

Trisomy 18

BIRTH DEFECT RESEARCH FOR CHILDREN



What is Trisomy 18?

Trisomy 18 is a genetic disorder which has its onset before birth and is usually fatal. In this disorder, chromosome 18 appears three times (trisomy) rather than the normal two times in the cells of the body. The most severe form of the disorder occurs when every cell in the body is affected. A less severe form, called the mosaic form, occurs when some cells have the normal chromosomal pair. Trisomy 18 is also called Edwards' Syndrome and Chromosome 18. It causes severe mental retardation and major physical abnormalities. Common findings include low birth weight, malformed and low-set ears, small jaw, hand abnormalities, congenital heart disease, hernias, feeding and breathing problems, and a weak infant cry.



Trisomy 18 (Edward's Syndrome)



How many children have Trisomy 18?

Trisomy 18 affects one in 3,000-11,000 newborns and affects girls three times more often than boys.

How do you know if your child has Trisomy 18?

Trisomy 18 can be detected before birth by several tests. These include maternal serum analysis or screening, ultrasonography, amniocentesis, and chorionic villus sampling. Serum analysis can show abnormal levels of certain hormones and proteins and ultrasonography can show an unusually shaped fetal trunk, small fetal head and chest, and excess amniotic fluid. Amniocentesis and chorionic villus sampling are methods of obtaining cells for examination and diagnosis. At birth, an unusually small placenta and physical abnormalities of the newborn, alert doctors to the possibility of Trisomy 18. It is diagnosed by a laboratory test called karyotyping which involves drawing the baby's blood or bone marrow for microscopic examination to study chromosome make-up.

What causes Trisomy 18?

Trisomy 18 occurs sporadically when, for unknown reasons, chromosomes fail to separate properly. Chances of conceiving a child with Trisomy 18 increase with maternal age. Although no lifestyle or environmental factors have been definitively identified as causing Trisomy 18, results of some studies have suggested that environmental factors may influence the risk for Trisomy 18.

How can you help a child with Trisomy 18?

Because there is no cure, treatment usually consists of supportive care with the goal of making the infant comfortable rather than prolonging life. Some abnormalities can be treated with surgery; however, extreme invasive procedures may not be in the best interest of an infant with a limited lifespan.

What's in the future for a child with Trisomy 18?

Fifty percent of the children with the most severe form of Trisomy 18 have an average lifespan of less than two months. Ninety to ninety-five percent die before their first birthday. The 5-10% who survive the first year are usually severely mentally retarded with limited verbal communication, although they can learn to recognize and interact with others. Children with the mosaic form of Trisomy 18 are usually less severely handicapped.

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