Cri-du-Chat (5p minus syndrome)

Genetics

-Partial or complete deletion of chromosome 5p; deletion 5p from band 5p15.2 to 5pter

-12% due to unequal segregation of a translocation or recombination involving a pericentric inversion in one of the parents

-85% sporadic de novo deletions (80% are on the paternal chromosome)

Clinical findings/Dysmorphic features

-Cat-like cry (abnormal laryngeal development); slow growth; ID; hypotonia; strabismus

-Facial features: microcephaly; round face; hypertelorism; micrognathia; epicanthal folds; low‐set ears; broad nasal bridge; short philtrum

Etiology

-Incidence ranges from 1:15,000 to 1:50,000 live-born infants; slight female predominance

Pathogenesis

-Loss of CTNND2 is associated with severe ID

Genetic testing/diagnosis

-Most on karyotype, few are submicroscopic and diagnosed by FISH

Others

-Cat-like cry only when deletion limited to band 5p15.32

-Study of deletions from 5p15.2 to 5p13 found no correlation with size and degree of ID