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(RESEARCH ARTICLE)

Commonly reported congenital anomalies in four regions of Ethiopia

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Abstract

Introduction: Congenital anomalies are structural, behavioral, and metabolic disorders that occur during intrauterine life, and can be identified prenatally, or later in infancy. The prevalence varies between studies as 3- 11 % of live births, 20-30% of stillbirths, and 7% of neonatal mortality. Many congenital anomalies are due to multifactorial inheritance, a combination of genes and environment.

Methodology: Cross sectional study was conducted October to December 2019 in health facilities supported by Transform: Primary Health Care project, five intervention regions (Amhara, Oromia, SNNP with Sidama and Tigray). Districts and health centers (HC) were randomly selected, with all hospitals included, minimum sample size was determined using standard approach.

Results: A total of 541 health facilities (110 primary hospitals, and 441selected HCs) were included in the study, 314 identified different congenital anomalies, 89 (28%) detected neural tube defect, 60 (19%) orofacial cleft, 40 limb defect (13%), 27 down syndrome (9%), 10 heart defect (3%) and 88 unclassifiedanomalies (28%).

Discussion: The highest proportion of congenital anomalies is CNS/ neural tube defects, followed by orofacial and limb/musculoskeletal anomalies. This indicates the need to improve folic acid supplementation during preconception and pregnancy period. Orofacial cleft, and musculoskeletal/limb anomalies, need timely corrective surgery.

Keywords: Congenital anomaly; Live births; Stillbirths; Neural tube defect

1 Introduction

Congenital anomalies are structural, behavioral, and metabolic disorders that occur during intrauterine life, and can be identified prenatally, or later in infancy. Abnormal form, shape or position of a body part, caused by mechanical intrauterine forces or extrinsic, due to oligohydramnios or breech position or intrinsic causes of deformation, such as fetal hypo mobility. It is one of the causes of post neonatal mortality, the prevalence varies between studies as 3- 11 % of live births, 20-30% of stillbirths, and 7% of neonatal mortality. Many congenital anomalies are due to multifactorial inheritance, a combination of genes and environment. More than 90% of congenital anomalies occur in low- and middle-income countries (LMIC) (1, 2, 3).

Every year an estimated 7.9 million children are born with major congenital anomalies, about 303,000 of them die within four weeks of birth every year, worldwide, and 3.2 million birth related disabilities, which may have significant impacts on individuals, families, health systems and society. The most common anomalies are heart defects, neural tube defects, musculoskeletal and Down's syndrome. Congenital anomalies are results of genetic, infectious, nutritional, or

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environmental factors. They can be prevented by vaccination, adequate intake of folic acid, iodine, and adequate antenatal care (3, 4, 5).

The etiology of birth defects are genetic (10–30%), environmental (5– 10%), multifactorial inheritance (20–35%), while 30–45% are unknown. Infectious agents appear to be the most important environmental factor in LMICs. Maternal factors include age, lifestyle, illnesses during pregnancy, antenatal care, use of alcohol, iodine deficiency, obesity, medication, exposure to chemicals, and non-use of peri-conceptual folic acid. Parental consanguinity, previous miscarriages and stillbirths, and inheritable congenital disease are other important factors. Mortality is very high among major congenital anomalies in LMICs rising to 20–85% (as against less than 10% in high-income countries). Significant number of survivors also suffer life-long disabilities, with birth defects accounting for 25.3 to 35.8 million disability-adjusted life years, worldwide (3,6,7, 8, 9).The purpose of this study was to assess commonly reported congenital anomalies and suggest preventive and curative measures.

2 Material and method

A cross sectional study was conducted linking with the project annual random follow up visits to health facilities of intervention areas. Randomly selected districts of the four target regions of Transform: Primary Health project (TPHC), (Amhara, Oromia, SNNP, Sidama and Tigray). The minimum sample size was determined using standard approaches, following the pattern: all Primary Hospitals (PHLs), District Health Offices (DHOs): Health Center (HC) 1: 2 .

