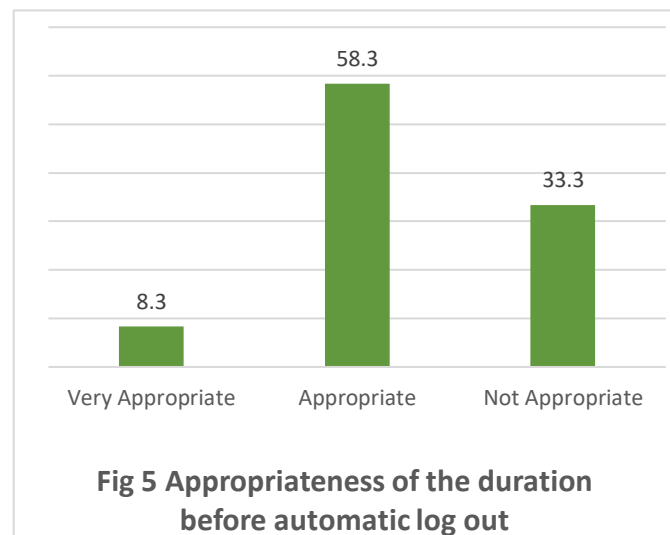
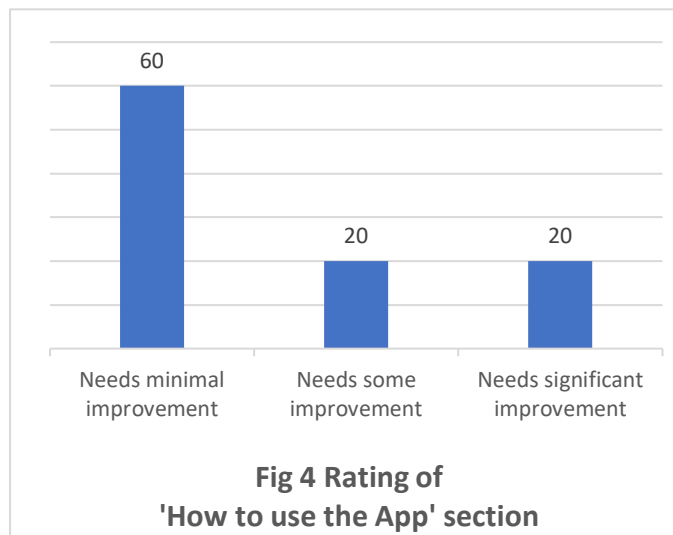
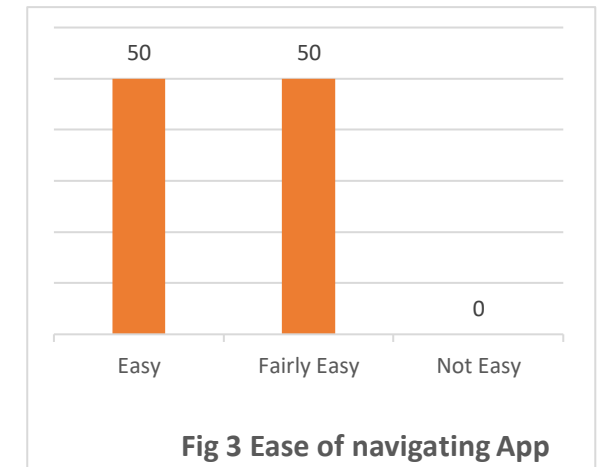
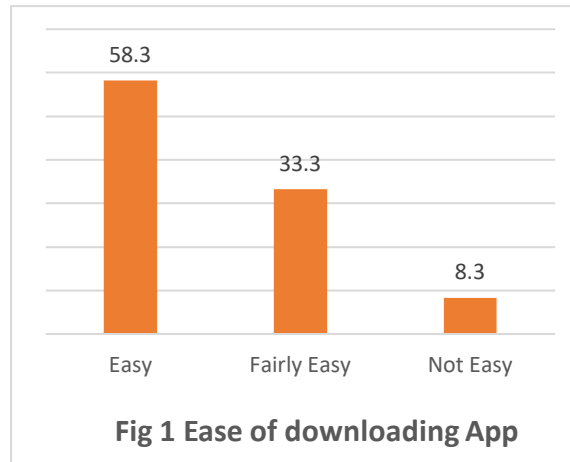
Most reviewers felt it was easy to download (58.5%) the app and easy to register (66.5%) to use the app (Fig 1 & 2). At the time of review, the Apple device users could access the app from the Apple Store, while Android users could only get the app from the App website and not Google Play Store. Apple users were generally satisfied with the process of searching and downloading the app from App Store while some Android devices users found it difficulty going through the download process outlined in the app website, including how to change the setting of their device to allow downloading from “unknown source”. Android users therefore strongly recommended that the app should be put in Google Play Store. Some reviewers also complained that the app was too heavy for mobile phone device and that it took a long time to download even with use of Wifi.

