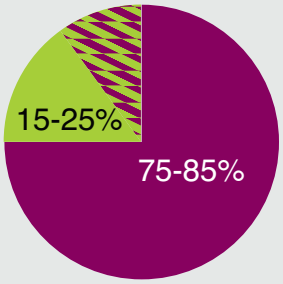
5.1.3 Causes of Congenital Anomalies

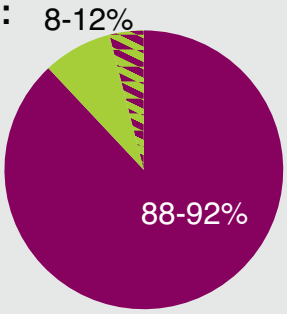
Recognized genetic conditions:
Chromosome and single gene causes

15% to 25% are due to recognized genetic conditions



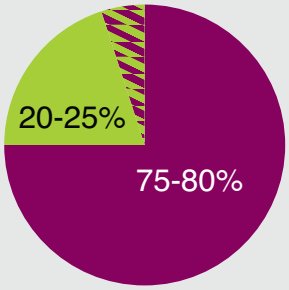
Environmental factors:
Maternal-related conditions, drug or chemical exposures

8% to 12% are due to environmental factors



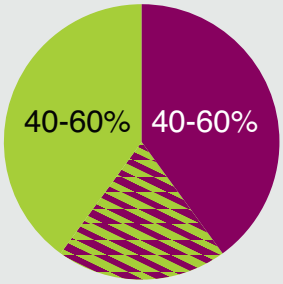
Multifactorial inheritance*

20% to 25% are due to multifactorial inheritance



Unexplained causes

40% to 60% are due to unexplained causes



* A congenital anomaly is considered to be multifactorial (or polygenic) in origin when there is a combined influence of (a number of) genes and environmental factors that interfere with normal embryologic development. Multifactorial inheritance is considered when there appears to be a genetic component but there is no clear Mendelian pattern of inheritance. Multifactorial inheritance is the underlying etiology of most of the common congenital anomalies.

Graphic created by CICH using data from Health Canada. Congenital Anomalies in Canada — A Perinatal Health Report, 2002. Ottawa: Minister of Public Works and Government Services Canada, 2002.

Some congenital anomalies are caused by mutations in a single gene or damage to a specific chromosome. Other congenital anomalies are due to exposure to environmental hazards or drugs during pregnancy. Others result from a combination of genetic and environmental influences. However, the cause of most congenital anomalies is unknown. Most children with congenital anomalies are born to mothers with no family history and no known risk factors.