Trisomy 18 (Edward's syndrome)

What is it?

Trisomy 18, or Edward's syndrome, is a congenital chromosomal condition. **Chromosomes** are located in the cells in our body. Typically, people are born with 23 pairs of chromosomes. Infants born with this syndrome have three copies of chromosome 18 in each cell, whereas most people only have two copies. The additional copies of chromosome 18 cause irregularities in the body. These can lead to poor development and eventually death. Many infants diagnosed with trisomy 18 die in the womb or do not live longer than one month. Only 5%-10% of affected individuals survive past one year. Symptoms of trisomy 18 can range in severity. Common symptoms include growth restriction, intellectual disabilities, feeding problems, and breathing issues. Other notable defects include skeletal and facial deformities (small mouth/jaw, cleft palate/lip, low-set ears), irregular head shape, clenched fists, overlapping fingers, and congenital heart defects.

How common is it?

About 1 in 5,000 infants born each year will be diagnosed with trisomy 18. It is more common in girls than boys.

What causes it?

Infants born with trisomy 18 have three copies of chromosome 18 in each cell. Most people have only two copies. The additional copies of chromosome 18 cause anomalies in the body. This can lead to poor development and eventually death. This condition is usually a random occurrence and may occur during the development of the fetus. It is rarely inherited from the mother and father.

How is it diagnosed?

Trisomy 18 is confirmed by chromosomal analysis, a test that looks at and determines the number of chromosomes in a person. Trisomy 18 can be diagnosed during pregnancy by fetal ultrasound and maternal blood test or after birth with a physical exam and chromosomal analysis.

How is it treated?

Treatment will vary from person to person. The goal is to improve the overall quality of life. Your child's doctor will discuss appropriate treatment options with you.

For more information:

U.S. National Library of Medicine, Genetics Home Reference
https://medlineplus.gov/genetics/condition/trisomy-18/#inheritance

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/trisomy-18-syndrome/

National Institutes of Health-Rare Diseases
https://rarediseases.info.nih.gov/diseases/6321/trisomy-18

BDPR Indiana Birth Defects and Problems Registry

Indiana Department of Health

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