

# Fact Sheet

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## Trisomy 18

### ***What is Trisomy 18?***

Trisomy 18 is a condition in which the cells of the body have three copies of chromosome 18, rather than the usual two.

Found inside the cells of the body, chromosomes are tiny thread-like structures that house our genetic traits. Most of our body cells have 46 chromosomes. Chromosomes come in pairs, for a total of 23 pairs. One member of each pair comes from our mom, and the other comes from our dad. The 23 pairs of chromosomes are numbered from 1 to 22. The 23<sup>rd</sup> pair of chromosomes is the sex chromosomes. The sex chromosomes determine whether we are a boy or a girl.

Trisomy 18 occurs when a problem during cell division leads to an extra copy of chromosome 18 in the cells. Instead of the typical 46 chromosomes, a child with Trisomy 18 has 47 chromosomes. This extra chromosome causes problems with the child's growth and health. Trisomy 18 occurs in both boys and girls.

### ***Is there more than one type of Trisomy 18?***

There are two main types of Trisomy 18:

- 1) **Classic Trisomy 18:** The first type of Trisomy 18 is known as classic Trisomy 18. Trisomy refers to the presence of three copies of a chromosome. In classic Trisomy 18 either the egg or sperm receives an extra copy of chromosome 18 as it is formed. Typically, the egg and sperm have only one copy of each chromosome (i.e., one copy of chromosome 18). When the typical egg and sperm unite, the resulting baby receives the full set of 46 chromosomes needed to grow. If an egg or sperm with two copies of chromosome 18 unites with an egg or sperm carrying one copy, the result is a child with three copies or Trisomy 18. Classic Trisomy 18 is the most common form of Trisomy 18, occurring in about 90 percent of cases.
- 2) **Mosaic Trisomy 18:** The second type of Trisomy 18 is called mosaic Trisomy 18. Mosaic refers to a child who has some cells with three copies of chromosome 18 and some cells with the normal two copies of chromosome 18. The two types of cells form after the egg and sperm have united. The features and problems common in Trisomy 18 may be milder in cases of mosaic Trisomy 18 since not all of the body cells carry the extra chromosome. About 10 percent of people with Trisomy 18 have a mosaic form.

### ***What are the features of Trisomy 18?***

Individuals with Trisomy 18 often share many common features. These features include a low birth weight, small head, small chin, cleft lip and/or palate, distinctive and low set ears, overlapping of fingers, clubfoot and rounded sole of foot, seizures, severe mental retardation, heart defects, kidney problems, and hernias.

***What types of problems occur in Trisomy 18?***

Trisomy 18 affects the growth of many parts of a baby's body. The effect on the body systems can be severe, and many of the body systems do not work as needed. As a result, children with Trisomy 18 usually do not do well. Sadly, about 30 percent of children with Trisomy 18 die within a month of birth, and about 90 percent die within one year.

***How common is Trisomy 18?***

Trisomy 18 occurs in about 500-1300 births in the United States each year. In Virginia, about 7 children are born yearly with Trisomy 18. Women over the age of 35 have a higher chance of having a child with Trisomy 18, but Trisomy 18 can occur with parents of any age.

***What causes Trisomy 18?***

Trisomy 18 is due to an extra copy of chromosome 18 in the cells of the body. Neither parent did anything to cause Trisomy 18. Neither parent could have done anything to prevent it.

Parents of a child with Trisomy 18 have a higher chance of having another child with Trisomy 18. This chance is usually less than 1 in 100. A genetic counselor or geneticist can help you to determine the risks for your family and situation.

***Where can I go for more information about Trisomy 18?***

Support Organization for Trisomy 13, 18 and Related Disorders

[www.trisomy.org](http://www.trisomy.org)

1-800-716-7638

March of Dimes Birth Defects Foundation

[www.modimes.org](http://www.modimes.org)

1-888-MODIMES (1-888-663-4637)

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