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ORIGIN, TREATMENT AND PREVENTION OF PATAU SYNDROME

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Abstract : Patau's syndrome is a serious, rare genetic disorder caused by having an additional copy of chromosome 13 in some or all of the body's cells. It's also called trisomy 13.Each cell normally contains 23 pairs of chromosomes, which carry the genes you inherit from your parents. But a baby with Patau's syndrome has 3 copies of chromosome 13, instead of 2.This severely disrupts normal development and, in many cases, results in miscarriage, stillbirth or the baby dying shortly after birth.Babies with Patau's syndrome grow slowly in the womb and have a low birth weight, along with a number of other serious medical problems.Patau's syndrome affects about 1 in every 4,000 births. The risk of having a baby with the syndrome increases with the mother's age. More than 9 out of 10 children born with Patau's syndrome die during the first year.

Keys words: Patau's syndrome, blood, treatment, drug.

Trisomy 13 (Patau syndrome) is a rare genetic condition that occurs when the 13th chromosome appears three times (trisomy) instead of two times in a person's DNA. Trisomy 13 affects the development of the face, brain and heart, along with physical growth abnormalities throughout a child's body. Symptoms of trisomy 13 can be life-threatening and the condition poses a risk or miscarriage or death before the child turns 1 year old.

Trisomy 13 occurs in an estimated 1 out of 10,000 to 20,000 live births. The mortality rate is high during a baby's first few days of life and many pregnancies result in a miscarriage due to life-threatening symptoms like heart problems and spinal cord abnormalities during fetal development. Only 5% to 10% of babies born with trisomy 13 survive past their first year. Trisomy 13 will affect how your child develops, which could cause physical growth abnormalities like a cleft palate, extra fingers or toes, low muscle tone and a small head. The condition also affects the development of your child's internal organs, which could lead to life-threatening symptoms. After your child is born, they'll likely spend some time in the neonatal intensive care unit (NICU) where your healthcare provider will offer life-saving medical care to make sure your baby has the best odds of survival based on their physical symptoms.

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Symptoms of trisomy 13 affect several different parts of the body and range in severity for each person diagnosed with the condition. Symptoms of trisomy 13 include:

Heart abnormalities present at birth (congenital).

Physical growth irregularities with many cases targeting the spinal cord.

Severe issues with cognitive function.

Underdeveloped internal organs.

A third chromosome 13 that attaches to a pair of chromosomes causes trisomy 13. A person with trisomy 13 has 47 total chromosomes.

There are normally 46 chromosomes in your body. Chromosomes carry DNA in cells, which works as an instruction manual to tell your body how to form and function. Genes are segments of your DNA that are chapters in your body's instruction manual.

Cells initially form in reproductive organs by dividing from one fertilized cell, which is a combination of the sperm and the egg. The newer cells divide and copy themselves with half the amount of DNA as the original cell. During this process of cell division, a trisomy (when a third chromosome joins a pair) can occur randomly as cells re-type the instruction manual word for word. Any time that there's a typo, symptoms of trisomy 13 occur because your cells don't have the instructions they need to form and function properly.

There are three possible ways for a trisomy to form at chromosome 13 depending on how the chromosomes come together.

Complete trisomy 13

Random copying errors where more genetic material connects to a chromosome than necessary (complete trisomy 13) during the formation of the sperm and egg before conception causes trisomy 13. People with trisomy 13 have three copies of chromosome 13 instead of two. The extra genetic material attached to chromosome 13 causes symptoms of the condition.

Translocation

In about 20% of trisomy 13 cases, symptoms occur when part of chromosome 13 attaches to a nearby chromosome when eggs and sperm form (translocation) during fetal development. In this case, there

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are two pairs of chromosome 13, and an additional copy of chromosome 13 forms and bonds with a nearby chromosome pair, not necessarily in the 13th position.

Mosaic trisomy 13

In rare cases, an extra copy of chromosome 13 appears in some cells in the body but not all cells. This means that some cells in the body have three chromosome 13's and others only have a pair of chromosome 13 (euploid). The severity of symptoms for a mosaic trisomy 13 diagnosis depends on how many cells have the third copy of trisomy 13. Symptoms are more severe if more cells have a third copy.

Diagnosis and Tests

How is trisomy 13 (Patau syndrome) diagnosed?

During the first trimester of pregnancy, your healthcare provider may offer genetic testing in addition to prenatal ultrasounds, with tests as early as 11 to 14 weeks to check for genetic changes, like a third chromosome attached to a pair. Confirmation of the diagnosis occurs after your baby is born, when your healthcare provider can physically examine your baby for symptoms and provide additional tests, if necessary.

References:

1. Disease Ontology (http://purl.obolibrary.org/obo/doid.owl) (англ.) — 2016. Проявления заболевания Лечение См. также Примечания
2. https://web.archive.org/web/20070929094344/http://schools.keldysh.ru/school1413/pro_2005/z/hrbol.htm
3. http://www.medkurs.ru/lecture2k/genetics/gl19/4288.html
4. http://rh-conflict.narod.ru/student/lectures/hrombol.htm