

Birth Defects

Birth defects, specifically malformations – congenital malformation, chromosomal anomaly, or persistent metabolic defect – are the leading cause of death in the first year of life. These defects also follow an infant into childhood and are a major cause of morbidity and mortality¹. A birth defect is defined as an abnormality of structure, function or body metabolism present at birth that results in physical or mental disability, or is fatal. As with most health related issues, genetics and environment play an equal role. About 11,000 babies in New York State are born each year with some form of birth defect. In 2004, 320 babies were born in the Capital Region (Albany- 135, Schenectady- 100, Rensselaer- 85) with a major congenital malformation.

The *Healthy People 2010 Objective (16-1f)* is to decrease the incidence of all birth defects to 1.1/1,000 live births. Although many of the causes of birth defects are still unknown and therefore unpreventable, a woman can take numerous steps to reduce her baby's risk of being born with birth defects. Some birth defect risks can be detected preconceptionally through genetic testing. Women who are planning to become pregnant and their partners can discuss their risks through genetic counseling during the preconception period. Although genetic testing may result in dilemmas regarding the possible termination of the pregnancy, if there is a history of birth defects is prevalent in ones family, additional information about the developing fetus is always useful.

¹ Lippig KA, Werler MM, Caron CI, Cook CA, Holmes LB. Predictive value of minor abnormalities: Association with major malformations. *J Pediatr* 1987; 110:530-537; New York State Department of Health Congenital Malformations Registry: Annual Report 1994 (Publication Year 1998); New York State Department of Health Congenital Malformations Registry: Annual Report 1994 (Publication Year 1999).