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# Editorial

## **BIRTH DEFECT**

Birth defect is a widely used term for a congenital malformation; birth defects are structural or functional abnormalities present at birth that can cause physical, intellectual and developmental disability, and other health problems. Some may be fatal, especially if not detected and treated early. Although some birth defects are inherited, others are a product of harmful environmental factors known as teratogens, and still others are multifactorial, resulting from a complex interaction of genetic and environmental influences. However, in approximately half of all birth defect cases, the causes are unknown<sup>1</sup>.

Birth defects affect an estimated 1 in 33 infants and result in approximately 3.2 million birth defect-related disabilities every year<sup>2</sup>. An estimated 270 000 newborns die during the first 28 days of life every year from congenital anomalies<sup>3</sup>. Congenital anomalies may result in long-term disability, which may have significant impacts on individuals, families, health-care systems and societies. The most common severe congenital anomalies are heart defects, neural tube defects and Down syndrome.

Researchers have identified thousands of different birth defects. There are more than 4,000 different known birth defects, ranging from minor to serious, and although many can be treated or cured, they're the leading cause of death in the first year of life. Metabolic defects affect 1 in 3,500 babies and usually involve a missing or incorrectly formed enzyme, a protein necessary for processing chemical substances in the body. Most children with a metabolic birth defect do not have any visible abnormality, but metabolic defects are usually harmful or can be even fatal<sup>4</sup>. Defects caused by congenital infections result when a mother gets an infection before or during the pregnancy. Infections that can cause birth defects include rubella, cytomegalovirus, syphilis, toxoplasma, parvovirus,

and, rarely, chickenpox<sup>5-7</sup>. Maternal infections such as syphilis and rubella are a significant cause of birth defects in low- and middle-income countries<sup>1</sup>. If the mother is infected during early pregnancy, rubella carries the highest risk for birth defects (approximately 20%). Most birth defects occur in the first 3 months of pregnancy, when the organs of the baby are forming. This is a very important stage of development; however, some birth defects occur later in pregnancy as during the last six months of pregnancy, the tissues and organs continue to grow and develop. Some women have a higher chance of having a child with a birth defect who smoke, or drink alcohol during pregnancy and when there is birth defect in family <sup>8-11</sup>.

Consanguinity increases the prevalence of rare genetic congenital anomalies and nearly doubles the risk for neonatal and childhood death. Iodine deficiency, folate insufficiency, obesity, or uncontrolled diabetes mellitus are linked to some congenital anomalies<sup>12-14</sup>. For example folate insufficiency increases the risk of having a baby with neural tube defects<sup>15</sup>.

Although congenital anomalies may be genetic, infectious or environmental in origin, most often it is difficult to identify the exact causes. Many congenital anomalies can be prevented, pre- and peri-conception and prenatal health care services can decrease the frequency of certain congenital anomalies. Primary prevention of congenital anomalies includes: ensuring an adequate dietary intake of vitamins and minerals and particularly folic acid and iodine, and abstaining from or restricting intake of harmful substances, particularly the use of alcohol and smoking, controlling pre-conceptional and gestational diabetes through counseling, weight management, diet and the administration of insulin when needed.

In 2010, the World Health Assembly issued a report on birth defects. The report describes the basic components for creating a national programme for the prevention and care of birth defects. It also recommends priorities for the international community to assist in establishing and strengthening these national programmes<sup>16</sup>.

As Bangladesh is making progress in maternal and neonatal health and reducing newborn deaths from common causes like sepsis, asphyxia, deaths due to congenital birth defects are gradually increasing in proportion. Now time has come to focus on the issue of congenital birth defects and its prevention and management.

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#### References

- Christianson A, Howson CP, Modell B, editors. March of dimes global report on birth defects: the hidden toll of dying and disabled children. New York: March of Dimes Birth Defects Foundation, White Plains, 2006
- Centers for Disease Control and Prevention. Update on Overall Prevalence of Major Birth Defects—Atlanta, Georgia, 1978-2005. MMWR Morb Mortal Wkly Rep. 2008;57(1):1-5
- WHO. Congenital anomalies. Fact sheet N°370. Updated January 2014, http://www.who.int/ mediacentre/factsheets/fs370/en/
- 4. Kochanek KD, Kirmeyer SE, Martin JA, Strobino DM, Guyer B. Annual summary of vital statistics: 2009. Pediatrics 2012; 129: 338-348
- M,Edlich RF, Winters KL, Long WB 3rd, Gubler KD. Rubella and congenital rubella (German measles). *J Long Term Eff Med Implants* 2005; 15(3): 319-328
- Centers for Disease Control and Prevention (CDC). Congenital syphilis - United States, 2003-2008. MMWR Morb Mortal Wkly Rep 2010; 59:413
- 7. Stegmann BJ, Carey JC.TORCH Infections. Toxoplasmosis, Other (syphilis, varicella-zoster,

parvovirus B19), Rubella, Cytomegalovirus (CMV), and Herpes infections. Curr Womens Health Rep. 2002 Aug;2(4):253-8

- Ericson, A., Källén, B., & Westerholm, P. Cigarette smoking as an etiologic factor in cleft lip and palate. American Journal of Obstetrics and Gynecology 1979;135: 348-351
- Hackshaw A, Rodeck C, Boniface S. Maternal smoking in pregnancy and birth defects: a systematic review based on 173 687 malformed cases and 11.7 million controls. Hum Reprod Update. 2011 Sep-Oct;17(5):589-604
- Hanson, J. W., Jones, K. L., & Smith, D. W. Fetal alcohol syndrome. Experience with 41 patients. Journal of the American Medical Association 1976; 235: 1458-1460
- Wong-Gibbons DL, Romitti PA, Sun L, Moore CA, Reefhuis J, Bell EM, Olshan AF.Maternal periconceptional exposure to cigarette smoking and alcohol and esophageal atresia +/- tracheoesophageal fistula. Birth Defects Res A Clin Mol Teratol 2008 Nov;82(11):776-84
- Correa A, Gilboa SM, Besser LM, Botto LD, Moore CA, Hobbs CA, Cleves MA, Riehle-Colarusso TJ, Waller DK, Reece EA.Diabetes mellitus and birth defects.Am J Obstet Gynecol. 2008 Sep;199(3):237
- Waller DK, Shaw GM, Rasmussen SA, Hobbs CA, Canfield MA, Siega-Riz AM, Gallaway MS, Correa A. Prepregnancy obesity as a risk factor for structural birth defects. Arch Pediatr Adolesc Med. 2007 Aug;161(8):745-50
- Potter, J. D., McMichael, A. J., & Hetzel, B. S. Iodization and thyroid status in relation to stillbirths and congenital anomalies. International Journal of Epidemiology1979; 8: 137-144
- 15. Youngblood ME, Williamson R, Bell K. 2012 Update on global prevention of folic acidpreventable spina bifida and anencephaly. Birth Defects Res A Clin Mol Teratol 2013; 97:658
- 16. WHO. Sixty-third World Health Assembly. Birth Defects. Report by the Secretariat.1 April 2010.