The main issues with registration process of the app were: more data was needed for registration than the usual username and password, causing delay in the registration process; the requirement for registration before using the app was not immediately obvious as one opens the app; pin was visible

when entering and some users wanted it invisible; and the registration and login process was not clearly separated.

Half of the reviewers felt the app was easy to navigate (Fig 3). Issues raised regarding app navigation include: app logs out while still in use; the Syndrome and examination video tabs were not obviously visible; and no dedicated button that takes user to the main home page or to the baby's image. Also, some reviewers were unsatisfied with the automatic logout time (5mins) set for the app, complaining it was too short (Fig 4). On average, users proposed that the time for automatic logout of the app after inactivity should be set at 15mins.

Most (60%) reviewers rated the section on "How to use the App" as appropriate (Fig 4). However, some reviewers thought there were items within the "About the App" section that could be moved over to "How to use the App" section. Some also thought the "About the App" section needs more structuring; that the images and labels were too tiny on a phone; and that the texts were too enlarged and difficult to read.



Clinical contents of the App

Overall, 100% of reviewers felt the clinical contents of the app was very appropriate (41,7%) or appropriate (58.3%) for non-expert users (Figure 6). On average, 66% of reviewers rated the contents of each body part as needing minimal improvement (Fig 7 a – j). In addition, they made the following suggestions/observations:

