Perspective

Addressing the public health challenge of birth defects in India

Manpreet Singh Khurmi[1], Medhavi Gupta[2], Gaurav Chaudhari[3]

From [1]Ministry of Health and Family Welfare, Government of India, [2]B J Medical College, [3]Department of Community Medicine, NHL Municipal Medical College, Ahmedabad, Gujarat, India

Correspondence to: Dr. Gaurav Chaudhari, C-0604, Samarpan Tower, Naranpura, Ahmedabad - 380 013, Gujarat, India. E-mail: drgaurav18@gmail.com

Received – 05 June 2014 Initial Review – 12 July 2014 Published Online – 28 December 2014

Abstract

Birth defects consisted of a group of diverse clinical conditions categorized on the basis of a congenital presentation and a partly or wholly genetic etiology. Although individually rare, birth defects affect 2-3% of all births in India. As India has the largest global annual births, in absolute numbers, India may harbor the largest number of affected children worldwide. There is a need of strategic research and interventions to bring down the rate of birth defects and associated economic burden and also a need to actively screen and identify them at early stage so necessary intervention (medical or surgical) could be initiated so as to reduce lifelong disability as a result of such defects. The recent child health screening and early intervention service initiative by the Government of India, the Rashtriya Bal Swasthya Karyakram under the National Health Mission is the first attempt in the direction toward providing services for some of the more prevalent birth defects and has the potential to alleviate suffering of affected children especially from rural areas. The data collected from this program could help policy makers to allocate sufficient funds aimed at treatment of birth defects and also develop behavior change communication strategies for prevention of the same.

Key words: *Birth defects, India, Intervention, Rashtriya bal swasthya karyakram, Screening*

Birth defects consist of a diverse group of rare disorders of pre- or post-conception origin. They arise either due to a genetic etiology (caused by chromosomal abnormalities and single gene disorders) or due to gene-environmental interactions, where the maternal physiological environment interacts with the genetic predisposition of the fetus to cause the defect [1]. Other birth defects arise because of intra-uterine developmental defects while the etiologies of many birth defects are unknown [2]. The information is summarized in Table 1 and Fig. 1.

Birth defects are traumatizing chronic conditions that result in physical or mental disability, or in lifelong medical conditions such as the hemoglobinopathies or bleeding disorders. Many birth defects are incompatible with life and therefore result in pregnancy losses and early neonatal mortality. The focus of maternal and child health intervention in India and other developing countries is on the reduction of child mortality due to preventable causes. Until now, birth defects had remained a low public health priority with no programs for the prevention and care for the affected [3].

The first move by the Government of India to address the issue of birth defects through the Rashtriya Bal Swasthya Karyakram (RBSK) is described in this article [4].

ESTIMATES OF THE MAGNITUDE OF BIRTH DEFECTS IN INDIA

Data on birth defects is obtained through surveillance that may be either hospital or population-based [5]. Genetic disorders are usually monitored through rare disease registries where all diagnosed patients are recorded. There is very little population-based research on birth defects in India.

A global assessment of the burden of birth defects was conducted by the March of Dimes, in 2006 [6]. India ranked 38th among all countries in terms of the prevalence rate of birth defects (India 64.3/1000; range 82/1000 in Sudan to 39.7/1000 in France). In terms of the absolute number of affected children, India had the highest estimated numbers of live or stillbirths due to birth defects (estimated 1,613,502 births). This number was one and a half times more than that of China, which had the second highest estimated global burden of children born with birth defects (963,997 affected children born annually). This data are not unexpected as India has the highest number of children born annually (25,112,000 when compared to 18,841,000 for China) and a higher birth rate (20.97 births/1000 population) when compared to China (12.29 births/1000 population) [7].

