Birth defects (structural, functional and metabolic disorder present from birth, may be diagnosed later) rising up as an important cause of infant mortality even in developing countries where infant mortality has been reduced to much extent. Seventy percent of birth defects are preventable through the application of various cost effective community genetic services.

Indian people are living in the midst of risk factors for birth defects, e.g., universality of marriage, high fertility, large number of unplanned pregnancies, poor coverage of antenatal care, poor maternal nutritional status, high consanguineous marriages rate, and high carrier rate for hemoglobinopathies. India being the second most populous country with a large number infant born annually with birth defects should focus its attention on strategies for control of birth defects. Many population based strategies such as iodization, double fortification of salt, flour fortification with multivitamins, folic acid supplementation, periconceptional care, carrier screening and prenatal screening are some of proven strategies for control of birth defects. Strategies such as iodization of salt in spite of being initiated for a long time in the past do have a very little impact on its consumption (only 50% were using iodized salt). Community genetic services for control of birth defects can be easily flourish and integrated with primary health care in India because of its well established infrastructure and personnel in the field of maternal and child health care. As there is wide variation for infant mortality rate (IMR) in different states in India, so there is a need of deferential approach to implement community genetic services in states those had already achieved national goal of IMR. On the other hand, states those have not achieved the national goal on IMR priority should be given to management of other causes of infant mortality.

Current Situation and Burden of Birth Defects

Birth defects can be defined as structural or functional abnormalities, including metabolic disorders, which are present from birth. The term congenital disorder is considered to have the same meaning and two terms are used interchangeably.\[1\]

According to March of Dimes (MOD) Global Report on Birth Defects,\[2\] worldwide 7.9 million births occur annually with serious birth defects and 94% of these births occur in the middle and low income countries. According to joint World Health Organization (WHO) and MOD meeting report, birth defects account for 7% of all neonatal mortality and 3.3 million under five deaths.\[1\] In India birth defects prevalence varies from 61 to 69.9/1000 live births.

Major birth defects include congenital heart defects, neural tube defects (NTDs) and Down syndrome, hemoglobinopathies and glucose-6-phosphate dehydrogenase deficiency, cause 20% of infant mortality and are responsible for a substantial number of childhood hospitalizations.\[3\] It has been estimated that 70% of birth are preventable.\[1\]

Risk Factors for Occurrence of Congenital Birth Defects in Indian Scenario

Maternal age at conception

In incidence of Down syndrome is related to fertility status of older (>35 years) female which constitute around 17% of the female population.\[4\] According to National Family Health Survey 3 (NFHS 3) report,\[5\] fertility rates among
women of age group of 35-49 are around 53 live birth per 1000 female of the same age group. Based on these estimates around 5.26 million births occur annually to these females. Sixty five percent of such births are of four or more order. These births are also at maximum risk for occurrence of Down syndrome. In India, annual birth of Down syndrome babies is around 37,000 taking incidence of down syndrome as 1.4/1000 live birth.[6]

Proportion of unplanned pregnancies and no antenatal care

Unplanned pregnancies and no antenatal care straightway means pregnancies not benefitted from preventive strategies against birth defects. According to NFSH 3,[5] report 22.8% of pregnant female didn’t receive any antenatal care and 33% received any type of ANC care at 4 month or even later; 10% pregnancies were mistimed and 11% were not wanted at all. Common reasons for unplanned pregnancies reported in studies[7] are contraceptive failure, lack of access to family planning information and services, personal or religious beliefs, inadequate knowledge about the risks of pregnancy following unprotected sexual relations, women’s limited decision making with regard to sexual relations and contraceptive use, and incest or rape.

Medical condition of mother

Exact prevalence of chronic conditions like diabetes, epilepsy, hypertension during pregnancy is not known, but it has been documented by study,[8] that about 8% of pregnant women need permanent drug treatment due to various chronic diseases and pregnancy-induced complications.

Rate of consanguineous marriages in the population

Based on recent estimates,[8] consanguinity rates in India varies from as low as 1% to 4% in the northern region to as high as 40-50% in the southern region. In comparison to a non-consanguineous couple, consanguineous are more likely to have (1) early age at marriage and at first birth (2) higher number of infants born (3) same or lower rates of abortion, (4) higher rates of postnatal mortality (5) higher rates of congenital malformations and genetic disorder.

Parent’s carrier status of a genetic disorder

Carrier frequencies for various genetic disorders like thalassemia, sickle cell anemia, and metabolic disorder are high among Indians. Carrier frequencies for sickle cell hemoglobin ranges from 17% to 30% or more in the population.[10] Hb E is found in the eastern half of the Indian sub-continent, and throughout South-East Asia, where in some areas, carrier rates may exceed 60% of the population. The carrier frequency for β thalassaemia ranges from 0.3% to 15%, while that for the milder forms of α thalassaemia varies from 15% to 80% (tribal population) in north eastern parts of India.[10]

Maternal nutritional status

Maternal deficiencies of iodine and folic acid and other macro and micro nutrient found to be associated with birth defects. According to NFHS 3, just over half (51%) household was using salt that was adequately iodized. Fifty-five percent of women were found to be anemic. Anemia is more prevalent for women who are breastfeeding (63%) and women who are pregnant (59%) than other. More than one-third (36%) of women have a BMI below 18.5, indicating a high prevalence of nutritional deficiency.

