Trisomy 13

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1. Introduction

Trisomy 13, also called Patau syndrome, is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body. Individuals with trisomy 13 often have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers or toes, an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate), and weak muscle tone (hypotonia). Due to the presence of several life-threatening medical problems, many infants with trisomy 13 die within their first days or weeks of life. Only five percent to 10 percent of children with this condition live past their first year.

2. Frequency

Trisomy 13 occurs in about 1 in 16,000 newborns. Although women of any age can have a child with trisomy 13, the chance of having a child with this condition increases as a woman gets older.

3. Causes

Most cases of trisomy 13 result from having three copies of chromosome 13 in each cell in the body instead of the usual two copies. The extra genetic material disrupts the normal course of development, causing the characteristic features of trisomy 13.

Trisomy 13 can also occur when part of chromosome 13 becomes attached (translocated) to another chromosome during the formation of reproductive cells (eggs and sperm) or very early in fetal development. Affected people have two normal copies of chromosome 13, plus an extra copy of chromosome 13 attached to another chromosome. In rare cases, only part of chromosome 13 is present in three copies. The physical signs and symptoms in these cases may be different than those found in full trisomy 13.

A small percentage of people with trisomy 13 have an extra copy of chromosome 13 in only some of the body's cells. In these people, the condition is called mosaic trisomy 13. The severity of mosaic trisomy 13 depends on the type and number of cells that have the extra chromosome. The physical features of mosaic trisomy 13 are often milder than those of full trisomy 13.

3.1 The chromosome associated with Trisomy 13

- chromosome 13

4. Inheritance

Most cases of trisomy 13 are not inherited and result from random events during the formation of eggs and sperm in healthy parents. An error in cell division called nondisjunction results in a reproductive cell with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of chromosome 13. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra chromosome 13 in each cell of the body.

Translocation trisomy 13 can be inherited. An unaffected person can carry a rearrangement of genetic material between chromosome 13 and another chromosome. These rearrangements are called balanced translocations because there is no extra material from chromosome 13. A person with a balanced translocation involving chromosome 13 has an increased chance of passing extra material from chromosome 13 to their children.

5. Other Names for This Condition

- Bartholin-Patau syndrome
- complete trisomy 13 syndrome
- Patau syndrome
- Patau's syndrome
- trisomy 13 syndrome

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