2.1 Sample Selection Procedures

In the study all PHLs were included, DHOs, HCs were selected by the regional Monitoring & Evaluation (M&E) Program Officers in consultation with the country office M&E team using simple random sampling technique. List of hospitals, DHOs, HCs were first prepared by each Cluster Offices (CLOs) and submitted to the regional program office to be used as a sampling frame forselection.

2.2 Data collection methods and processes

Data were collected using checklists for each of the two levels which are hospitals, and HCs. Each data collector observed and verified whether all the necessary standards were in place. They were TPHC, CLO and Regional Program Office (RPO) staff. They attended training on how to administer the checklist List.Questionnaires on congenital anomalies were prepared based on the WHO recommendation, limited to readily identifiable, and easily recognized by physical examination at birth or shortly after birth. Congenital anomaly identification and documentation is part of the national BEmONC (Basic Emergency maternal Obstetric, and Newborn Care) training for midwives, in addition to their pre-service curriculum.

2.2.1 Data quality control

Sample checklists (10%at each level) were double-entered to verify the quality of data entry, and the matching report showed excellent agreement (99.0%).

2.3 Data processing and analysis

All completed checklists from delivery registers were submitted to the TPHC Country office for data entry and processing. A team of data entry clerks performed data entry, by editing, coding of open-ended questions, and random verification of entered data. Data encoding, entry and processing were managed by the Country Office (CO) M&E team, and analyzed using SPSS version 16.0.

3 Results

A total of 541 health facilities (110 primary hospitals, and 441 randomly selected HCs) were included in the study, 314 (58%) identified congenital anomalies, 89 (28%) detected neural tube defect, 60 (19%) orofacial cleft, 40 limb defect (13%), 27 down syndrome (9%), 10 heart defect (3) and 88 (28) unclassified (others) anomalies (figure 1).

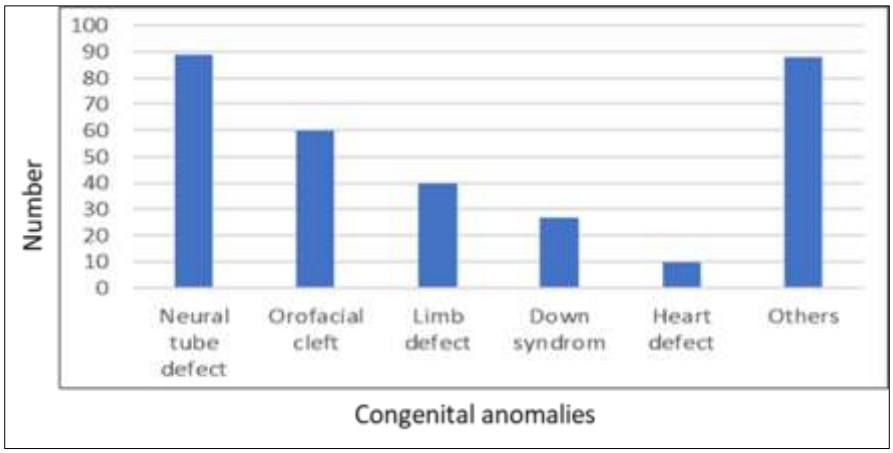

Figure 1 Number of facilities detected congenital anomalies

36% of hospitals, and 11% HCS detected neural tube defect, 26% of hospitals and 7% of HCs identified orofacial cleft, 12% of hospitals and 6% of HCs detected limb defect, 16% of hospitals and 2% of HCs have seen downs syndrome, 5% of HCs and 1% of HCs have seen heart defect. Unclassified anomalies were seen in 35% of hospitals and 11% of HCs (figure 2).