Table 1: Proportion of different types of birth defects in high income countries

Cause	Approximate percentage of affected births %
Pre-conception	
Multifactorial malformations such as neural tube defects	~20-30
Chromosomal anomalies	~6
Single gene disorders	~7.5
Post-conception	
Teratogens	7-8
Intrauterine abnormalities	~2
Cause unknown	~50

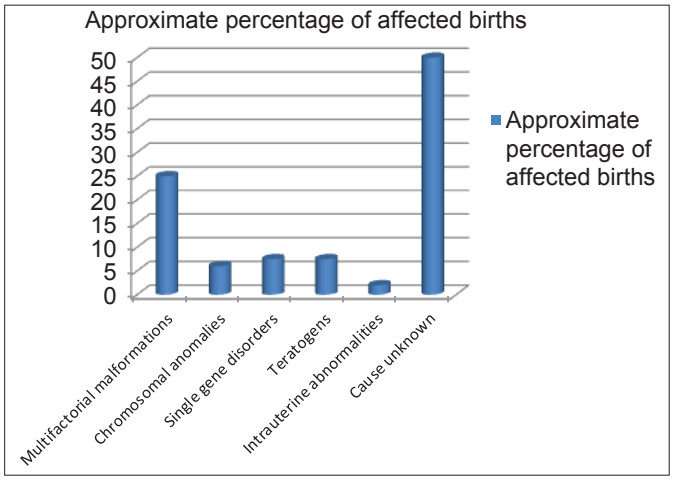

Figure 1: Proportion of different types of birth defects in high income countries

BIRTH DEFECTS AND NEONATAL MORTALITY

Another source of data on the magnitude of birth defects comes from the estimates of early neonatal mortality. The Million Child Death study conducted in 2005 reported that the neonatal mortality rate (NMR) due to congenital anomalies was 1.2/1000 births, accounting for 32,000 neonatal deaths in that year [8].

The Lancet Child Mortality data estimated that the NMR due to congenital anomalies was 3/1000 births, affecting 76,000 births in India in 2010 [9] Globally, countries of the South East Asia region of the World Health Organization harbor the largest burden of neonatal mortality due to congenital anomalies. India harbors 77% of the burden of neonatal mortality due to congenital anomalies, reiterating that globally, India has the largest magnitude of neonatal mortality due to congenital anomalies.

BIRTH DEFECTS PREVENTION THROUGH PRECONCEPTION CARE

Interventions delivered during the preconception period provide an opportunity for the prevention of birth defects as well as for improving pregnancy outcome. From the perspective of prevention, birth defects can be prevented using low-cost tools of preconception care, targeted at the modifiable risk factors of birth defects. The preventive strategies include correction of Folate deficiency (as well as Iron and Iodine deficiency), management of high-risk pregnancies (such as women with diabetes, epilepsy, thyroid disease), increasing awareness regarding teratogenic risk exposures (environmental exposures, risk of self-prescription, need for routine ante-natal care) and healthy pregnancy practices. Genetic disorders like sickle cell anemia or thalassemia, as well as conditions like mental retardation that may arise due to chromosomal anomalies can be prevented using genetic testing. Genetic counseling, the process by which genetic testing is offered, is considered to be resource intensive. That genetic counseling can be offered and can be effective when delivered through the services of non-governmental organizations (NGOs) is reflected by the activities of the Hemophilia Federation of India. This NGO provides services for patients and refers parents for genetic counseling. A study on 20 year trends of hemophilia in the state of Maharashtra indicates that this lay genetic counseling has brought about a statistically significant reduction in the number of families having more than one son with hemophilia [10-12].

