Triploidy

Genetics

-69,XXY > 69,XXX (69,XYY very rare)

-sporadic, no increased risk of recurrence

Clinical findings/Dysmorphic features

-Dysplastic calvaria (skullcap) with large posterior fontanelle (incomplete skull ossification), classic 3/4 finger syndactyly, ASD, VSD, hydrocephalus, holoprosencephaly

Etiology

-The frequency of triploidy in live births is 1:10,000; males represent 51-69 % of all cases

-More than 99% lost in first trimester; accounts for 6-10% of all spontaneous abortions and ~20% of all chromosomally abnormal spontaneous abortions

Pathogenesis

-85% are diandric (2 paternal, 1 maternal) --> well grown fetus, slightly smaller head size, large placenta (partial mole), usually do not survive to term; 0.5% risk of gestational trophoblastic disease (abnormal growth of cells inside a woman's uterus), 0.1% risk of choriocarcinoma

-15% are digynic (2 maternal, 1 paternal): growth retarded fetus with macrocephaly, small & fibrotic placenta, can survive to birth

-Complete mole (diploid – all paternal): 15% risk of gestational trophoblastic disease; 3% risk of choriocarcinoma

-Ovarian teratomas arise through duplication of egg genome (contain all germ layer)

Genetic testing/diagnosis

-Prenatal US, maternal serum hCG low

-Karyotype