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A Review on Situation of Congenital Disorders and Access to Community Genetics Services in Bangladesh

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Abstract

About 2-3% of all live births suffer from congenital abnormality globally and 70% of those are preventable through community genetics services. There is no population-wide data on congenital disorders in Bangladesh. The objectives of this article are: to identify the prevalence of the common congenital disorders, to assess the factors influencing congenital disorders and to assess the situation of access to the community genetics services in Bangladesh. A review has been conducted with formal and informal searches. The estimated prevalence of congenital abnormalities is about 2-4% in live births along with still born and aborted fetus. Neural tube defects are the commonest congenital abnormality. Other common defects involve gastrointestinal, cardiovascular, genitourinary and musculoskeletal system. Down's syndrome shares majority, nearly 71%, of all chromosomal abnormality. The ethnic variations in the prevalence of Beta thalassemia trait and HbE trait are respectively 4.1% and 6.1% in Bengali students of nine and ten grade and that in the tribal students of same grade 4.2% and 41.7%. The electrophoretic patterns and distributions of hereditary hemoglobin disorders in cases are: thalassemia trait-47.14%, HbE-beta thalassemia-30.47%, HbE trait-13.3%, HbE disease-5.71% and thalassemia major-3.33%. The prevalence of haemophilia-A is 0.4/100,000 males that of congenital hypothyroidism is 1/(1300-1353) newborns. Key factors associated with the burden of congenital disorders in Bangladesh are: high prevalence of carriers of genetic diseases along with consanguineous marriage, inadequate antenatal check-up, high rate (nearly 72%) of unskilled home delivery and lack of community genetics services. Political and professional prioritization of community genetics services through the existing primary health care resources is a priority social, health and economic interest for effectively addressing the problem of congenital disorders in Bangladesh.

Keywords: Community genetics, Congenital disorders, Bangladesh

Introduction

Mapping and deciphering of the DNA sequence of all the structural and functional genes of human genome are the revolutionary advancement of the technology. Molecular basis of all human diseases is a growing interest of medicine today [1]. The emerging clinical genetics reveals that all diseases have either major or minor genetic as well as environmental components with varying degrees of influence and interactions among them [2]. An apparently complete concept of human genetics and genetic disorders needs an account of anthropological, ethnological, sociological, cultural and religious dimensions of human history and civilization along with scientific knowledge of genetics [1].

Genetic diseases along with congenital malformations, fetal disease, intrauterine growth retardation and disability are the categories of the congenital disorders. An estimated 5% of all pregnancies end with childbirths having significant congenital and/or genetic disorders [1]. In 2004, an estimated approximately 260,000 neonatal deaths were due to congenital disorders which occupied nearly 7% of all neonatal mortality globally [3]. These findings clearly indicate that congenital disorders are one of the public health concerns and genetic diseases (e.g. single gene, chromosomal and multifactorial abnormalities) are the commonest of those.

Congenital disorders (also called birth defects) are associated with damaging health, economic, psychological and social consequences to the individual, family and health sector. The scientific basis of many congenital abnormalities and their prevention, especially of the genetic diseases, have distinct individual, social, religious, cultural and legal dimensions; for example, consanguineous marriage, selection of partner, family size, access to and preference of family planning, abortion law, age of marriage and parenthood. Other than genetic factors, there are some common environmental factors related to birth defects, such as maternal infections (rubella, TORCH, syphilis), nutritional deficiency (e.g. folic acid) and antenatal exposure to harmful drugs and radiation. Further, some disorders such as maternal diabetes, hypertension have both environmental and genetic components and are associated with congenital malformations. Over 70% of all congenital disorders are preventable [4].