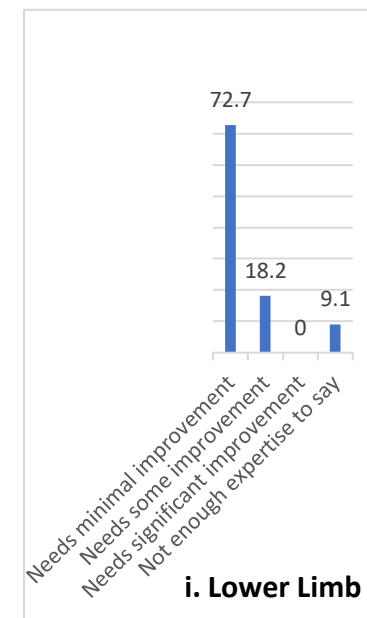
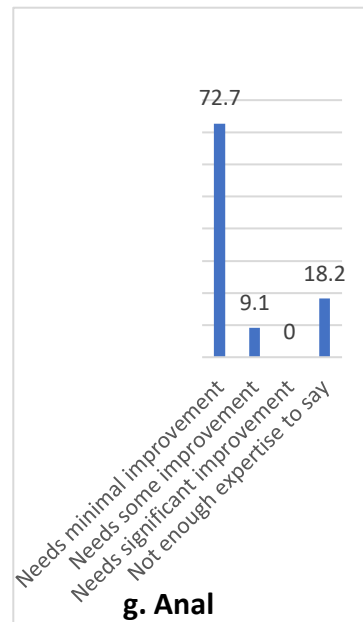
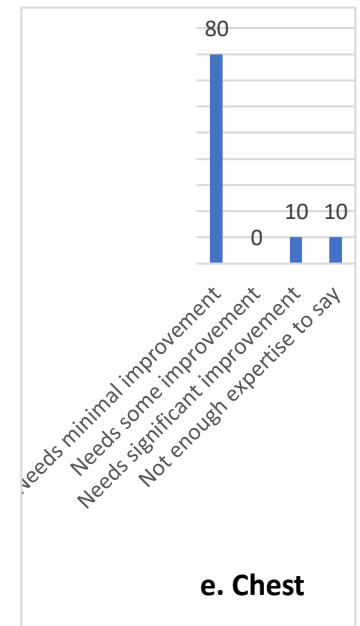
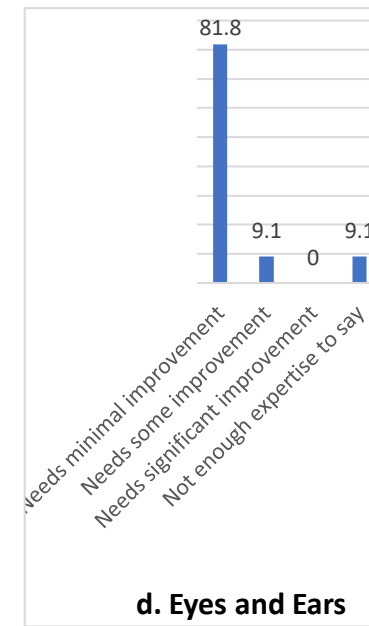
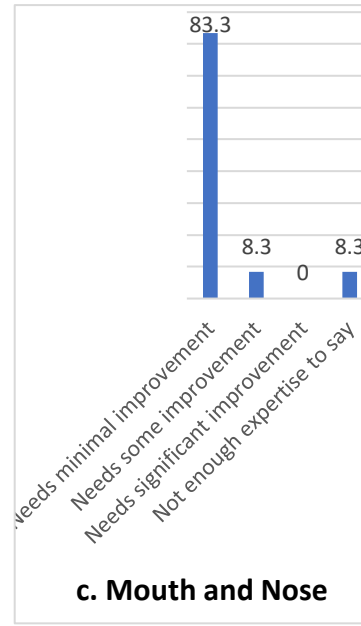
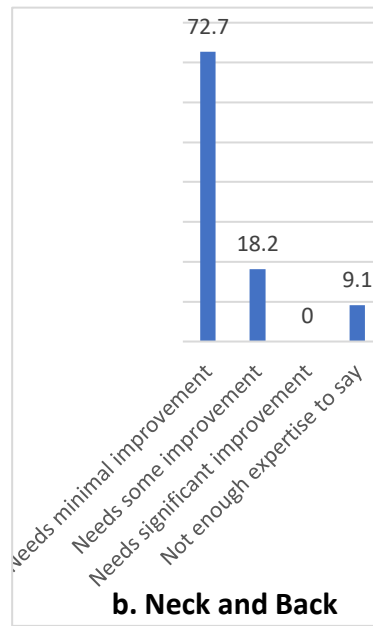
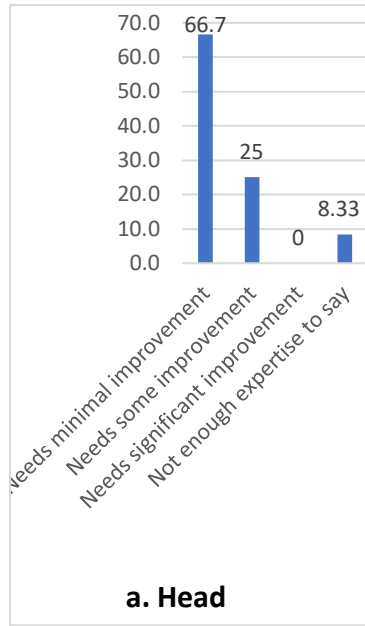
- *Main image selection page should contain only drawings or photos of a particular anomaly and not both*
- *ICD codes for minor anomalies should be added so they can be reported if not isolated*
- *State whether a condition is compatible with life*
- *Remove statement that says “anencephaly is lethal”*
- *App anomalies not harmonised with WHO atlas: e.g some birth defects present in the app are not included in atlas because they are too rare*
- *Some figures need more explanation on what viewers should focus on regarding the malformation*
- *Skeletal dysplasias quite difficult to differentiate by non-expert*
- *In case of sub-categories of birth defects, extend text of label to describe each*
- *Synonyms of birth defect terms should be placed in parentheses*

Also, reviewers proposed several conditions that could be added to the app (Table 1).

Table 1: Additional anomalies suggested by Reviewers

Body part	Suggested CA additions
Head	- <i>Cystic hygromas, and sacral pits with or without a background</i>
Neck & Back	<ul style="list-style-type: none"> - <i>Hidden bifid spine (minor, spina bifida occulta)</i> - <i>Vascular tumors or vascular anomalies</i> - <i>Pterigium colli/webbing of neck</i> - <i>lumbosacral pits,</i> - <i>scoliosis due to a congenital bone malformation,</i> - <i>Fusion or hemi vertebra or failure of vertebral segmentation with scoliosis</i> - <i>Branchial cleft/sinus/fistula/cyst (minor)</i>
Mouth & nose	<ul style="list-style-type: none"> - <i>Aglossia (Q38.30), to describe at least one anomaly of tongue / mouth cavity, besides the clefts.</i> - <i>Micrognathia (currently part of the Pierre-Robin Sequence) could be a standalone malformation?</i>
	<i>In “mouth other”</i> <ul style="list-style-type: none"> - <i>Macrostomia,</i> - <i>Microstomia, which can be observed inside the mouth: frénulas</i> - <i>Malar hypoplasia (or elsewhere?),</i>
Eye & ears	<ul style="list-style-type: none"> - <i>Anquiblepharon/ epiblepharon</i> - <i>Hyper or ocular hypotelorism, pustules and preauricular appendages.</i>

