



Patau's syndrome Guide

What is Patau's syndrome

There is no such thing as a typical person with Patau's syndrome. Like all people, they vary a lot in appearance, personality and ability. Patau syndrome, also known as trisomy 13 and trisomy D, is a chromosomal abnormality, a syndrome in which a patient has an additional chromosome 13 due to a nondisjunction of chromosomes during meiosis.

The extra chromosome 13 disrupts the normal course of development, causing heart and kidney defects amongst other features characteristic of Patau syndrome. Like all nondisjunction conditions (such as Down syndrome and Edwards syndrome), the risk of this syndrome in the offspring increases with maternal age at pregnancy, with about 31 years being the average. Patau syndrome affects approximately one in 10,000 live births.

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Most cases of Patau's syndrome result from trisomy 13, which means each cell in the body has three copies of chromosome 13 instead of the usual two copies. A small percentage of cases occur when only some of the body's cells have an extra copy, resulting in a mixed population of cells with a differing number of chromosomes; such cases are called mosaic Patau.

Patau syndrome can also occur when part of chromosome 13 becomes attached to another chromosome (translocated) before or at conception. Affected people have two copies of chromosome 13, plus extra material from chromosome 13 attached to another chromosome. With a translocation, the person has a partial trisomy for chromosome 13 and often the physical signs of the syndrome differ from the typical Patau syndrome.

Most cases of Patau syndrome are not inherited, but occur as random events during the formation of reproductive cells (eggs and sperm). An error in cell division called non-disjunction can result in reproductive cells with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of the chromosome. 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 of the body's cells. Mosaic Patau syndrome is also not inherited. It occurs as a random error during cell division early in fetal development. Patau syndrome due to a translocation can be inherited.

An unaffected person can carry a rearrangement of genetic material between chromosome 13 and another chromosome. This rearrangement is called a balanced translocation because there is no extra material from chromosome 13. Although they do not have signs of Patau syndrome, people who carry this type of balanced translocation are at an increased risk of having children with the condition.

What are the symptoms?

The features and problems that develop in children with Patau's syndrome vary from child to child. Typically, of those fetuses that do survive to gestation and subsequent birth, common abnormalities include:

- Nervous system M
- Mental & motor challenged
- Microcephaly
- Holoprosencephaly (failure of the forebrain to divide properly)
- Structural eye defects, including microphthalmia, Peters anomaly, cataract, iris and/or fundus (coloboma), retinal dysplasia or retinal detachment, sensory nystagmus, cortical visual loss, and optic nerve hypoplasia
- Meningomyelocele (a spinal defect)
- Musculoskeletal and cutaneous
- Polydactyly (extra digits) and overlapping of fingers over thumb
- Low-set ears
- Prominent heel
- Deformed feet known as rocker-bottom feet
- Omphalocele (abdominal defect)
- Abnormal palm pattern
- Cutis aplasia (missing portion of the skin/hair)
- Cleft palate
- Urogenital
- Abnormal genitalia
- Kidney defects
- And possible heart defects (ventricular septal defect) and single umbilical artery.

Screening for Patau's Syndrome

If you are looking for information on pre-natal testing, we recommend a Nuchal Translucency scan. A nuchal scan measures the amount of fluid at the back of the baby's neck. Together with a blood test a nuchal scan can estimate the risk of your baby having a chromosomal abnormality such as Patau's syndrome.

A nuchal scan is a non-invasive test (procedure which does not require incision into the body) that can only be performed between 11 weeks +4 days to 13 weeks +6 days of pregnancy. The blood test measures two concentrations BHCG (hormone) and PAPP-A (protein). Research suggests BHCG levels are higher and PAPP-A levels are lower in cases of Patau's syndrome. A nuchal scan is not however a diagnostic test, it just gives a risk factor. Some private clinics in the UK

A number of Baby Premier clinics offer OSCAR (one stop clinic for assessment of fetal risk) for Patau's syndrome screening. This means that the nuchal scan, blood test and result are done during one visit. The entire process usually takes around 2 – 2 1/2 hours. Other clinics provide a similar service but on a two-stop appointment system.

What's the treatment?

Medical management of children with Trisomy 13 is planned on a case-by-case basis and depends on the individual circumstances of the patient. Treatment of Patau syndrome focuses on the particular physical problems with which each child is born. Many infants have difficulty surviving the first few days or weeks due to severe neurological problems or complex heart defects. Surgery may be necessary to repair heart defects or cleft lip and cleft palate. Physical, occupational, and speech therapy will help individuals with Patau's syndrome reach their full developmental potential.

NICE recommendations

The National Institute for Clinical Excellence (NICE) have recommended nuchal scanning combined with blood test during the first trimester of pregnancy to be the most effective non-invasive screening test.

In the future NICE would like all women to be offered first trimester screening for Patau's syndrome so that mums-to-be do not have to go through with unnecessary invasive tests. Unfortunately nuchal screening is not widely available at most NHS antenatal clinics and some clinics do not offer the combined blood test.

Advantages of nuchal screening:

- No risk to you and your baby
- No waiting lists
- Immediate results and advice
- Avoid unnecessary worry
- Peace of mind
- Time to make an informed choice

How can I get more information on Nuchal Testing for Down's syndrome?

Choosing whether to have a nuchal scan the tests is an important decision, for you and for your baby. If you are faced with this decision, you need to make sure you reach the right decision for you.

For more information on nuchal screening or any obstetric medical ultrasound scans please contact the Baby Premier advice and bookings clinic on **0845 345 7262** or email **info@babypremier.co.uk**

Baby Premier is the obstetric and gynaecological division of SMI (Specialist Medical Imaging Ltd), a medical ultrasound company established in 2004. Baby Premier is an accredited ultrasound service that offers a full range of obstetric and gynaecological ultrasound examinations to both self-funding and privately insured patients.