Fact Sheet

Division of Child and Adolescent Health
Pediatric Screening and Genetic Services
Virginia Genetics Program
1-800-523-4019
www.vahealth.org/genetics

VDH VIRGINIA DEPARTMENT OF HEALTH
Protecting You and Your Environment
www.vdh.virginia.gov

Trisomy 13

What is Trisomy 13?

Trisomy 13 is a condition in which the cells of the body have three copies of chromosome 13, rather than the usual two copies.

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 23rd pair of chromosomes is the sex chromosomes. The sex chromosomes determine whether we are a boy or a girl.

Trisomy 13 occurs when a problem during cell division leads to an extra copy of chromosome 13 in the cells. Instead of the typical 46 chromosomes, a child with Trisomy 13 has 47 chromosomes. This extra chromosome causes problems with the child's growth and health. Trisomy 13 occurs in both boys and girls.

Is there more than one type of Trisomy 13?

There are three types of Trisomy 13:

1) **Classic Trisomy 13:** The first type of Trisomy 13 is known as classic Trisomy 13. Trisomy refers to the presence of three copies of a chromosome. In classic Trisomy 13 either the egg or sperm receives an extra copy of chromosome 13 as it is formed. Typically, the egg and sperm have only one copy of each chromosome (i.e., one copy of chromosome 13). 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 13 unites with an egg or sperm carrying one copy, the result is a child with three copies or Trisomy 13. Classic Trisomy 13 is the most common form of Trisomy 13, occurring in about 75 percent of cases.

2) **Translocation Trisomy 13:** The second type of Trisomy 13 is translocation Trisomy 13. In translocation Trisomy 13, an extra piece or complete extra copy of chromosome 13 gets stuck on another chromosome. The result is three copies of chromosome 13 in the cells. In about three-fourths of cases, this translocation occurs as the egg and sperm unite to form the baby. In the other one-fourth of cases, this translocation came from one of the parents. Translocation Trisomy 13 accounts for about 20 percent of cases of Trisomy 13.

3) **Mosaic Trisomy 13:** The third type of Trisomy 13 is called mosaic Trisomy 13. Individuals with mosiac Trisomy 13 have two distinct groups of cells: cells with the typical 46 chromosomes and cells with an extra copy of chromosome 13. The two types of cells form after the egg and sperm have united. The features and problems common in Trisomy 13 may be milder in cases of mosaic Trisomy 13 since not all of the body cells carry the extra chromosome. About 5 percent of people with Trisomy 13 have a mosaic form.

What are the features of Trisomy 13?
Individuals with Trisomy 13 often share many common features. These features include a small head with a sloping forehead, cleft lip and/or palate, abnormal and low set ears, deafness, missing ribs or thin ribs, small eyes, extra fingers and toes, prominence of heels, seizures, severe mental retardation, heart defects, hernias, and genital problems.

What types of problems occur in Trisomy 13?
Trisomy 13 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 13 usually do not do well. Sadly, about 45 percent of children with Trisomy 13 die within a month of birth, and about 90 percent die within one year.

How common is Trisomy 13?
Trisomy 13 occurs in about 400-1000 births in the United States each year. In Virginia, about 6 children are born yearly with Trisomy 13. Women over the age of 35 have a higher chance of having a child with Trisomy 13, but Trisomy 13 can occur with parents of any age.

What causes Trisomy 13?
Trisomy 13 is due to an extra copy of chromosome 13 in the cells of the body. Neither parent did anything to cause Trisomy 13. Neither parent could have done anything to prevent Trisomy 13.

Parents of a child with Trisomy 13 have a higher chance of having another child with Trisomy 13. If the first child has classic Trisomy 13, this chance is about 1 in 4,000. If the first child has translocation Trisomy 13, the chance ranges from less than 1 to 15 in 100 depending on the exact translocation. 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 13?

Support Organization for Trisomy 13, 18 and Related Disorders
www.trisomy.org
1-800-716-7638

March of Dimes Birth Defects Foundation
www.modimes.org
1-888-MODIMES (1-888-663-4637)

* *This publication was supported by grant number U50/CCU321127-02 from the Centers for Disease Control (CDC). Its contents are solely the responsibility of the authors and do not necessarily represent the official views of the CDC.*