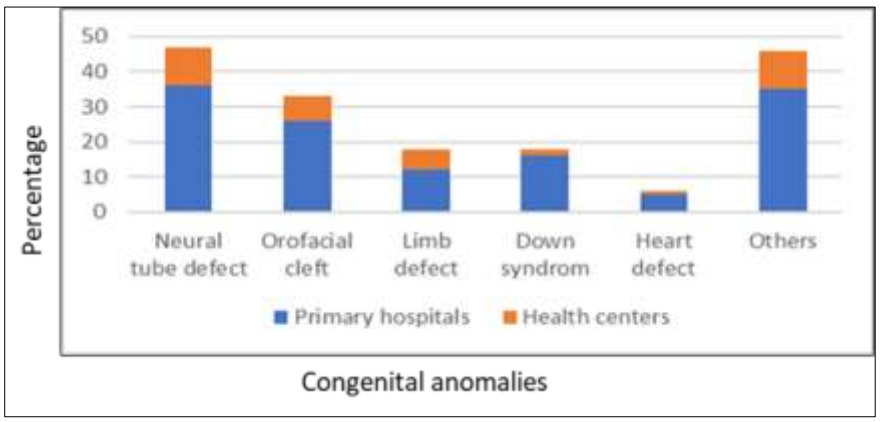

Figure 2 Proportion of health facilities detected congenital anomalies

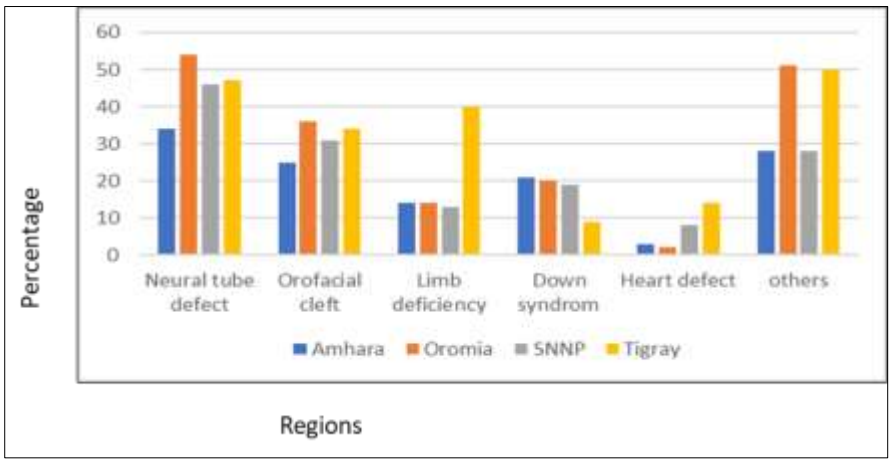

Figure 3 Regional variation congenital anomalies

Regional variations higher proportion of neural tube defect were seen, in Oromia 54% health facilities, low in Amhara 34%, Limb defect higher in Tigray 40%, lowest in SNNP 13%, orofacial cleft highest in Oromia and Tigray, 36 and 34 respectively, lowest in Amhara 25% (Figure 3).

4 Discussion

The highest proportion of health facilities detected neural tube defect which was 28% followed by orofacial cleft 19%, 13% limb defect, 9% downs syndrome, and 3% heart defect. Unclassified were 28%. Higher numbers of congenital anomalies were reported from hospitals, than HCs which can be explained by the higher number of deliveries happening in hospitals and better diagnosing capacity of the HWs (doctors, specialists), and availability of imaging instruments like ultrasound in hospitals. Regional variations were seen, neural tube defects and orofacial clefts predominate in Oromia and Tigray, and limb defects are highest in Tigray.

Different studies done in Ethiopia showed similar results, the prevalence of congenital anomalies was 1.4-2%, the highest proportion 31-40%were CNS/ neural tube defects, 11-34% orofacial and 20% -23% limb/musculoskeletal system (10, 11,12). The result of this study is similar to our finding as the highest anomaly being CNS/ neural tube defect, the second orofacial defect and the third musculoskeletal or limb anomalies. This shows that the problems are similar in the country.

