VACTERL Association

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genetic conditions

1. Introduction

VACTERL association is a disorder that affects many body systems. VACTERL stands for vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities. People diagnosed with VACTERL association typically have at least three of these characteristic features. Affected individuals may have additional abnormalities that are not among the characteristic features of VACTERL association.

Defects in the bones of the spine (vertebrae) are present in 60 to 80 percent of people with VACTERL association. These defects may include misshapen vertebrae, fused vertebrae, and missing or extra vertebrae. In some people, spinal problems require surgery or cause health problems, such as back pain of varying severity, throughout life. Sixty to 90 percent of individuals with VACTERL association have narrowing or blockage of the anus (anal atresia). Anal atresia may be accompanied by abnormalities of the genitalia and urinary tract (genitourinary anomalies). Heart (cardiac) defects occur in 40 to 80 percent of individuals with VACTERL association. Cardiac defects can range in severity from a life-threatening problem to a subtle defect that does not cause health problems. Fifty to 80 percent of people with VACTERL association have a tracheo-esophageal fistula, which is an abnormal connection (fistula) between the esophagus and the windpipe (trachea). Tracheo-esophageal fistula can cause problems with breathing and feeding early in life and typically requires surgical correction in infancy. Kidney (renal) anomalies occur in 50 to 80 percent of individuals with VACTERL association. Affected individuals may be missing one or both kidneys or have abnormally developed or misshapen kidneys, which can affect kidney function. Limb abnormalities are seen in 40 to 50 percent of people with VACTERL association. These abnormalities most commonly include poorly developed or missing thumbs or underdeveloped forearms and hands.

Some of the features of VACTERL association can be subtle and are not identified until late in childhood or adulthood, making diagnosis of this condition difficult.

2. Frequency

VACTERL association occurs in 1 in 10,000 to 40,000 newborns.

3. Causes

VACTERL association is a complex condition that may have different causes in different people. In some people, the condition is likely caused by the interaction of multiple genetic and environmental factors. Some possible genetic and environmental influences have been identified and are being studied.

The developmental abnormalities characteristic of VACTERL association develop before birth. The disruption to fetal development that causes VACTERL association likely occurs early in development, resulting in birth defects that affect multiple body systems. It is unclear why the features characteristic of VACTERL association group together in affected individuals.

4. Inheritance

Most cases of VACTERL association are sporadic, which means they occur in people with no history of the condition in their family. Rarely, families have multiple people affected with VACTERL association. A few affected individuals have family members with one or two features, but not enough signs to be diagnosed with the condition. In these families, the features of VACTERL association often do not have a clear pattern of inheritance. Multiple genetic and environmental factors likely play a part in determining the risk of developing this condition and how severe the condition will be in an individual.

5. Other Names for This Condition

- VATER association

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