INTERNATIONAL MULTIDISCIPLINARY JOURNAL FOR RESEARCH & DEVELOPMENT
SJIF 2019: 5.222 2020: 5.552 2021: 5.637 2022:5.479 2023:6.563
eISSN 2394-6334 https://www.ijmrd.in/index.php/imjrd Volume 10, issue 10 (2023)

PATAU SYNDROME'S CURRENT OCCURRENCE STATISTICS AND SIMTOMS OF THE DISEASE

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Annotation: Patau's Syndrome is also known as Trisomy 13. It is a chromosome-based, rare genetic disorder in which the patient has an extra copy of chromosome 13 in some or all of the cells in the body. Normally the baby should have two copies of the chromosome but in this case, there are three. The presence of the extra chromosome causes abnormal development of the fetus, often resulting in a miscarriage or stillbirth.

Key words: Patau syndrome, cytogenetic analysis, autosomal trisomies.

Patau syndrome, also called trisomy 13, is the least common and most severe of the viable autosomal trisomies. Median survival is fewer than 3 days. First identified as a cytogenetic syndrome in 1960, Patau syndrome is caused by an extra copy of chromosome 13, a medium-length acrocentric chromosome. Cytogenetic analysis is a necessary step in the prenatal diagnosis of Patau syndrome. Referral to a geneticist or genetic counselor is important for appropriate counseling regarding recurrence risks, etiology, prognosis, and the availability of local area resources for support.

Immediately obtain conventional cytogenetics for any child or neonate with suspected Patau syndrome, unless cytogenetic diagnosis has been made prenatally.
Antecedents to the prenatal diagnosis of Patau syndrome include abnormal first or second trimester aneuploidy screening and abnormal prenatal ultrasonographic findings, including birth defects or growth restriction. In addition to conventional cytogenetics, fluorescent in-situ hybridization (FISH) on interphase cells could be used to obtain a more rapid prenatal diagnosis.
Initiate appropriate imaging studies when holoprosencephaly or cardiac or renal anomalies are clinically suspected.
Because of the high frequency of structural defects in Patau syndrome, perform cardiac evaluations on patients who survive the neonatal period.
Once a diagnosis of Patau syndrome is made, pregnancy management varies according to the gestational age at diagnosis.
At previable gestational ages, the option of pregnancy termination should be among those discussed. The gestational age limits for this procedure are state-specific and subject to the training and skill of the physician available to perform the pregnancy termination.
When patients choose not to proceed with pregnancy termination or when the pregnancy has progressed to a viable gestational age such that pregnancy termination is no longer an option (except in rare locations throughout the United States), attention should be focused on whether the labor should be induced or spontaneous.
Surgical interventions in Patau syndrome are generally withheld for the first few months of life because of the high mortality rates of babies with the condition. Carefully weigh decisions about extraordinary life-prolonging measures against the severity of the neurologic and physical defects that are present and the likelihood of postsurgical recovery or prolonged survival.

The chances of survival of a baby with Patau's syndrome are minimal. One in sixteen thousand are affected by the syndrome. Around 90% of babies born with Trisomy 13 will not survive beyond their first birthday. Many are born with such severe medical conditions that they die within a week after being born. The 10% of babies born with a less severe form of Patau's Syndrome may live longer but will be afflicted with a range of health issues.

Due to the presence of the extra chromosome 13 in the body's cells, the proper development of the baby is impossible. Some possible symptoms of a baby born with this genetic disorder include:

- Congenital heart defects
- Brain and spinal cord abnormalities
- Microcephaly or head size is smaller than normal
- Micrognathia or smaller lower jaw size than normal
- Cutis Aplasia or missing skin on the scalp
- Deformation of ears accompanied with deafness
- Microphthalmia or poorly developed eyes
- Anophthalmia or absence of one or both eyes
- Coloboma or hole, split or cleft in the iris
- Polydactyly or extra toes or fingers may be present
- Nasal passages may not be properly developed
- Cleft lip or an opening in the lip
- Cleft palate of an opening in the roof of the mouth
- Hypotonia or weak muscle tone
- Rounded bottom to the feet also known as rocker-bottom feet
- Capillary Haemangiomas or raised red birthmarks
- Exomphalos or intestines found outside the body in a sac of membrane
- Hernias such as umbilical hernia or inguinal hernia
- Cysts in the Kidneys
- Intellectual disability
- Skeletal abnormalities in the limbs

Not all babies born with Patau's syndrome will display all these symptoms. However, they will have many of the symptoms mentioned in the list leading to medical health issues.

Cause and Diagnosis

The extra copy of chromosome 13 is generated by random events. The formation of the eggs or sperm may show translocation of this chromosome which causes trisomy 13. This is a condition that may be inherited or may occur randomly during conception. There is nothing that the parents can do to avoid this abnormal development. Statistically speaking, the older the woman is at the time of conception, the more likely there is to be a genetic problem with the fetus. This is why women are advised to conceive before they reach the age of thirty. The condition of their eggs is not as robust as they age.

The diagnosis of a genetic disorder is done by screening the pregnant woman at 10 to 14 weeks. The combined test will check for Down's syndrome, Edward's syndrome and Patau's syndrome.

It involves a blood test combined with an ultrasound scan. If the fetus is found to be at risk, additional tests may be done by using a sample of cells from the fetus. People with a family history of trisomy 13 should opt for the tests.

The two ways to get fetal cells are *amniocentesis* and *chorionic villus sampling*. In amniocentesis, a sample of the amniotic fluid is removed from the womb as it tends to contain the cells shed by the developing fetus. In chorionic villus sampling cells will be taken from the placenta which links the blood supply of the mother to the developing fetus. Both tests are highly invasive and include a risk of miscarriage.

Treatment

There is no definitive treatment for Patau's syndrome. As it cannot be cured, the treatment is usually based on the symptoms that ail the baby. The aim of the medical staff at the hospital is to minimize the discomfort of the baby and ensure that it is able to feed. Very often, the baby will be unable to respond to normal stimuli due to its abnormal development.

The care of the baby includes counseling the parents about what to expect. A support system is essential to help the new parents deal with the stark reality of the fact that their baby is not going to live beyond a year if it makes it past the first week in the hospital. It can be an emotionally traumatic experience for the parents.

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