Realizing the burden of congenital disorders the WHO expert team has endorsed the Community Genetics Services (CGS). The goals of the endorsement are to reduce the burden of congenital disorders dealing with the underlying complex issues relating to those through evidence-based intervention while respecting the autonomy of the risk population group in the developing countries. Prevention of congenital disorders, as suggested by the WHO expert panel, includes: primary prevention (e.g. premarital and pre-conception genetic screening and counselling, pre-implantation screening, carrier detection and prenatal screening of fetus of risky couples), secondary prevention (e.g. newborn screening, testing and follow-up care) and tertiary prevention (e.g. surgical treatment and rehabilitation). The key pre-requisites of the CGS are: prioritizing the issue politically and professionally, availability of skilled human resources, financial and technical supports, cost-effective implementation of the evidence-based interventions through integrating the service with the existing primary health care (PHC) system and community mobilization. However, in many developing countries CGS are either not prioritized or in an infantile stage because of the competing interests, lack of resources, and legal, religious and cultural limitations [4].

Bangladesh is one of the developing countries of South-East Asia. The country is an intimate part of civilization of Indian subcontinent because of the long history of shared geography, politics, anthropology and culture. Being a part of the historical undivided Indian subcontinent, the land was invaded and ruled-over by various rulers and empires from Middle-East and Europe. Social interaction with invaders and subsequent genetic diversification among the indigenous people became rampant. Unexpected consequences of exchange of mutant genes cannot be ignored across generations. Congenital disorders are common in the countries of this historically and culturally linked region including Bangladesh [1].

Bangladesh ranks the world's densest populated country with over 151 million people. Muslim as the majority constitutes nearly 90% of the total population. Annual population growth rate is 1.36%. Prevalence of contraceptive use and home delivery by traditional birth attendants is about 61% and 72% respectively. Infant mortality rate is 33/1000 live births [5]. Being a low income country, the public allocation to health sector is low only 3.6% of the gross domestic product. Private health expenditure (PvtHE) constitute about two-thirds (65.6%) of the total health expenditure. Of the PvtHE about 97% is households' out-of-pocket payments [6]. The healthcare delivery system of the country is organized into primary, secondary and tertiary health care levels. PHC workers occupy nearly two-thirds (61,312) of a total of 93,312 health workforces under the Directorate-General of Health Services [5]. Shortage of human resources is a chronic problem of the healthcare system.

In this context taking into account the public health importance of congenital disorders, this document aims to: (i) identify the prevalence of the common congenital disorders in Bangladesh, (ii) assess the factors contribute to the prevalence of congenital disorders and (iii) assess the situation of access to the community genetics services in Bangladesh.

Methods

The study is mainly based on available literature on congenital disorders in Bangladesh. Very few articles are published in peer-reviewed journal on congenital disorders in Bangladesh. To find all possible sources of information on the issue, we conducted both formal and informal searches on WHO website, PubMed, google and google scholar.

Findings

Prevalence of the common congenital disorders in Bangladesh

No national population-wide data on the congenital disorders in Bangladesh is available. Most of the available data are tertiary level healthcare facility-based. A specialized facility-based study during January to December 2007 on 1630 deliveries including live births, stillborn and abortions found a total of 60 (i.e. 3.68%) congenital abnormalities. Prevalence of congenital abnormalities in male was 2.15 times higher than in female newborns. Congenital abnormalities were found higher in primi para (63.33%) and age group 25-29 years (53.33%). The commonest congenital abnormalities detected were: neural tube defect (NTD) 46.67%; in them, 33.33% was identified having hydrocephalus, the most common complication. NTD was followed by defects of the urinary system (23.33%), gastrointestinal system (6.68%) and musculoskeletal system (5%). No definite genetic disorders were investigated. It was reported that about 92% of those pregnant women had had irregular antenatal care [7]. Another tertiary level hospital-based study on 6040 admitted neonates during April 2011 to March 2012 reported the prevalence of congenital abnormalities of 1.75%. The combined alimentary tract, head and neck, and trunk anomalies were the highest (20.75%) that was followed by the anomalies of the central nervous system (19.81%), cardiovascular system (18.87%), musculoskeletal system (12.26%), genitourinary system (11.32%), chromosomal abnormalities (6.6%) and others 10.37%. Down's syndrome was the commonest (71%) among known chromosomal abnormalities. Congenital abnormalities were nearly 20% higher in male than in female neonates. This study reported maternal exposure to the following known risk factors: antiepileptic drugs, vitamin-A, maternal diabetes, history of smoking, previous delivery of malformed baby, young mother and consanguineous marriage [8].