BIRTH DEFECTS UNDER THE RBSK

Birth defects prevention has received attention for the first time in India as it has been incorporated as a component of a newly launched child health screening and early intervention services program (RBSK) [4] RBSK has been launched in February 6, 2013. This new program has been envisaged to screen and treat or manage a set of health conditions, including defects at birth, nutritional deficiencies, diseases and developmental delays including disability. The program intends to cover more than 27 crore children from birth to 18 years in a phased manner. The program has identified 30 health conditions for screening and management including birth defects like neural tube defect, Down's syndrome, cleft lip and palate, club foot, developmental dysplasia of the hip, congenital cataract, congenital deafness, congenital heart diseases, retinopathy of prematurity, deficiency conditions like anemia, Vitamin A deficiency, Vitamin D deficiency, severe acute malnutrition, goiter, common childhood diseases like skin conditions, otitis media, rheumatic heart disease, reactive airway disease, dental caries, convulsive disorders, developmental delays and disabilities like vision impairment, hearing impairment, neuro-motor impairment, motor delay, cognitive delay, language delay, behavior disorder (Autism) and learning disorder. In addition, sickle cell anemia, thalassemia and congenital hypothyroidism would be covered based on epidemiological data and incidence and prevalence of the same [13].

All newborns born at public health facilities and at home would be screened for birth defects by health personnel and ASHA respectively, 6 weeks to 6 years at Anganwadi centers

and 6 years to 18 years enrolled in government and government-aided schools. These children would then be referred to an appropriate facility for further management. In order to implement this program of such a grand magnitude, dedicated Mobile Block Health Teams would be recruited and stationed at the block level (three per block) consisting of two AYUSH Doctors (one male, one female), one ANM/Staff Nurse and one Pharmacist. (Ayurveda, Yoga, Unani, Siddha and Homoeopathy, i.e. AYUSH are rationally recognized systems of medicine and have been integrated into the national health delivery system).

In addition, a District Early Intervention Centre (DEIC) would be operationalized, consisting of a multidisciplinary team viz. Medical Professionals (Pediatrician, Medical Officer and a Dental doctor), Physiotherapist, Audiologist and Speech Therapist, Psychologist, Optometrist, Early Interventionist cum Special Educator cum Social Worker, Lab Technician, Dental Technician, Manager and a Data Entry Operator and provide referral services to children referred for confirmation of diagnosis and appropriate treatment. DEIC staff would visit all newborns delivered at District Hospital including those admitted in Special Newborn Care Units irrespective of their sickness for hearing, vision, congenital heart diseases before discharge, and will ensure all children born sick or preterm or low birth weight or with birth defects or referrals related to developmental delays are followed up. Furthermore, laboratory technicians of the DEIC would screen children for inborn errors of metabolism and other disorders at the district level and will ensure the linkage with tertiary level care facilities.

The Mobile Block Health Teams will undertake at least twice yearly visits to anganwadi centers to screen children aged 6 weeks to 6 years and, at least once a year; they will visit all government and government-aided schools to screen children in the age group of 6-18 years. The children identified as requiring further management will be referred to DEIC for confirmation of their diagnosis and further care.

Potential challenges to implement RBSK include the operationalization of DEIC, training of DEIC staff in various confirmatory tests and monitoring of data expected to cover over 270 million children. The success of the program would be based primarily on three pillars viz. recruitment of human resources and their capacity building, regular and uninterrupted supply of necessary logistics i.e. training manuals, registers, formats etc. and information, education and communication, including behavior change, communication to spread awareness about this new initiative [13,14]. In the near future, birth defects reporting could be incorporated into the Mother Child Tracking System and hence that the burden of these conditions on the public health system, including their impact on early neonatal mortality can be estimated. Data availability would help program managers with focused planning and including appropriate budget planning for addressing the challenge of birth defects in their State Programme Implementation Plans. Once the actual data would be made available, focused interventions on prevention could be upscaled.

CONCLUSION

Development of a national birth defects program with components of prevention, care, surveillance, advocacy, research and training is needed, as in absolute numbers, birth defects may account for a significant burden in India. Development of appropriate interventions should take into consideration the sensitivity, confidentiality and support needed for referral of affected families. The role of research for the characterization of this newly emerging field of birth defects is extremely important in guiding policy for a national birth defects program. Prevention of genetic disorders through genetic counseling is urgently required as these conditions are extremely traumatic to patients and families.

