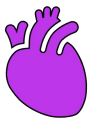
VACTERL Association

A group of anomalies that occur in the newborn
- Approximately **1 in 10,000 to 40,000 births**
- The cause of VACTERL association is **unknown**
- The mode of inheritance is **sporadic**
- Greater frequency in **infants of diabetic mothers**

DIAGNOSIS
- VACTERL is a **clinical diagnosis**
- **≥ 3 features** required for diagnosis
- **Diagnosis of exclusion** – other possible causes of birth defects need to be ruled out

Vertebral defects
- Present in 60-80% of cases
- Includes hemivertebrae, hypoplastic vertebrae, extra or missing vertebrae, congenital scoliosis, spina bifida
- Can cause back pain later in life

Anal atresia
- Present in 60-90% of cases
- Narrowing or lack of patency of the anus
- May be accompanied by genitourinary abnormalities
- No passage of meconium, abdominal distension, vomiting



Cardiac defects
- Present in 40-80% of cases
- Subtle to life threatening defects
- Common defects: ventricular septal defect, atrial septal defect, tetralogy of fallot, patent ductus arteriosus

Tracheo-esophageal fistula
Esophageal atresia
- Present in 50-80% of cases
- TEF is an abnormal connection between the trachea and the esophagus
- EA is a congenital condition characterized by the incomplete development of the esophagus
- TEFs are most often found in conjunction with EA
- May present with vomiting, copious oral secretions, feed intolerance, coughing, cyanosis, or respiratory distress

Renal anomalies
- Present in 50-80% of cases
- Renal aplasia, renal dysplasia, vesicoureteral reflux, displaced or mal-positioned kidneys
- May not cause problems immediately
- Nephrology and urology consultation may be necessary

Limb abnormalities
- Present in 50% of cases
- Radial aplasia – underdevelopment of the radius
- Polydactyly (additional digits), missing digits, lower limb hypoplasia, syndactyly (webbing)

A **syndrome** involves a collection of signs and symptoms that consistently occur together due to an underlying cause, while an **association** refers to a statistical tendency for certain features to co-occur without an underlying cause.

MANAGEMENT
- Treatment is directed toward the **specific malformations and related symptoms**, which varies greatly.
- Many of the structural abnormalities (heart defects, anal atresia, TE fistula, radial defects, etc.) can be **surgically repaired**. Some surgeries occur in the neonatal period, while others occur later in life.
- Repeat surgeries might also be needed to further correct structural defects.

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