

## Review Article

# Birth defects: Risk factors and consequences

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**Abstract.** Birth defects (BDs) or congenital anomalies include all structural and functional alterations in embryonic or fetal development resulting from genetic, environmental or unknown causes, which result in physical and/or mental impairment. BDs occur in about 3% of newborn babies and in most cases of pregnancy loss. BDs are a very complex and heterogeneous group of single or multiple changes that, in most cases, are of unknown etiology. Among the risk factors are advanced maternal and paternal ages, parental consanguinity, teratogenic agents such as infectious agents and drugs, and poor nutrition, in particular folic acid deficiency. One of the consequences of these defects is the high death rate within the first year of life. Information on BDs is becoming increasingly more important throughout the world so that preventive measures can be taken. Knowledge of BDs enables the development of therapeutic and preventive strategies besides adequate genetic counseling.

**Keywords:** Congenital anomalies, congenital malformation, chromosomal abnormalities

## 1. Classification and etiology

Congenital anomalies or birth defects (BDs) include all functional and structural alterations in embryonic or fetal development resulting from genetic, environmental or unknown causes that originate during pregnancy, are present at birth and that cause physical or mental impairment [1].

BDs are a very complex and heterogeneous group of single or multiple changes that, in most cases, are of unknown etiology. The genetic etiology of BDs is relatively well known and includes monogenic causes (about 20% of cases), chromosomal alterations (approximately 25%), which may be numerical or structural and multifactorial causes (about 50% of cases). The etiology of environmental factors responsible for

disruptive effects exists in approximately 5% of cases and these cases are the most difficult to investigate. Aneuploidy is the cause of BDs in more than 30% of all cases; most are monosomy of chromosome X and trisomies of chromosomes 16, 21 and 22, and are responsible for about 60% of gestational losses. Although trisomies of chromosomes 13, 18 and 21 are responsible for many pregnancy losses, they also have an impact at birth, as they are present in approximately 0.3% of all births together with trisomy of chromosome X [2–4].

Thus, genetic factors are among the most common causes of BDs and are one of the main causes of infant mortality. BDs are observed in about 3% of newborns and are the cause of 50% of spontaneous neonatal deaths [5–8].

Stillbirths occur in between 1 in 1000 and 1 in 160 pregnancies and are related to maternal, fetal, and placental causes; in half of the cases, no cause can be determined. BDs occur in 1–4% of these cases with genetic diseases being a common cause [9,10]. Stillbirths can

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# Birth Defects Risks And Consequences

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