KEY MESSAGES

1. In absolute numbers, birth defects may be an unrecognized public health problem in India, requiring immediate attention as there are very few services for affected patients from the lowest socio-economic strata.
2. Preconception care can not only help prevent about half of the birth defects [14], but also reduce prematurity, stillbirths and improve pregnancy outcome by providing women knowledge on healthy pregnancy habits.
3. Research on the epidemiology of birth defects is immediately needed in order to characterize this invisible public health problem in India.
4. District Early Intervention Centers provide an opportunity for diagnosis and care for affected rural children, but a national level dialogue is immediately needed in order to ensure a feasible and sustainable service.

REFERENCES

1. Boyle CA, Cordero JF. Birth defects and disabilities: A public health issue for the 21st century. Am J Public Health. 2005;95(11):1884-6.
2. Disease Control Priorities Project Controlling Birth Defects: Reducing the Hidden Toll of Dying and Disabled Children in Low-Income Countries. Available from: http://www.dcp2.org/main/Home.html. [Last accessed on 2014 Sep 12].
3. Christianson A, Modell B. Medical genetics in developing countries. Annu Rev Genomics Hum Genet. 2004;5:219-65.
4. Operational Guidelines on Rashtriya Bal Swasthya Karyakram (RSBK). Ministry of Health and Family Welfare, Government of India; 2013. Available from: http://www.nrhm.gov.in/nrhm-components/rmnch-a/child-health-immunization/rashtriya-bal-swasthya-karyakram-rbsk/2013-12-19-08-30-24.html. [Last accessed on 2014 Sep 12].
5. Penchaszadeh VB, Christianson AL, Giugliani R, Boulyjenkov V, Katz M. Services for the prevention and management of genetic

disorders and birth defects in developing countries. Community Genet. 1999;2(4):196-201.
6. Christianson A, Howson CP, Modell B. March of Dimes Global Report on Birth Defects. 2006 March of Dimes Birth Defects Foundation, White Plains. Available from: shttp://www.marchofdimes.com/downloads/birth_defects_report-pf.pdf. [Last accessed on 2014 Sep 12].
7. Census of India, 2011 Vital Statistics. Available from: http://www.censusindia.gov.in/vital_statistics/SRS_Bulletins/MMR_release_070711.pdf. [Last accessed on 2014 Sep 12].
8. Liu L, Johnson HL, Cousens S, Perin J, Scott S, Lawn JE, et al. Global, regional, and national causes of child mortality: An updated systematic analysis for 2010 with time trends since 2000. Lancet. 2012;379(9832):2151-61.
9. Million Death Study Collaborators, Bassani DG, Kumar R, Awasthi S, Morris SK, Paul VK, et al. Causes of neonatal and child mortality in India: A nationally representative mortality survey. Lancet. 2010;376(9755):1853-60.
10. Nakade J, Potnis-Lele M, Kar A. Impact of genetic counselling? The potential utility of haemophilia surveillance data in developing countries. Haemophilia. 2013;19(6):e388-90.
11. Seshadri S, Oakeshott P, Nelson-Piercy C, Chappell LC. Prepregnancy care. BMJ. 2012;344:e3467.
12. Kerber KJ, de Graft-Johnson JE, Bhutta ZA, Okong P, Starrs A, Lawn JE. Continuum of care for maternal, newborn, and child health: From slogan to service delivery. Lancet. 2007;370(9595):1358-69.
13. Biswas T. India launches programme for child-health screening. Lancet. 2013;381(9869):e8.
14. Czeizel AE, Intôdy Z, Modell B. What proportion of congenital abnormalities can be prevented? BMJ. 1993;306(6876):499-503.

Funding: None; Conflict of Interest: None Stated

How to cite this article: Khurmi MS, Gupta M, Chaudhari G. Addressing the public health challenge of birth defects in India. Indian J Child Health. 2014;1(3):95-8.

Doi: 10.32677/IJCH.2014.v01.i03.001