What is a VACTERL?

VACTERL association is a disorder that affects many body systems. VACTERL is an acronym for the first letters of the main symptoms that make up the condition. At least three VACTERL features need to be present for the diagnosis to be considered.

Vertebral defects

These affect 60 – 80% of infants born with VACTERL association to differing degrees. The vertebrae (the bony part of the spine) may be fused or have abnormal shape or there may be an additional or reduced number of vertebrae.

Anorectal anomalies

This is when the anus (back passage opening) has not developed normally and may either be absent or very narrow so faecal matter cannot be passed from the body in the usual way. This affects between 60 - 90% of infants born with VACTERL association. It can be detected on an ultrasound scan prenatally with the presence of swelling of the baby's lower abdomen and the absence of the back passage sphincter.

Cardiac defects

Between 40 - 80% of infants with VACTERL association will have a heart defect, although the severity of the defect can vary. The most commonly seen heart defects are ventricular septal defects (a ‘hole in the heart’) and a group of heart abnormalities called Tetralogy of Fallot.

Tracheoesophageal fistula/esophageal atresia

These affect between 50 - 80% of infants born with VACTERL association. Tracheo-esophageal fistula (TEF) is where there is an abnormal connection between the esophagus and the trachea (windpipe). Esophageal atresia (EA) is where there is a narrowing of the esophagus so food cannot pass from the throat to the stomach.

Renal abnormalities

These abnormalities can differ in severity and may include absent kidney(s) or other forms of abnormal kidneys such as a horseshoe kidney or cystic kidneys. Around 50 - 80% of infants born with VACTERL association have some degree of kidney problem.
Limb abnormalities

This affects around 50% of infants born with VACTERL association. It often affects the thumb, the forearm, or hand, which may be missing or underdeveloped.

Other abnormalities have been reported in infants with VACTERL and it is important that the fetus is scanned thoroughly in the prenatal period. All infants are also given a comprehensive physical examination after they are born to check for any other problems that might not have been obvious prenatally.

How does VACTERL happen?

It is not known precisely what causes VACTERL association. It does not appear to have a genetic origin as nearly all infants born with VACTERL association do not have any family history of the same symptoms/condition. It affects all races and genders equally, affecting 1 in 10,000 to 40,000 births.

Should I have more tests done?

Many women will choose to have more tests done to know more about the condition of the baby. The tests are available depending on where you are. Tests to ask about include:

- An amniocentesis to look for problems with the number of chromosomes and some of the problems within the chromosomes. This is done by removing a small amount of amniotic fluid surrounding the fetus. A more detailed genetic test called prenatal exome sequencing may be offered to you after discussion with the clinical geneticist. This is to exclude any other genetic conditions that present similarly to VACTERL association.

- A fetal echocardiography is a specialised ultrasound of the baby’s heart during the pregnancy, during the pregnancy, which uses sound waves that “echo” off of the structures of the baby’s heart.

- A detailed ultrasound scan - to monitor any worsening of the abnormality and look for any other abnormality that may become apparent as the baby grows and gets more mature.
What are the things to watch for during the pregnancy?

Babies with VACTERL association are at risk of problems during pregnancy. Babies with tracheoesophageal fistula or esophageal atresia may accumulate extra amniotic fluid around the baby. This condition is called polyhydramnios. It can stretch the uterus too much and cause early labor well before the due date. Knowing this in advance, your doctor can help with decreasing the risk of an early birth.

What does it mean for my baby after it is born?

The prognosis of VACTERL is determined by the number, severity, and nature of abnormalities present. Your baby will likely need corrective surgeries, and this will be discussed further with the paediatric surgeon before or after your baby is born.

Will it happen again?

If no genetic reason is found (which is likely to be the case in VACTERL), the risk of this happening again is very low.

What other questions should I ask?

- Is VACTERL associated with other abnormalities?
- Should I do a genetic test?
- How often will I have ultrasound examinations done?
- Is surgery during the pregnancy available?
- Is my baby one that could benefit from surgery inside the womb?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

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