Trisomy 13 (Patau syndrome)

Genetics

-Full/mosaic/partial trisomy of chromosome 13

Clinical findings/Dysmorphic features

-Sloped forehead, malformed ears, cleft lip and palate

-Eye anomalies: microphthalmia, iris coloboma, hypotelorism

-CNS: Holoprosencephaly, microcephaly, severe ID, meningomyelocele (type of spina bifida), agenesis of the corpus callosum, enlarged cisterna magna (opening to cerebellum)

-CHD: ventral septal defect, hypoplastic left heart, double-outlet right ventricle

-Renal abnormalities: hydronephrosis, polycystic kidneys, hydroureter

-Genitalia abnormalities: Male --> cryptorchidism, hypospadias, anomalous scrotum; Female --> bicornuate uterus, duplicated system

-Postaxial polydactyly; IUGR; cutis aplasia; seizures, HL; omphalocele (abdominal organs are outside of the body)

Etiology

-Least common of the live born trisomy disorders (1/15,000-25,000 liveborn infants)

-44% die in the first month, >70% die within one year

Pathogenesis

-75% are due to maternal nondysjunction, 20% to a translocation, and 5% to mosaicism

Genetic testing/diagnosis

-Karyotype is diagnostic

Others

-Mosaic T13 with broad phenotype (full T13 - mild ID/physical features and longer survival)