Exposures to teratogens before and after conception

According to NFHS 3, 11% of reproductive age group female was using any form of tobacco and its usage was almost equal among pregnant and non-pregnant women. Children of women who smoke during pregnancy are found to have multiple birth defects 1.5-2 times more than expected.[11] Alcohol, which is associated with fetal alcohol syndrome, usage among reproductive age group female was 2.2%. According to study on drug utilization pattern during pregnancy,[12] exposures to radiation and category X drug occur in 4% and 5.71% pregnant female respectively during the first trimester. Exposures to other categories of drugs during their first trimester vary from 55.28% for category A to 6% for category D drugs respectively. Moreover, easy availability of drugs coupled with inadequate health services, compound the problem of intake of non-prescribed drugs and self-medication.[13]
Strategies for Prevention of Birth Defects in Indian Context

Under the Action Plan for Global Strategy for the Prevention and Control of non-communicable diseases 2008-2013, prevention and care of the birth defects was given due emphasis. According MOD and WHO report, 70% of the birth defects are preventable if the evidence-based community genetics services are used. Community genetics services include a number of activities for the diagnosis, care and prevention of genetic diseases at the community level. The goal of community genetic services would be to maximize the chances for having healthy babies.

Community genetic services have been given low priorities in India because of the paucity of resources, inadequacy of data on the burden of birth defects and insufficient number of trained health personnel. Moreover, various cultural, religious and social issues play a major role in management of the birth defects. As India has already reduce the IMR to a level of 47/1000 live births. It has been seen that as the infant mortality will go down proportional death form birth defects would rise up.

A number of states in India had achieved an IMR less than 30/1000 live birth (National Health Policy 2002 goal), i.e., Kerala (13), Goa (10), Delhi (30), Tamilnadu (24), Maharashtra (28), Chandigarh (22), Pondicherry (22), Lakshadweep (25), Andaman and Nicobar (25), Daman and Diu (23) etc., Community genetic services can be started on deferential approach basis; to implement community genetic services in states those who had already achieved national goal of IMR. On the other hand, states those have not achieved the national goal on IMR priority should be given to management of other causes of infant mortality.

Some of these strategies have already been implemented in India, but these are not up to the mark such as iron folic acid supplementation during pregnancy. More than 95% of all NTDs are first in occurrence, with a
small proportion being repeated events in women.\textsuperscript{[22]} In India as more than half of the pregnancies are unplanned thereby such pregnancies rarely benefited from providing folic acid supplementation for prevention of NTD as NTDs are caused by failure of the neural tube to close between 21 and 28 days following conception, has already occurred at the time of first ANC care. Fortification of food commodity with multivitamins including folic acid has been found to be a successful strategy for the prevention of NTDs in a number of other countries. In India work has also been in this direction under the micronutrient initiative (MI), an international organization aimed to eliminate micronutrient deficiencies in the world’s most vulnerable populations. Under MI, India flour fortification network (IFFN) was set up to eradicate the micronutrient malnutrition in India. IFFN has been working actively with various stakeholders like governments development agencies, health and medical research institutes, flour millers associations and nongovernmental organizations to adopt the flour fortification. Around 44.52 million Indians now have access to fortified flour supplied in various states.\textsuperscript{[23]} Most challenging job is to provide the accesses to marginalised and neglected population on a regular basis through public distribution system. More ground level research and attention is needed towards the universal iodization and double fortification of salt as even after the 50 years of completion of iodine deficiency disorder control program 50% of the population not using adequately iodized salt.

**Birth defects registry**

Although a few small hospital-based studies added to birth defects statistics, but nationwide prevalence of birth defects is not known. There is a need of more systematic surveillance for birth defects in India as India is second most populous country with around 27 million children born every year. In view of this Fetal Care Research Foundation established the Birth Defects Registry of India (BDRI) in 2001. BDRI currently covers 28 states and 3 union territory including Delhi, Chandigarh and Pondicherry. Reported prevalence of birth defects provided by BDRI in 2010 was 84.2/10000, much lower than estimated prevalence of at least 2%.\textsuperscript{[24]} True prevalence can be found out by establishing more population and hospital based registry. Moreover, various nationwide surveys currently conducted for collection of RCH statistics can be used to collect information on birth defects which can be used a base for further research and program development.

**National program for commonly occurring birth defects or hemoglobinopathies**

A national program for thalassemia and sickle cell anemia should be initiated on priority basis as the carrier rate for both conditions is high in India. Moreover, it is associated with great expense and difficulties in providing optimal treatment for patients, and the high mortality from untreated cases.\textsuperscript{[25,26]} Thalassemia treatment services are available in India, but these are not in reach on every needy individual because of high cost. This indicates towards high priority for initiation of a national program for prevention of hemoglobinopathies based on the proven cost effective strategies experienced by other countries. Policy should determine the carrier status of subjects before reproduction, or during early pregnancy, so that prenatal diagnosis can be obtained in the at risk couples.

Other major area need attention are commitment from policy makers to handle the problem, education of the public about genetic disorder and their prevention, training of medical professionals in taking comprehensive and specific medical family history and genetic counseling and trained health staff in the field of medical genetics.

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