VACTERL (VATER) Association

Genetics

-Unknown, eventually: FGF8, FOXF1, HOXD13, LPP, TRAP1, ZIC3

-Sporadic

Clinical findings/Dysmorphic features

-Vertebral defects (V), 70%

-Anorectal malformations/Anal atresia (A), 33%

-Cardiac defects (C), 75%: VSD, PDA, TOF

-Tracheoesophageal fistula with or without esophageal atresia (TE), 70%

-Renal malformations (R), 50%

-Limb (L), 70%: polydactyly, humeral hypoplasia, radial aplasia, proximally placed thumb

Etiology

-Frequency: 1/10,000 to 1/40,000

Pathogenesis

-unknown

Genetic testing/diagnosis

-Diagnosis requires 3 of 7 features and it is a diagnosis of exclusion --> rule out aneuploidy with karyotype, Fanconi anemia with DEB testing, and Townes- Brocks syndrome by SALL1 seq

Others

-A variant is VACTERL with hydrocephalus which can be AR or XL