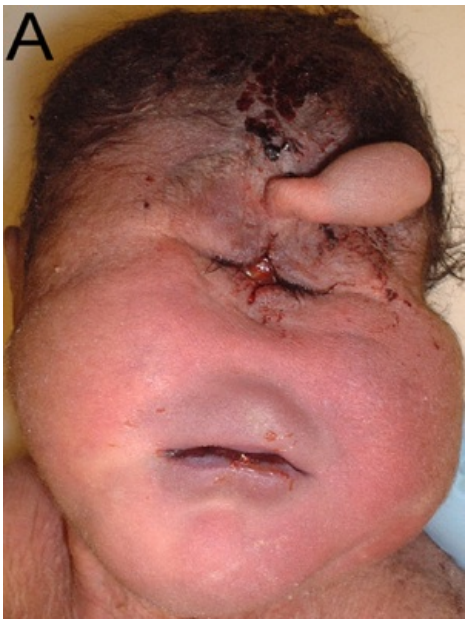
Patau Syndrome

Bartholin-Patau syndrome, also called autosomal Trisomy 13, is a very severe condition first described by *Dr. Klaus Patau* in 1960. This genetic disorder is associated with severe intellectual disability and physical abnormalities in many parts of the body. The affected individuals often have heart defects, brain or spinal cord abnormalities, microphthalmia (very small or poorly developed eyes), cleft lip, extra fingers and/or toes, among other features.



A 37 2/7 week gestational age male infant with Patau syndrome.

Causes

Trisomy 13 is a chromosomal condition in which the sex ratio at birth is slightly higher in females than in males. This fact could be related to the decreased survival rate among male fetuses.

Like most other trisomies (aneuploidy), *Patau syndrome* increasing incidence is associated with advanced maternal age, and the additional chromosome usually arises from nondisjunction in maternal meiosis resulting in three copies of 13 chromosome genetic material. This is a noninheritate case, is the result of random events during meiosis.

The extra genetic material disrupts the normal course of development, causing the characteristic features of *trisomy 13*. However, some cases derived from germinal mosaicism or by balanced chromosomal rearrangements (Robertsonian translocations), where the extra material is attached to another chromosome, are situations in which the trisomy can be inherited.

The person who carries the balanced translocation involving chromosome 13 has an increased chance of passing extra material from chromosome 13 to their children.

File:Patauhand.PNG

The same male infant with Patau syndrome demonstrating polidactyly.

Symptoms

The small percentage of babies with full *Patau's syndrome* and who survived birth, may express the following internal and external conditions:

- Low Apgar scores.
- Hands and feet showing polydactyly (postaxial).
- Facial defects:
    - Mouth: cleft lip and/or palate.
    - Nose: absent, malformed or proboscis (prominent).
    - Ears: malformed ears.
    - Eye: structural eye defects (microphthalmia, iris coloboma or even absence of the eyes).
- Hands: abnormal palm pattern.
- Muscle: Hypotonia (decreased muscle tone).
- Hernias: umbilical hernia, inguinal hernia.
- Severe central nervous system malformation:
    - Severe mental retardation.
    - Cephalic disorder: arhinencephaly and holoprosencephaly.
    - Spinal cord defects.
- Cardiac defects.
- Urogenital defects:
    - Abnormal genitalia (e. g. undescended testicle).
    - Kidney defects.

Links

Related articles

- Turner Syndrome
- Klinefelter Syndrome

Sources

- HALDEMAN-ENGLERT, Chad. *Trisomy 13* [online]. [cit. 2010-10-26]. <http://www.nlm.nih.gov/medlineplus/ency/article/001660.htm>.
- BEST, Roger G. *Patau Syndrome* [online]. [cit. 2010-10-26]. <http://emedicine.medscape.com/article/947706>.