



# VACTERL association: information for families

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In medical terms, an 'association' is a group of symptoms that appear together more often than would be expected by chance. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of VACTERL association (also known as VATER association) and where to get help.

VACTERL is an acronym made up of the first letters of the main symptoms that make up the condition:

- Vertebral defects
- Anorectal anomalies
- Cardiac defects
- Tracheo-oesophageal fistula/oesophageal atresia
- Renal abnormalities
- Limb abnormalities

Some children may be described as having VATER association instead of VACTERL association if they do not have any cardiac defects or limb abnormalities.

## What causes VACTERL association?

We do not know exactly what causes VACTERL association. It is clear that all the symptoms happen very early in pregnancy but there is no clear cause, genetic or environmental. It does not seem to have a genetic (inherited) component as most children born with VACTERL association do not have any close relatives with similar symptoms. According to published literature, it affects all races and males and females in equal numbers, affecting 1 in 10,000 to 40,000 births.

## What are the symptoms of VACTERL association?

As described earlier, VACTERL is an acronym made up of the first letters of the main symptoms that make up the condition.

**Vertebral defects** – these affect between 60 and 80 per cent of babies born with VACTERL association to varying degrees. The vertebrae (bones that make up the spine) may be misshapen or fused or missing or there may be additional vertebrae in place. These vertebral problems do not usually cause any problems immediately after birth.

**Anorectal anomalies** – this means that the anus has not formed properly and may be either closed over or very narrow so faeces cannot be passed from the body in the usual way. This affects between 60 and 90 per cent of babies born with VACTERL association. Babies will not be able to pass meconium – the dark faeces passed in the first few days of life – in the usual way. This can cause a swollen abdomen and vomiting. If there is an abnormal passage joining the anus to the vagina or urethra, the faeces will be able to partially pass out of the body, near the vagina in girls or through the urethra in boys. As the faeces can only be partially removed from the body, this

will not relieve the pressure in the abdomen, causing swelling and vomiting.

**Cardiac defects** – Between 40 and 80 per cent of babies with VACTERL association will have a cardiac or heart defect, although the severity of the defect varies from child to child. Ventricular septal defects (a 'hole in the heart') are most commonly seen in VACTERL association, as well as a heart defect called Tetralogy of Fallot.

#### **Tracheo-oesophageal fistula/oesophageal atresia**

– These affect between 50 and 80 per cent of babies born with VACTERL association.

Tracheoesophageal fistula (TOF) is where part of the oesophagus is joined to the trachea (windpipe). Oesophageal atresia (OA) is where a short section at the top of the oesophagus (gullet or foodpipe) has not formed properly so is not connected to the stomach. This means food cannot pass from the throat to the stomach.

**Renal abnormalities** – These abnormalities can vary in severity and may include an absent kidney or kidneys that are formed abnormally such as a horseshoe kidney or cystic kidneys. Around 50 to 80 per cent of babies born with VACTERL association have some form of kidney problem, although this may not cause any problems immediately after birth.

**Limb abnormalities** – This affects around 50 per cent of babies born with VACTERL association. It is often the thumb or the forearm and hand that are affected, which are missing or underdeveloped.

Other abnormalities have been reported in babies born with VACTERL association – all babies are given a thorough physical examination to check for any other problems.

## **How is VACTERL association diagnosed?**

VACTERL association can be diagnosed before birth (prenatally) but this tends only to happen if any of the problems are visible on ultrasound.

More commonly, VACTERL association is diagnosed after a baby has been transferred to a specialist unit for treatment of the tracheo-oesophageal fistula/oesophageal atresia, anorectal anomalies or cardiac defects.

Most doctors believe that a baby has to have three of the symptoms listed above to be diagnosed with VACTERL association. If doctors suspect that your child has VACTERL association, they will carry out a full examination and order further imaging scans to confirm or rule this out.

## **How is VACTERL association treated?**

Babies born with VACTERL association will usually have been transferred to a specialist centre soon after birth due to the immediate problems caused by the tracheo-oesophageal fistula/oesophageal atresia, anorectal anomalies or cardiac defects. The initial aim is to stabilise the baby with intravenous fluids as feeding may be difficult or impossible. If an anorectal anomaly is present, the surgeons will usually create a loop stoma (an artificial way of disposing of waste matter) usually in the days after birth. The tracheo-oesophageal fistula/oesophageal atresia will also be corrected soon after admission along with any serious cardiac defects.

Once the immediate symptoms have been corrected, babies will usually be discharged home to grow and develop. All babies born with VACTERL association will require regular monitoring on a long term basis – both to check that the operations carried out soon after birth have been successful but also to monitor any other symptoms that might cause problems in the future. This monitoring will usually continue until adulthood.

## **What is the outlook for children with VACTERL association?**

The outlook for children born with VACTERL association is variable, depending on the symptoms present, although it is improving all the

time as surgery and long term monitoring improves.

Children with VACTERL association rarely have learning disabilities so most will grow up to attend mainstream school.

## **Further information and support**

The VACTERL Association Support Group is an organisation offering support and advice to anyone affected by VACTERL/VATER association. Visit their website at [www.vacterl-association.org.uk](http://www.vacterl-association.org.uk) or Facebook page at [www.facebook.com/VacterlAssociationSupportGroup](https://www.facebook.com/VacterlAssociationSupportGroup)

TOFS – Tracheo Oesophageal Fistula Support – supports families of children born with tracheo-oesophageal fistula. Call them on 0115 961 3092 or visit their website at [www.tofs.org.uk](http://www.tofs.org.uk)