

VACTERL - an overview

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What is VACTERL?

The term 'VACTERL' describes a group of anomalies which often occur together in newborn babies. It is an acronym for:

- Vertebral (spinal) defects
- Anorectal atresia (failure of the anus and lower end of the gut to form)
- Cardiac (heart) defects
- Tracheo-oesophageal fistula with or without Esophageal atresia (American spelling of 'oesophageal')
- Renal (kidney) anomalies
- Limb defects.

VACTERL was once simply 'VATER' but the longer term is now preferred, since it includes cardiac defects – which over 70% of these children have – and acknowledges that the limb problems are not only in the radius bone of the forearm. 'VATER' was originally described in the mid 1970's, so children born before this date may not have been screened for these problems. Other synonyms for VACTERL are Kaufman syndrome, PIV and PIAVA.

What is a VACTERL child?

To qualify as a 'VACTERL' child, three of the seven components mentioned above must be present. There may also be other characteristics which occur more frequently in affected children than the rest of the population; these include ear abnormalities, genital anomalies, cleft lip and/or palate, thumb abnormalities, various changes in gut development and in the fetus, and the presence of a single artery in the umbilical cord (normally there are two).

No one baby is likely to display all these features and no two individuals are likely to be affected in exactly the same way.

What causes VACTERL?

For every 6,250 births, one child will have VACTERL, making it relatively rare.

Large numbers of patients are required to scientifically study any medical condition, so the small numbers of children with VACTERL, and their wide variety of features, has made research difficult. Consequently, little is known.

Growth of the fetus in the womb is enormously complex; in spite of the massive scientific efforts aimed at understanding the intricate mechanisms involved, many pieces of the jigsaw are still missing.

The cause of VACTERL is unclear. One theory suggests that cells are disrupted at an early stage of development. There are only three types of cell in the three-week-old fetus, one of which is called a mesodermal cell. These cells go on to form the gut wall (including the oesophagus), the kidneys and bone (including the spine and the skeleton of the limbs). In principle, a change in a mesodermal cell could result in a change in any of these body parts, which would help explain the wide variety of abnormalities present in VACTERL.

Efforts have been made to identify agents, such as drugs, which could adversely affect these cells, but so far nothing has been proved.

Chromosomes are the inherited structures of DNA which carry the genes to determine everything about an individual. Two chromosomal abnormalities (the more common of which is Edwards syndrome) may result in the features of VACTERL – two rare cases in which the cause of VACTERL is known. In order to exclude this, the chromosomes of all VACTERL babies are examined.

Can VACTERL be detected before birth?

The regular use of ultrasound to examine the fetus is greatly increasing the number of abnormalities detected before birth.

Many VACTERL babies will not be picked up during a routine scan, but in pregnancies which merit detailed ultrasound investigation it is more likely that skeletal abnormalities, kidney defects and a single umbilical artery will be seen.

Perhaps, as ultra-sound scanning continues to improve, ante-natal diagnosis of VACTERL will become the norm.

This information has been written for the parents of TOF children by TOFS (Tracheo-Oesophageal Fistula Support) – helping children born unable to swallow.

If you have any feedback on this leaflet, please use our leaflets feedback form which is available from either the TOFS office or our web site.

TOFS relies on money from membership fees, voluntary donations and other sources of charitable income to fund its activities.

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TOFS does not offer specific medical advice to parents. We work only in a supportive role, offering emotional and practical support to meet the needs of parents and providing a source of information which complements that given by the specialist hospital.

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Related leaflets from TOFS which you might like to read:

1. Conditions occurring with TOF/OA
2. VACTERL - vertebral anomalies
3. VACTERL - anal anomalies
4. VACTERL - cardiac anomalies
5. VACTERL - renal anomalies
6. VACTERL - limb anomalies

These are all available from the TOFS web site (www.tofs.org.uk) or from TOFS office.

TOFS also publishes a book, 'The TOF Child,' which is suitable for both parents and medical professionals. Details are available from TOFS.

What are the exact problems in VACTERL?

VERTEBRAL ABNORMALITIES

Vertebrae are flat bones which are stacked on top of each other to form the spine. If they are deformed, an abnormal spinal shape may result ('scoliosis' or 'kyphosis'). Abnormal bone development may also be accompanied by abnormalities in the associated muscles and nerves; treatment depends on the severity of the deformity.

ANORECTAL ATRESIA

This is a broad term which describes a range of abnormalities. At one extreme, children have an intact bowel with a blind end ('low imperforate anus'). At the other extreme, the bowel stops quite some distance short of what should be the anal opening, and there are often abnormal connections between the bowel and the bladder or vagina.

CARDIAC ABNORMALITIES

'Ventricular septal defect' (VSD) is the commonest type of cardiac abnormality, accounting for more than three-quarters of all cases. It is a hole in the wall that separates the two large chambers of the heart. Consequently, the heart does not function efficiently. The child is symptom-free if the hole is small, but in more severe cases children can become breathless and fail to thrive. The defect may close spontaneously but sometimes surgery is required.

There are many other heart defects which may occur in isolation or accompany a VSD.

TRACHEO-OESOPHAGEAL FISTULA WITH OESOPHAGEAL ATRESIA

VACTERL children are classed as TOFs because this is a feature of the condition, but their problems and treatments differ due to their multiple abnormalities.

RENAL ANOMALIES

These fall into two main categories; total failure of one or both kidneys to form, and 'other' anomalies.

Absence of both kidneys has sinister implications and may be detected antenatally by ultrasound. A single absent kidney is however entirely symptomless, because the remaining kidney can fully compensate for the deficiency.

The 'other' category is vast, including a large number of different problems. These are not always associated with malfunction, but problems may arise if the kidney cells do not work efficiently or if kidney infections result.

LIMB ABNORMALITIES

The forearm (the part of the arm between the elbow and wrist) contains two bones, the ulna and radius. Partial or total failure of the radius (and the muscles which attach to it) to develop is frequently seen in VACTERL children. Characteristically this causes the hand to lie at a right-angle to the forearm; usually the thumb is also malformed or absent. Splints, or even surgery, may be required to correct the defect.

Limb abnormalities are not always restricted to the forearm – the feet or legs can also be affected.

The VACTERL Child

Hospitals are a second home for VACTERL children. They receive intensive medical attention, both to treat the immediate problems and to monitor their progress. This involves frequent consultations with a variety of specialist doctors, in addition to intensive general health surveillance, such as regular growth checks.

The impact of such prolonged hospitalisation should not be underestimated, although VACTERL children have comparable intelligence to their peers.

Ultimately, the future of an affected child will be determined by the nature of their initial problems and the success of their treatment. Residual problems may cause long-term disability, the severity of which will vary between individuals and depends on the number and type of defects.

In the majority of cases, continued medical support will be required, but this does not prevent a VACTERL child from leading an active, independent and fulfilled life.

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