Chest	<ul style="list-style-type: none"> - Q67.6- Pectus excavatum, Congenital Funnel Chest and - Q67.7- Pectus carinatum, Congenital pigeon chest - Costal grill; and pectoral hypoplasia / agenesis, that can be observed alone or associated with other anomalies. - Thorax in keel, excavated thorax - Cantrell pentalogy
Abdomen	<ul style="list-style-type: none"> - Constriction band sequence spectrum that involves abdominal/thoracic region. - Congenital malformations of the digestive system: esophageal atresia, atresia of the small intestine. - Umbilical hernia - Limb body wall complex/body stalk anomaly
Anal	<ul style="list-style-type: none"> - (Q43.5)- Ectopic anus - Absence, atresia and congenital stenosis of the rectum and anus with / without a fistula (imperforation of the rectum and anal imperforation)
Genitourinary	<ul style="list-style-type: none"> - Potter sequence
	<ul style="list-style-type: none"> - Peno-scrotal and Scrotum Transposition in Scarf - And cryptorchidism, epispadias, hypoplastic scrotum.
Upper limb	<ul style="list-style-type: none"> - Macrodactylia - Q74.04
Syndromes	<ul style="list-style-type: none"> - Overgrowth syndromes such as Beckwith-Wiedemann-(Q87.30), hemihyperplasia or Sotos - Cornelia de Lange syndrome-(Q87.12), - Treacher Collins syndrome - (Q75.4 or Q87.0A), - Klippel Feil syndrome - (Q76.1) - Goldenhar syndrome (oculoauriculovertebral spectrum) (Q87.04) - Q87.26 VACTERL syndrome - Velocardiofacial syndrome - Robin Syndrome, - Alagille Syndrome, - Turner Syndrome, - Treacher-Collins Syndrome
Other Minor anomalies	<ul style="list-style-type: none"> - preauricular appendages, - macroglossia, - congenital bowing of the long bones
Other	<ul style="list-style-type: none"> - Congenital heart defects - OEIS Complex - Congenital defects in skin: hemangiomas, skin appendages, congenital nevi, hypertrichosis.



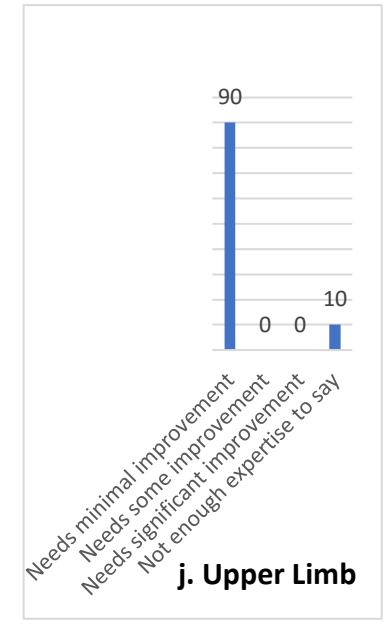
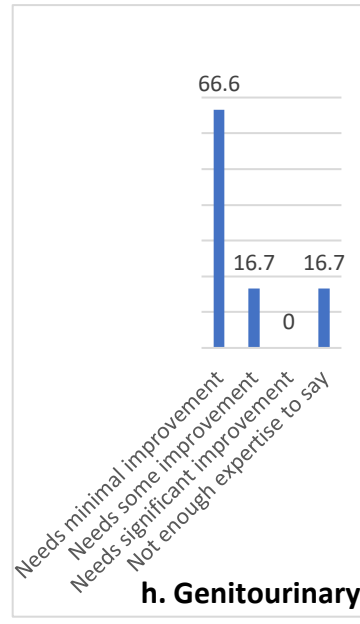
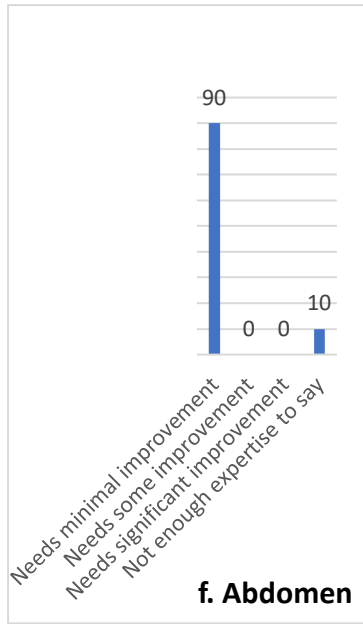


Figure 7 a - j Rating of clinical contents for different body parts

Section on Microcephaly

Most (80%) reviewers found the z-score tables and head circumference measurement videos for microcephaly were useful tools (Fig 8 & 9); 60% believed the app has potential to improve recording of microcephaly (Fig 10); and 63.6% felt minimal improvement was needed for the microcephaly section of the app (Fig 11). The following were suggested to improve the microcephaly section:

