Trisomy 18 (Edwards syndrome)

Genetics

-Full (94%), mosaic (<5%), or partial (~2%) trisomy of chromosome 18q

Clinical findings/Dysmorphic features

-Clenched hands: 2 over 3 and 5 over 4

-IUGR, ID

-Rocker bottom feet, small fingernails, short sternum

-Craniofacial disproportion: micrognathia, prominent occiput, microphthalmia (small eye/eyes) -CHD: VSD, ASD, PDA (patent ductus arteriosus); multiple dysplastic valves

-Generalized muscle spasm; renal anomalies

Etiology

-Second most common autosomal trisomy; 1/6000 liveborn infants; 60% female

-95% spontaneously abort; 50% die in first week, 90% die by one year

Pathogenesis

-Less than 1% due to a translocation

-Maternal nondysjunction (97%), mosaicism (10%)

Genetic testing/diagnosis

-Echo, abdominal US; maternal serum screen: low AFP, hCG, and UE3

Others

-Causes of death: central apnea, cardiac failure, respiratory insufficiency

-Maternal age effect