A cross sectional study among 735 students (ethnicity: Bengali 687 and tribal 48) of grade nine and ten from all administrative divisions of Bangladesh was conducted to identify Beta thalassemia trait and HbE trait by haemoglobin electrophoresis technique.

This study reported that in Bengali students the prevalence of Beta thalassemia trait and HbE trait was 4.1% and 6.1% respectively and that in the tribal students were 4.2% and

41.7%. Two Bengali and one tribal student were positive for HbS/D bands [9]. Another tertiary level facility-based study among 210 cases (male:female=51:49) of age 2-72 years during January to December 2007 was conducted to identify the electrophoretic pattern and distribution of hereditary haemoglobin disorders and reported as follows: thalassemia trait-47.14%, HbE-beta thalassemia-30.47%, HbE trait-13.3%, HbE disease-5.71% and thalassemia major-3.33% [10]. A conservative estimation of WHO as cited in Amin [11] reported that about 3% and 4% populations of Bangladesh are carriers of Beta thalassemia and HbE thalassemia and yearly more than 2000 children born with thalassemia [11]. The reported prevalence of haemophilia A, an X-linked bleeding disorder, was 0.4/100,000 males in 2006 [12].

Congenital hypothyroidism is an important cause of irreversible mental and physical disability if remained undetected then untreated. A pilot programme screened 2,600 newborns, mostly delivered at the hospitals of Dhaka city and detected 2 cases of congenital hypothyroidism with an incidence of 1/1,300 newborns. Another study in the southern Bangladesh also detected nearly a similar incidence of congenital hypothyroidism (1/1,353 newborns) [13]. A case-control study in Dhaka city reported a significantly higher prevalence of rubella seropositive in the children with hearing impairment of seropositive mothers than that in control children and mothers [14].

Since there is no population-wide data, the above facility-based study results could be a tool to assess the burden of birth defects in Bangladesh in comparing with the global distribution of common birth defects **(Table 1)**.

Factors contribute to the prevalence of congenital disorders

There found no available population-based study to identify the underlying factors of congenital disorders in Bangladesh.

However, the available information identify following factors as the potent contributors of congenital disorders: running of the mutant genes across generations, high prevalence of consanguineous marriage, religious and social limitations of abortion, lack of facilities for carrier screening, premarital screening, genetic counselling, general ignorance about genetic literacy, lack of prenatal screening facility.

Community genetics services are not politically and professionally prioritized because of the other competing health burdens. Inadequate access to and/or utilization of antenatal check-up with consequent low detection of high risk pregnancies and high prevalence of unskilled home deliveries often undermine primary prevention (for example: neural tube defect) and timely detection (such as Down's syndrome) of congenital disorders. The association between congenital disorders and high prevalence of early marriage with young motherhood is strongly predictive. A significant proportion of congenital disorders remain undiagnosed and unreported in Bangladesh because of lack of facility, skilled manpower and population awareness.

Table 1 Estimated global prevalence of some common birth defects

Disorders	Estimated prevalence	Sources
Down syndrome		[15]
LMICs	2-3/1000 live births	
HICs	1.2/1000 live births	
Congenital heart defects	4-8/1000 live births	
Neural tube defects	300,000 new-borns (estimated annual global total)	
Congenital hypothyroidism	1/4000 live births	[16]
Haemoglobin disorders		[17]
Significant variant[a]	5.2% of the population	
α^+ thalassaemia[b]	20.7% of the population	
Any variant[c]	24.0% of the population	

Notes: [a]Significant variants includes HbS, HbC, HbE, HbD etc. β thalassaemia, α^0 thalassaemia; [b]α^+ thalassaemia includes both hetero and homozygous α^+ thalassaemia and [c]includes (i) coincidence of α and β variants, and (ii) harmless combination of β variants.