- Make clearer that the WHO z-scores table are for term births and Intergrowth for preterm
- Add Spanish version of head circumference video,
- Cut section of WHO head circumference measurement video that says “round up measurements to nearest 0.1 cm.
- Intergrowth video has irrelevant information (e.g at the beginning) and needs editing
- Add images of cases of newborns with malformed heads

Section on Congenital Zika Syndrome (CZS)

Most (75%) reviewers said the app has the potential to improve recording of CSZ and that minimal improvement was needed for the CZS section (Figure 12 & 13). The following suggestions/observations were made to improve the CZS section:

- Make the CZS section more visible; e.g. include link to CZS in main general syndrome description section.
- Add links through to the relevant syndromes that can be associated with microcephaly; e.g CZVS, CRS, cCMV, FASD etc
- Add a page on algorithm for CZS case ascertainment
- Improvement of recording of CZS by the app would depend on access to head circumference tape measures, support for providers and training.

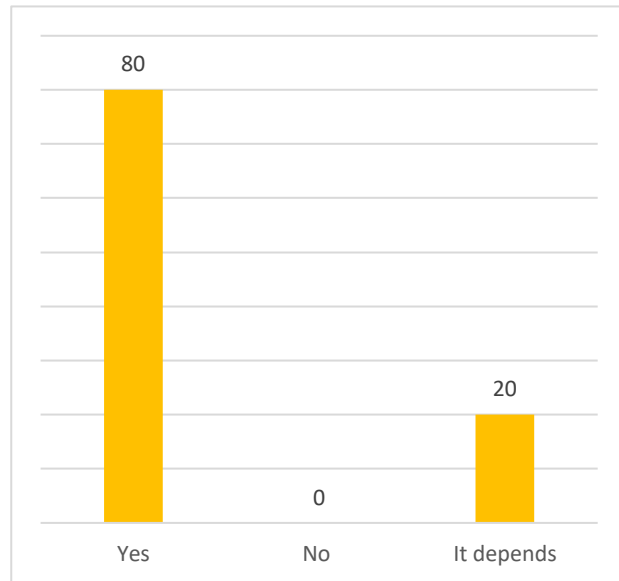


Fig 8 Usefulness of head size z-score table

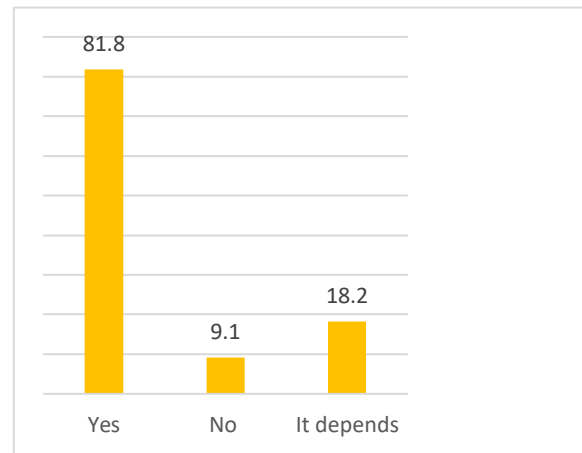


Fig 9 Usefulness of head circumference videos

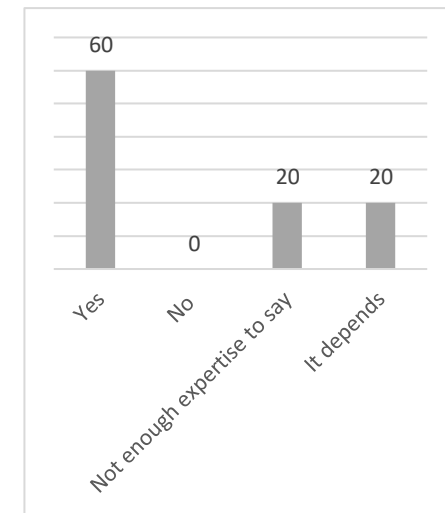


Fig 10 Potential of the App to improve recording of Microcephaly

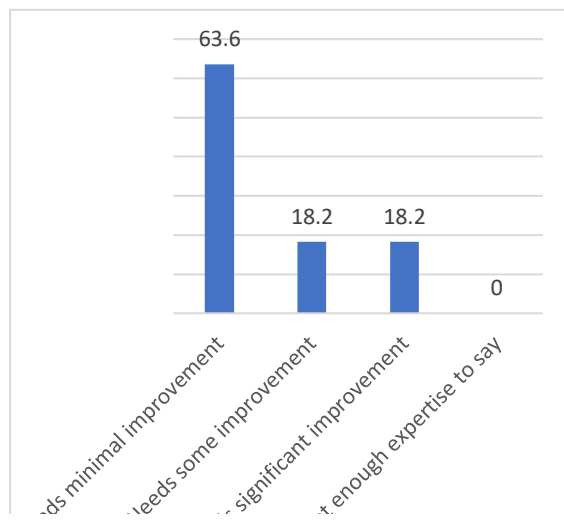


Fig 11 Overall assessment of Microcephaly section

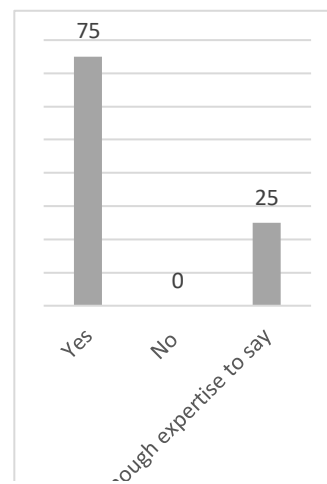


Fig 12 Potential of the App to improve recording of Congenital Zika Syndrome

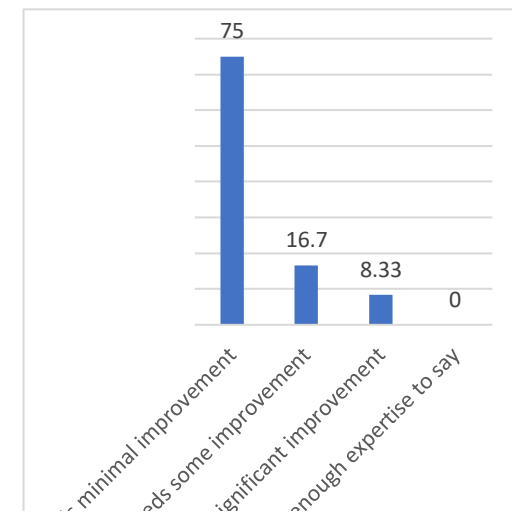


Fig 13 Overall assessment of Congenital Zika syndrome section

Section on Syndromes

