



Edward's syndrome Guide

What is Edward's syndrome

There is no such thing as a typical person with Edward's syndrome. Like all people, they vary a lot in appearance, personality and ability. Edward's syndrome (also known as Trisomy 18) is a genetic disorder caused by the presence of all or part of an extra 18th chromosome. It is named after John H. Edwards, who first described the syndrome in 1960 and is the second most common autosomal trisomy, after Down syndrome, that carries to term.

Edwards syndrome is caused by the presence of three—as opposed to two—copies of chromosome 18 in a fetus or infant's cells. The incidence of the syndrome is estimated as one in 3,000 live births. The incidence increases as the mother's age increases. The syndrome has a very low rate of survival, resulting from heart abnormalities, kidney malformations, and other internal organ disorders.

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Annually in England and Wales, there are around 495 diagnoses of Edwards' syndrome, of which 92% were made prenatally. There were 339 terminations, 49 stillbirths/miscarriages/fetal deaths, 72 unknown outcomes, and 35 live births. Because approximately 3% of cases of Edwards' syndrome with unknown outcomes are likely to result in a live birth, the total number of live births is estimated to be 37 (2008/09 data are provisional).

Only 50% of liveborn infants live to 2 months, and only 5–10% survive their first year of life. Major causes of death include apnea and heart abnormalities. It is impossible to predict the exact prognosis of a child with Edwards syndrome during pregnancy or the neonatal period. The median lifespan is 5–15 days. One percent of children born with this syndrome live to age 10, typically in less severe cases of the mosaic Edwards syndrome.

Edwards syndrome occurs in approximately 1 in 3,000 conceptions and approximately 1 in 6,000 live births; 50% of those diagnosed with the condition prenatally will not survive the prenatal period. Although women in their 20s and early 30s may conceive babies with Edwards syndrome, the risk of conceiving a child with Edwards syndrome increases with a woman's age. The average maternal age for conceiving a child with this disorder is 32½.

What are the symptoms?

The features and problems that develop in children with Edwards' syndrome vary from child to child. Typically, a child will have a small head with characteristic facial features including a small jaw and mouth, upturned nose, widely spaced small eyes with narrow eyelid folds and drooping of the upper eyelids, and low-set, malformed ears.

The hands may be clenched, with the second and fifth fingers overlapping the other fingers, and the thumbs may be underdeveloped or absent. Webbing of the second and third toes may also occur. In addition to these features, all systems of the body may be affected. Structural malformations of the heart, kidneys, brain, digestive tract and genitals may be present and cause the child difficulties. For example, children with the syndrome often have trouble feeding and breathing, and experience delay in growth and development. Infections of the lungs and urinary system are also common.

Health and development for people with Edward's syndrome

Infants born with Edwards syndrome may have some or all of the following characteristics: kidney malformations, structural heart defects at birth (i.e., ventricular septal defect, atrial septal defect, patent ductus arteriosus), intestines protruding outside the body (omphalocele), esophageal atresia, mental retardation, developmental delays, growth deficiency, feeding difficulties, breathing difficulties, and arthrogyposis (a muscle disorder that causes multiple joint contractures at birth).

Some physical malformations associated with Edward's syndrome include small head (microcephaly) accompanied by a prominent back portion of the head (occiput); low-set, malformed ears; abnormally small jaw (micrognathia); cleft lip/cleft palate; upturned nose; narrow eyelid folds (palpebral fissures); widely spaced eyes (ocular hypertelorism); drooping of the upper eyelids (ptosis); a short breast bone; clenched hands; underdeveloped thumbs and or nails absent radius, webbing of the second and third toes; clubfoot or Rocker bottom feet; and in males, undescended testicles.

In utero, the most common characteristic is cardiac anomalies, followed by central nervous system anomalies such as head shape abnormalities. The most common intracranial anomaly is the presence of choroid plexus cysts, which is a pocket of fluid on the brain that is not problematic in itself but may be a marker for Trisomy 18. Sometimes excess amniotic fluid or polyhydramnios is exhibited.

Screening for Edward'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 Edwards'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 Edward'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 Edward'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.

How are risk factors evaluated?

Edwards' syndrome occurs in around one in 6,000 live births and around 80 per cent of those affected are female. However, the majority of babies with the syndrome die before birth. It affects people from all cultural backgrounds and becomes more likely with increasing maternal age.

What's the treatment?

There's no cure for Edwards' syndrome, but medical treatment of symptoms is provided as required. Treatment focuses on providing good nutrition, tackling infections - which arise frequently - and helping the heart to function better.

Many babies with Edwards' syndrome have difficulties with feeding, so food may be given via a nasogastric tube or directly into the stomach through a gastrostomy. Where limb abnormalities affect movement, physiotherapy and occupational therapy can help. Emotional support for parents and other members of the family is vital, as babies with Edwards' syndrome have a shortened life expectancy. Few survive beyond their first year.

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 Edward'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 Edward'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.