Situation of access to the community genetic services in Bangladesh

Bangladesh is facing the rising burden of non-communicable diseases related deaths and disability; whereas, the issues of malnutrition and communicable diseases (for example, diarrhoea, tuberculosis) are still unsettled [17]. The CGS has yet to be prioritized by policymakers and professionals. Clinical genetics is still a subject of least interest and/or importance in the undergraduate medical curriculum. Further, general populations are not aware of genetic as well as environmental risk of developing congenital disorders. Trained human resources are one of the pre-requisite of CGS, which is in a crucial need in the healthcare sector of the country. Moreover, overall facilities for genetic counselling along with carrier, prenatal and newborn screening for genetic and/or congenital abnormalities are in serious dearth. Most of the screening and diagnostic facilities; for example, Hb-electrophoresis to screen thalassemia, screening for congenital hypothyroidism, diagnosis of haemophilia are established in the capital Dhaka city including some megacities. Most of those facilities are privately owned and out of affordability of the majority people in need. For this, the rural residents who constitute about 72% of the total population remain out of access to the community genetic services in Bangladesh.

Conclusion

Prevalence of congenital disorders is alarming in Bangladesh. Among congenital disorders, Beta thalassemia,

HbE, neural tube defect, congenital hypothyroidism, haemophilia, Down's syndrome and congenital rubella syndrome are common in Bangladesh. Both genetic and environmental factors are associated with congenital disorders. However, the high prevalence of Beta thalassemia and HbE trait with culture of consanguineous marriage and lack of established community genetic services, the prevalence of thalassemia major is a potential social, health and economic threat for the country. In Bangladesh, most of the available community genetic services are based on secondary and tertiary prevention and often out of affordability of the general population.

In Bangladeshi context, where approximately 72% of all deliveries are conducted by traditional birth attendant at home and lack of proper diagnostic facilities majority of the foetal or early childhood deaths relating to birth defects remain unknown. However, proper primary health care along with secondary or tertiary level referral care could correct over 40% birth defects [18]. To overcome the barriers of other competing interests in health sector, prevention of birth defects and care of the affected children have been recommended to be ensured cost-effectively with existing resources [15]. Thus, community, primary care staff, promotion of lay-support organizations, training of nurses and doctors should be institutionalized. Proper neonatal examination should be ensured. Premarital and pre-conceptual counseling, family planning service and antenatal screening of foetus needs to be developed. For proper addressing the issue national birth defects surveillance system has also been recommended by experts [15].

Bangladesh is in huge need of community genetics services. To overcome the challenge of congenital disorders, the issue has to be prioritized politically and professionally. For the cost-effective implementation of the components of the community genetic services, the existing resources of primary health care system has to be reorganized for primary prevention through proper training for the community health workers along with good referral and surveillance system. Universal coverage of congenital disorder-focused skilled maternity care has to be instituted. Community mobilization should be ensured involving key stakeholders namely, patients and families, health workers of all levels, faith-based organizations, civil society organizations, media, international experts and donors.