For most (83.3%) reviewers, it was easy to move between the syndrome and birth defects tabs (Fig 14). Most (70%, Fig 15) of them wanted more syndromes added (see Table 1 for suggested additions). A few others (23%) wanted some syndromes removed from the app (Fig 16). They argue conjoint twins and Poland syndrome do not belong to the syndrome group; acephalous acardia is rare and should be removed, and that FAS is difficult to diagnose, especially in low resource settings and should be removed. One reviewer suggested that a separate page be created for FAS and another that a description about the characteristic face features of Trisomy 21 be added.

Section on minor anomalies

Generally, most (91.7%) reviewers agreed to the rationale to include some minor anomalies to guide their exclusion from birth defects surveillance data (Fig 17). However, one reviewer suggested that to make the app compatible with the WHO Atlas, minor anomalies should not be included.

Section on neonatal examination video

Neonatal examination video was generally considered very useful (Fig 18). One reviewer suggested the videos should be brought to one location and links to the videos should be provided in sections where they are mentioned.

Potential uses of the basic version of the App

All reviewers agreed the app would be useful in the training of health professionals (Fig 20). Most reviewers said they would use the App for training of students and junior birth defects research staff. Some also said they would use the App in clinical settings as a quick reference guide for the assessment of newborn and for the accurate description and coding of birth defects.

The majority (88.8%) of reviewers were convinced the app can improve on data collection for birth defects diagnosis (Fig 19) and 78% believed it will be useful tool to help tract future zika epidemics (Fig 21). Overall, 63.6% of the reviewers believed the app needed minimal improvement, while 36.4% believed some improvement was still needed (Fig 22).

Fig 23 shows the percentage of devices used by reviewers to explore the app, 50% of which were iPhones.

