Trisomy 18

Background and Genetic Basis

The goal of this article is to briefly describe some of the features of Trisomy 18. Trisomy 18 is the most common abnormality involving chromosome 18. It is also the most severe. Most children with Trisomy 18 die before or shortly after birth. In this way, Trisomy 18 is very different from other abnormalities involving chromosome 18.

Of course, families of children with Trisomy 18 are welcome in the Chromosome 18 Registry & Research Society. However, babies and children with Trisomy 18 typically have more severe medical problems than other babies and children in the Registry. Many families want to connect with other families that are dealing with the same challenges and questions. There are several organizations besides the Chromosome 18 Registry & Research Society that are dedicated to Trisomy 18. See the section "For Additional Information" at the end of this article.

Our Mission:

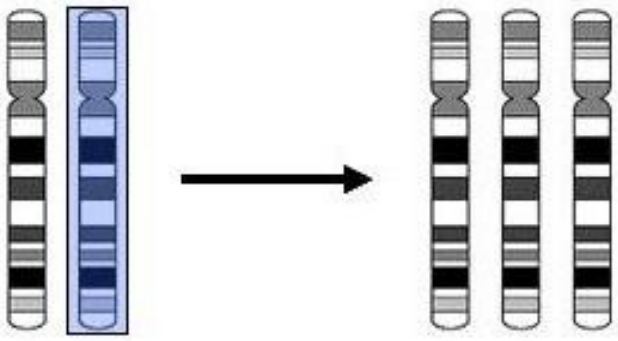
To help individuals with chromosome 18 abnormalities overcome the obstacles they face so they might lead healthy, happy, and productive lives.

Background

Trisomy 18 occurs in about 1 of every 6000-8000 live births. It is sometimes called "Edwards syndrome", after the physician that first described the condition.

Genetic Basis

Trisomy 18 occurs when there are three copies of chromosome 18 in every cell of the body. Typically, humans have two copies of chromosome 18. Sometimes, a baby will inherit an extra chromosome 18 from a parent. Therefore, the baby has three copies of chromosome 18 rather than two copies.



In the diagram above, the chromosome shaded in blue has been duplicated. This leads to one extra copy of chromosome 18.

Characteristics of Trisomy 18

Babies with Trisomy 18 typically have complex medical problems. Birth defects are very common in these babies. They may have defects of the heart, brain, spinal cord, and other internal organs. Cleft lips and palates are also more common in babies with Trisomy 18. They may have hand and foot anomalies. Babies with Trisomy 18 may have difficulty regulating their breathing and temperature. They may also be unable to feed properly.

Because of the large number of potential medical problems, babies with Trisomy 18 are usually considered "medically fragile". Most children with Trisomy 18 die before or shortly after birth. Five to ten percent of babies with Trisomy 18 live past their first birthday.

Family Planning and Genetic Counseling

Many parents wonder, "If we have another child, what is the likelihood that our next child will have Trisomy 18?"

In general, the likelihood of having a second child with Trisomy 18 is low. Studies suggest that the probability is 1% or less. However, as women age, this figure may increase slightly. This is because we know that older women are more likely to have a baby with a chromosome change than younger women.

If you have questions about the implications of a chromosome change for other family members, including future children, we recommend contacting a genetics provider.

For Additional Information

The information provided here is general information based on the literature as well as the experiences in the Chromosome 18 Clinical Research Center. However, every person with Trisomy 18 is different. Therefore, this information should not replace professional medical advice, diagnosis, or treatment. If you have questions or concerns, you may find it helpful to talk with a clinical geneticist or genetic counselor. You can locate a genetics provider at one of these sites:

GeneTests Clinic Directory (www.geneclinics.org)

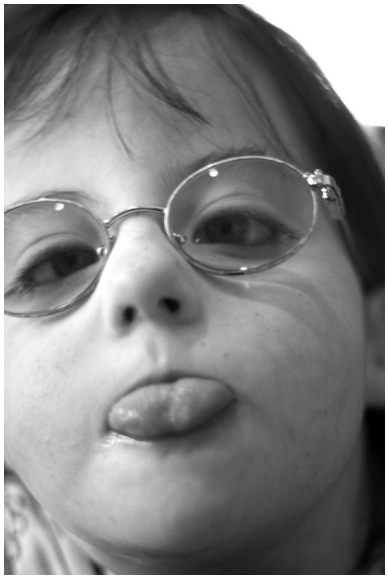
National Society of Genetic Counselors (www.nsgc.org)

You may also find it useful to contact groups that are specifically dedicated to individuals with Trisomy 18:

SOFT (www.trisomy.org)

SOFT (United Kingdom) (www.soft.org.uk)

Trisomy 18 Foundation (www.trisomy18.org)



Photographs courtesy of a Registry family.

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If you found this information helpful, or would like to learn more about the Chromosome 18 Registry & Research Society, we encourage you to become a member of our organization.

To become a member, visit our website at www.chromosome18.org or call us at 210-657-4968.

With your help, we will achieve our mission :

To help individuals with chromosome 18 abnormalities overcome the obstacles they face so they might lead healthy, happy, and productive lives.