The prevalence of congenital anomalies in different studies in Nigeria was 2.1 -6.3%, the pattern varied from one region to the other, anomalies of gastrointestinal system 12%-33%, Orthopedic 34%, CNS 12-27%, cardiovascular 12%, and unclassified up to 33% (13). In our study neural tube defects and orofacial clefts predominate. Detecting internal structural anomalies, like congenital heart disease, gastrointestinal malformations, and kidney require imaging techniques, or other specialized procedures, were not available at primary health care level. A study done in South African blacks also showed incidence of 3%, of which musculoskeletal 61%, CNS including neural tube defects 14%, and chromosomal 9%. Major congenital anomalies are significantly reduced in women' who take periconceptual multivitamin supplements, but women who are malnourished are at greater risk. This shows that the underlying cause could be poor socio-economic status of the population and the deficient health care facilities (14). In a study done in Iran the prevalence ranged from 1.13% to 3.9%, disparities might be attributed to highly polluted industrial zones. Anomalies of the nervous system were the most common defects (24%) , followed by heart disease anomalies(18%) (15).

In a study done in Bangladesh, unlike our study, congenital heart disease was commonest 59%, club foot was 9%, down syndrome was 8%, and total death with congenital anomalies was 6% (16). In Japan multiple congenital anomalies were 7% of total congenital anomalies, and mortality was found to be higher than isolated ones (9.8% vs 3.7%) (17). In Lebanon 2.4% were congenitally malformed, cardiovascular defects and limb anomalies were mostly detected, followed by respiratory system and chromosomal anomalies. Association was found with parental consanguinity and alcohol consumption of the mother were found (18).

The prevalence of congenital abnormalities in Europe was 2.5%, some congenital anomalies that were continuing to increase especially CHD (Congenital Heart Disease). Risks factors for these anomalies include diabetes and obesity, both of which are known to be increasing, smoking and alcohol consumption were additional factors. Public health prevention policies should therefore continue to focus on these two conditions. Decrease in NTDs did not occur for the last ten years, preventive measures need to be strengthened (19,20).

The reasons for reduction of infant mortality due to congenital anomalies in western countries has been the establishment of epidemiological surveillance networks of congenital diseases, improvement of diagnosis (including prenatal and early postnatal diagnosis), appropriate medical and surgical management of newborns. In developing countries, especially in sub-Saharan Africa such registries are non-existent. The reported incidence and mortality rates associated remained high, with very little information on other epidemiological data (21).

Limitations

Health workers/ midwives attending delivery in hospitals and HCs were not given special training on detection of anomalies, and data was collected from delivery registers. There can be mistakes, especially in identifying any of the internal congenital anomalies like heart, kidney, and intestinal defects. The other limitation of this study was that the data was collected by project staffs.

5 Conclusion

Despite all the limitations of properly identifying the congenital anomalies in lower health facilities, Ethiopia has a high proportion of congenital anomalies like other African countries and some Asians. The proportion of neural tube defects is highest indicating the need to improve folic acid supplementation during preconception and pregnancy period. Orofacial cleft, and musculoskeletal/limb anomalies, need timely corrective surgery.This study recommends the need to conduct more research using imaging techniques or specialized procedures to identify internal structural anomalies.

Recommendations

- The highest congenital anomaly was neural tube, indicating the need to improve folic acid supplementation preconception and pregnancy period.
- Orofacial cleft, and musculoskeletal anomalies require corrective surgical intervention.
- Internal structural anomalies require imaging techniques, or specialized procedures, equipment and training at primary health care level will help to make correct diagnosis.
- Establishment of surveillance networks will help to decrease congenital anomalies

Compliance with ethical standards

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Disclosure of conflict of interest

All authors declare that they have no conflict of interest.

Statement of ethical approval

There was agreement between Transform Primary Health Project and districts to collect data while conducting random follow up visit.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

Author's contribution

All authors contributed to the research from the proposal writing to draft preparation and finalization.

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Data was collected integrated with random follow up visit in the project program.

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