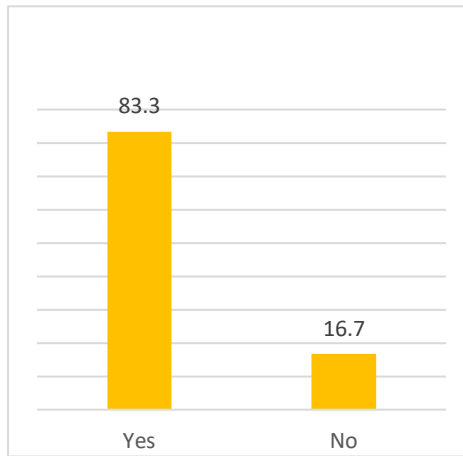


Fig 14 Ease of moving between syndromes and birth defects section

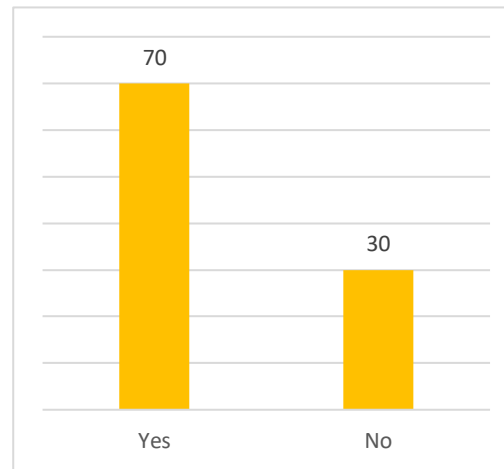


Fig 15 Are there extra syndromes which

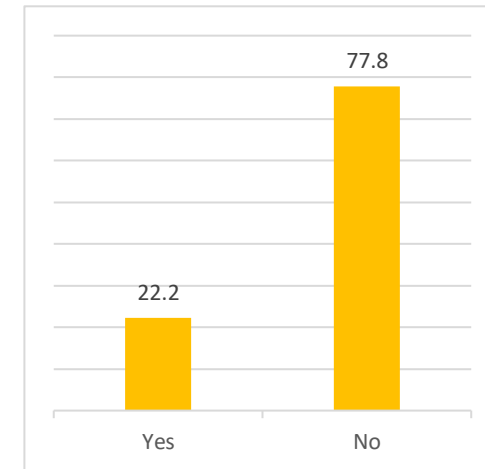


Fig 16 Are there syndromes which should be excluded?

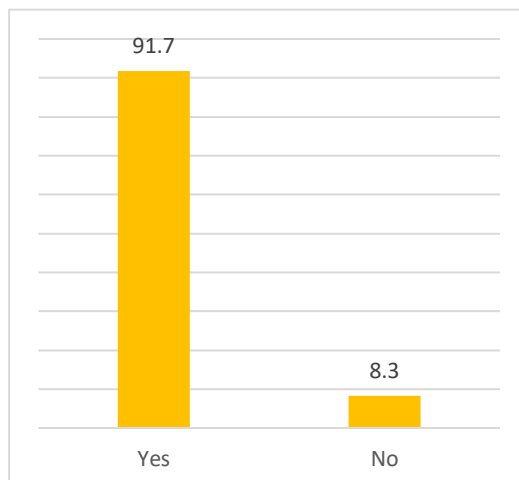


Fig 17 Agreement with inclusion of selected minor anomalies

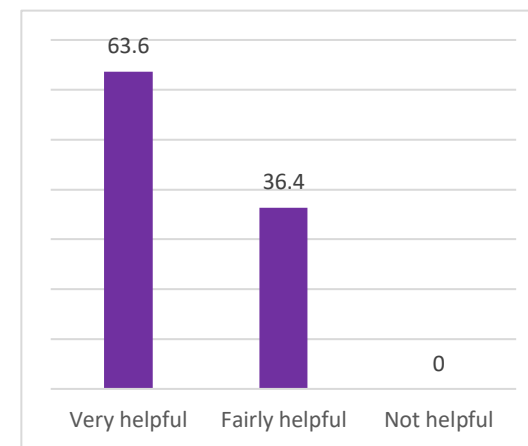


Fig 18 Is the neonatal examination video helpful?

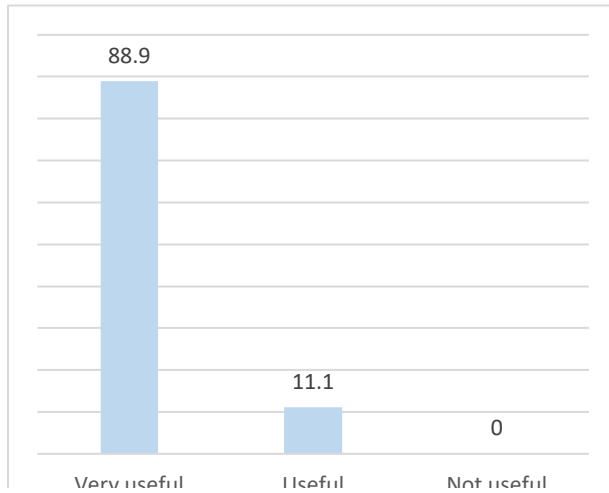


Fig 19 Usefulness of the App to improve data collection on birth defect diagnoses

