

TRISOMY 18

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TRISOMY 18

(formerly called Edwards Syndrome or Trisomy E)

Condition and Description

Trisomy 18 is a disorder in which babies are born with 3 copies of the #18 chromosome instead of 2. For an unknown reason, this accident occurs at the time of conception and all cells in the body will have this structure in 95% of the children with Trisomy 18. In 5-10% of infants with Trisomy 18, their cells may contain only a portion of the extra chromosome (called the mosaic type of Trisomy 18). Infants with Trisomy 18 will be small at birth and have a recognizable appearance caused by the extra chromosome: prominent back of the head, small eyes, mouth and jaw, unusual ears, clenched fist with overlapping fingers and thumbs, small fingernails, clubbed feet, a short breast bone (sternum) and extra skin folds at the back of the neck. There is no known cause of Trisomy 18. About 90% of babies do not survive beyond the first year and many live only a few days.

Prevalence

Trisomy 18 occurs at a rate of 1 in 3000-6000 live births. It is somewhat more common as the mother ages but can occur randomly to parents of any age. The vast majority of babies affected are girls.

Common Associated Conditions

Over 90% of infants with Trisomy 18 have a congenital heart defect. The type of heart defect varies, but most would be a ventricular

septal defect (opening between the lower heart chambers), an atrial septal defect (opening between the upper chambers), a patent ductus arteriosus (a blood vessel from fetal life that does not close), or a problem with the function of a heart valve. In about 10% of babies the heart defect is extremely severe and life-threatening (hypoplastic left heart, where the main pumping chamber is not formed). Other problems include apnea (the brain does not send a message to the body to breathe), difficulty feeding, under-developed lungs, joint and bone abnormalities (10% of babies), hearing loss (50% of babies), cleft lip (5-10% of babies), and eye defects (10% of babies). Children with this condition are also prone to develop Wilm's tumor (a tumor of the adrenal gland in the abdomen).

Short-term Treatment and Outcomes

Parents of a baby with Trisomy 18 face many difficult decisions regarding the care of their child. Some parents of severely affected infants elect to give comfort care and take their baby home to provide the best and most comfortable circumstances possible.

Long-term Treatment and Outcomes

Other families might choose to have treatment to improve the child's chances of survival and quality of life. However, this is very rarely an option, typically because of the severity of the baby's heart defects.



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Common Complications

Babies with Trisomy 18 will be small in size and will grow very slowly even if given good nutrition. They may have difficulty feeding and are prone to pneumonia from aspirating milk into their lungs (gastroesophageal reflux). Seizures, urinary tract infections and delayed development are also concerns for many babies with this condition.

Implications for Children's Development

Decision-making for children with chromosomal defects is very difficult and parents need information and support. Comfort care with the support of hospice or palliative care staff would be recommended for most families who have a baby born with Trisomy 18. Many resources are available on the internet to provide information to families in these difficult circumstances.

The birth of a severely affected child is very difficult and families will find support from other families who have experienced similar circumstances. Support groups can be found on the internet as well as in some communities. Children who do survive may make some developmental gains such as rolling over, recognizing their parents and expressing pleasure or pain in some way. A few may progress to be able to enter school with the help of early intervention and special education adaptations.

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