

Prevention of Birth Defects: A Priority
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Editorial

Under the aegis of the United Nation's Millennium Development Goal 4 (MDG 4), the global under five childhood mortality rate (U5MR) has declined from 12.7 million in 1990 to 6.3 million in 2013. But despite these gains, the Infant Mortality Rate (IMR) in 2013 was 4.6 million (74% of U5MR) [1]. One of the leading causes of this high mortality and morbidity are Birth Defects (BD) or Congenital Malformations (CM).

Birth defects are abnormalities affecting body structure or function that are present from birth. Some of these defects are major and life-threatening, but majority are minor defects affecting bodily function in various ways. They can occur in any family and can have devastating effect on the parents and the family. An estimated 8 million babies (6% of worldwide births) are born with BD each year and along with preterm births they account for over 4 million deaths annually. An analysis of the data from the World Health statistics 2015 reveals that as the number of under 5 deaths due to malaria, diarrhoea and measles have declined between 2000 and 2013, U5MR due to CM has witnessed a gradual increase [2].

The proportion of births with birth defects as well as the absolute number of births are much higher in the third world countries than in high-income countries because of sharp differences in maternal health and other significant risk factors, including poverty, a high percentage of older mothers, a greater frequency of consanguineous marriages and the survival advantage against malaria for carriers of sickle cell, thalassemia, and glucose-6-phosphate dehydrogenase (G6PD) deficiency genes. Several studies have attributed consanguinity as an independent risk factor of BD, independent of ethnic origin [3-4]. The studies also highlighted the fact that the risk of birth defects was higher in population with high frequency of consanguineous marriages. For instance, the reported incidence of consanguineous marriages in India varies from 1.4% in the state of Assam to 30.8% in Pondicherry and 26.4% in Maharashtra [5-7].

Exact cause of many of the BD is not known. Some genetic and environmental factors, either alone or in combination may be responsible. The five most common birth defects of genetic and partially genetic origin are: congenital heart disease, neural tube defects, hemoglobin disorders such as Thalassemia and Sickle cell disease, Down's syndrome and Glucose 6-phosphate dehydrogenase deficiency. These defects account for about 25% of more than 7000 birth defects of genetic and partial genetic origin. Non-genetic or post-conception birth defects are more common in low and middle income countries, where teratogenic environmental factors play a significant role. Dietary factors, life-style choices, environmental pollutants, poor living conditions, use of non-prescription medications before and during pregnancy etc are some of such factors.

Early diagnosis, appropriate care and intervention and proper preventive strategies in early pregnancy go a long way in bringing down the death and disability rates among these children. It is now known that 70% of birth defects can either be prevented, or that affected children can be offered care that could be life saving or would reduce the severity of disability. Preventive strategies include taking multivitamin tablets with folic acid daily by the mother since peri-conceptual period, maintaining a healthy weight, having regular checkups, learning about family history and genetic risk, managing maternal medical conditions like diabetes and minimizing unnecessary medication exposure during pregnancy.

The World Health Assembly report, 2010 describes the basic components for creating a national programme for the prevention and care of birth defects before and after birth [8]. It also recommends priorities for the international community to assist in establishing and strengthening of these national programmes. WHO, in collaboration with few other international agencies have developed normative tools, including guidelines and a global plan of action, to strengthen medical care and rehabilitation services to support the implementation of the Convention on the Rights of Persons with Disabilities. WHO also supports countries to integrate medical care and rehabilitation services into overall primary health care and development of community-based rehabilitation programmes. It is time the healthcare policy makers formulate effective programmes and strategy for prevention of BDs so as to root out this evil from our society.

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