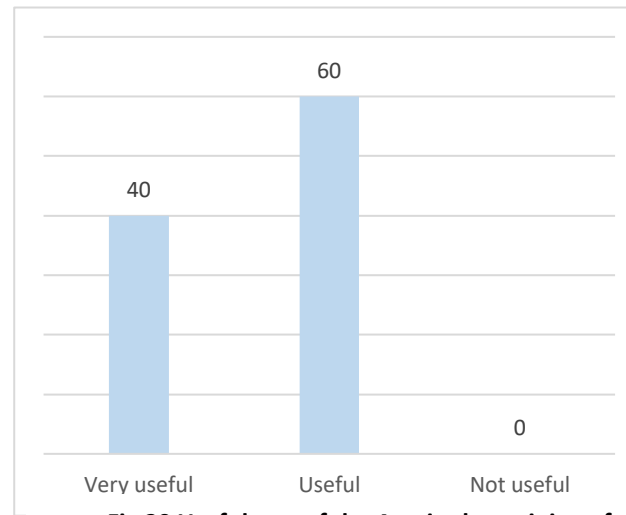


Fig 20 Usefulness of the App in the training of health professionals about birth defects

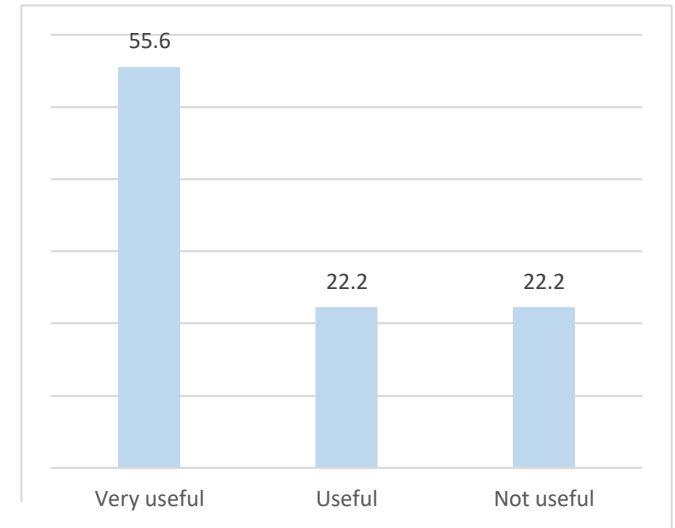


Fig 21 Usefulness of the App in tracking future Zika epidemics

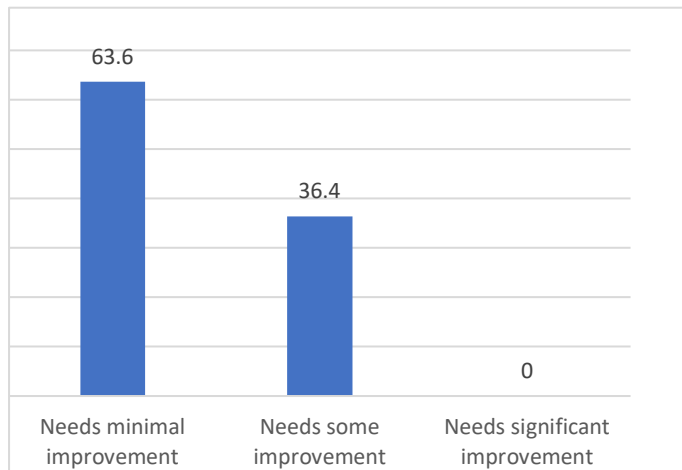


Fig 22 Overall functioning and contents of the App

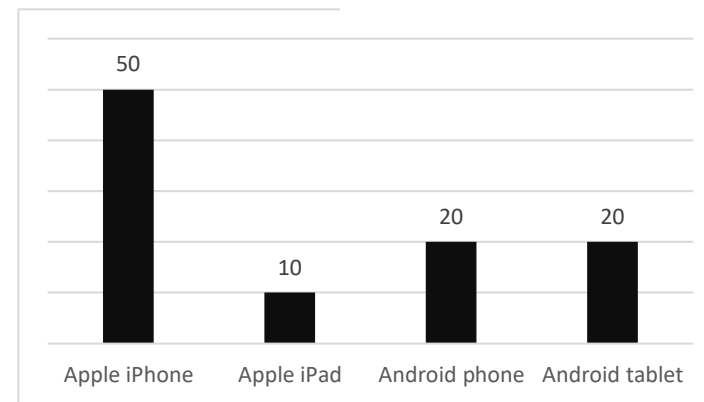


Fig 23 Device used to download the App

Contact

Chair of the International Committee for Congenital Anomaly Surveillance Tools: Professor Helen Dolk h.dolk@ulster.ac.uk; Research Associate Dr Leke Aminkeng az.leke@ulster.ac.uk