
Genetic Services Policy Project

Multiple Congenital Anomalies

What are multiple congenital anomalies (MCA)?

Infants with multiple congenital anomalies (MCA) are typically infants with:

- two or more major malformations (e.g., a neural tube defect, cardiac defect, missing limb), or
- three or more minor malformations (e.g., syndactyly, a club foot, abnormally formed pinnae).

Common abnormalities include cardiac defects, cleft lip/cleft palate, neural tube defects, musculoskeletal defects, abnormalities of the eye, and gastrointestinal or genitourinary defects.

Who is affected by MCA?

Birth defects affect 3 to 5% of all newborns in the United States; 1% of newborns have multiple defects or syndromes.

What causes MCA?

Approximately 40-60% of congenital malformations have no known origin. About 20% of birth defects are likely to result from genetic and environmental factors combined.

- 7.5% are caused by single gene mutations
- 6% are caused by chromosome abnormalities
- 5% are caused by maternal illness and/or substance use

Several factors must be considered in assessing MCA etiology: maternal health history, prenatal history, family history, and careful and detailed physical examination of the infant.

How are MCA detected?

Prenatal screening may identify a fetus at risk for MCA:

- Multiple marker maternal serum screening at 15 to 18 weeks gestation can identify fetuses at risk for MCA associated with chromosomal aneuploid or open lesions.
- Ultrasound at 19 to 21 weeks gestation allows sonographers to measure morphology.

Fetal MRI, fetal echocardiographs, and amniocentesis with cytogenetic or molecular testing may follow a positive prenatal screen to identify potential diagnoses and to predict prognosis. A woman or couple can then make more informed decisions about the pregnancy.

MCA are frequently not identified through prenatal screening but are noted at the time of birth.

- Clinical geneticists document malformations, signs, and symptoms in order to compare against known syndromes.
- Genetic testing may help to rule out or confirm a suspected single gene disorder or chromosomal aneuploid if maternal and pregnancy history are inconclusive.
- Biochemical studies, molecular testing, chromosomal testing and/or fluorescent in-situ hybridization (FISH) testing may help to provide a diagnosis.

What are standard treatments and therapies for MCA?

While prognosis and therapies for affected infants depend on the nature of the condition, infants with MCA usually need complex medical and surgical management. Care is tailored to clinical need and could range from palliative care to surgical and nutritional interventions. Families are typically referred to health care facilities that specialize in treating children.

Many health care professionals may be involved in evaluating, diagnosing and treating a fetus or infant with MCA: primary care providers (obstetrical or pediatric), clinical geneticists, genetic counselors, dysmorphologists, and varied pediatric specialists (e.g. cardiologists, urologists, neurologists, surgeons).

What are costs associated with MCA?

There are no estimates of costs associated with MCA because MCA may represent a multitude of conditions and birth defects. However, an analysis conducted in 1992 provides an estimate of cost of illness for cerebral palsy and for 17 structural birth defects in the United States. For 1992, the combined estimated cost of the 18 conditions in the United States was \$8 billion. The three conditions with the highest lifetime costs were cerebral palsy, Down's syndrome, and spina bifida.

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