References

1. Kumar D (2004) Genetic Disorders of the Indian Subcontinent. (1stedn). The Netherlands: Kluwer Academic Publishers, 2004. The Ethical, Legal and Social Implications of genetic testing and screening [Online] [Internet]. 1st edn. Kumar D, editor. Netherlands: Kluwer Academic Publishers; 2004. Available from: http://books.google.com.bd/books?id=bpl0LXKj13QC&pg=PA81&lpg=PA81&dq=congenital+disorders+in+bangladesh&source=bl&ots=BOeu3Hii-k&sig=n8LlY_Dr63IiGba1TrDrOD0faUA&hl=bn&sa=X&ei=6yFsVIebO47KaOS1gugB&ved=0CEcQ6AEwBQ#v=onepage&q=congenital disorders in banglad
2. (2006) WHO. The Ethical, Legal and Social Implications of genetic testing and screening [Online]. 2006; Available from: Available at: http://www.who.int/genomics/publications/GTS-MedicalGeneticServices-oct06.pdf [Accessed 24/09/2015]
3. (2004) WHO. Global Burden of Diseases 2004 Updates [Online]. 2004; Available from: Available at: http://www.who.int/healthinfo/global_burden_disease/GBD_report_2004update_full.pdf?ua=1 [Accessed 24/09/2015]
4. (2010) WHO. Community genetics services [Online]. 2010; Available from: Available at: http://whqlibdoc.who.int/publications/2011/9789241501149_eng.pdf [Accessed 24/09/2015]
5. (2014) Ministry of Health and Family Welfare. Health Bulletin 2014. Available from: Available at: http://www.dghs.gov.bd/images/docs/Publicaations/HB_2014_2nd_Edition_060115.pdf [Accessed 21/11/2015] [Accessed]
6. (2014) WHO. Global health expenditure database: Bangladesh [Online]. Available from: Available from: http://apps.who.int/nha/database/Key_Indicators_by_Country/Index/en?COUNTRYK EY=84674 [Accessed 24/09/2015]
7. Fatema K, Begum F, Akter N, Zaman S (2012) Major Congenital Malformations Among The Newborns in BSMMU Hospital. Bangladesh Med J 40: 7-12.
8. Islam MN, Siddika M, Bhuiyan MKJ, Chowdhury AM (2013) Pattern of Congenital Anomalies in Newborns in a Tertiary Level Hospital in Bangladesh. J Surg Pakistan (International) 18.
9. Khan WA, Banu B, Amin SK, Selimuzzaman M, Rahman M, et al. (2005) Prevalence of Beta thalassemia trait and Hb E trait in Bangladeshi school children and health burden of thalassemia in our population. DSH J 21: 1–7.
10. Uddin MK, Aziz MA, Sarder MH, Hossain MZ, Bhyua MF, et al. (2007) Electrophoretic Pattern of Hereditary Haemoglobin Disorders in Bangladesh. J Dhaka Med Coll 19: 39-42.
11. Amin SK (2011) Prevention of Thalassaemia by Genetic Counseling. Anwer Khan Mod Med Coll J 2.
12. Stonebraker JS, Bolton-maggs PH, Soucie JM, Brooker M (2010) A Study of Variations in the Reported Hemophilia B Prevalence Around the World. Haemophilia 20–32.
13. Rasul CH, Lucky SN, Miah SR, Moslem F (2008) Congenital Hypothyroidism in the Southern Bangladesh. J Teach Assoc RMC, Rajshahi 21: 18-22.
14. Rahman MM, Khan AM, Hafiz MM, Ronny FMH, Ara S, et al. (2002) Congenital hearing impairment associated with rubella: Lessons from Bangladesh. Southeast Asian J Trop Med Public Health 33: 811-817.
15. Christianson A, Howson CP, Modell B (2006) Global Report on Birth Defects' The hidden toll of dying and disabled children [Internet]. march of Dimes Birth Defects Foundation.
16. Hasan M, Nahar N, Ahmed A, Moslem F (2003) Screening for congenital hypothyroidism--a new era in Bangladesh. Southeast Asian J Trop Med Public Health. 34: 162-164.
17. Modell B, Darlison M (2008) Global epidemiology of haemoglobin disorders and derived service indicators. Bull World Health Organ 86: 480-487.
18. Christianson A, Modell B (2004) Medical Genetics in Developing Countries. Annu Rev Genomics Hum Genet 5: 219-265.