Pallister-Killian mosaic syndrome

Genetics

-Isochromosome 12p or i(12p)

-100% de novo

Clinical findings/Dysmorphic features

-Hypotonia in infancy and early childhood; ID

-Sparse scalp hair; high forehead; coarse face; widely spaced eyes; broad nasal bridge; highly arched palate; epicanthal fold; large, low-set ears with thick and outwards-protrude lobes

-Hypopigmented streaks of skin; extra nipples; seizures; droopy upper eyelids, crossed eyes (strabismus); joint contractures; cognitive delays; heart defects; rounded cheeks; wide mouth with thin upper lip and a large tongue

Etiology

-150 cases reported

Pathogenesis

-Some cells with isochromosome 12p --> four copies of all genes on p arm of chromosome 12

Genetic testing/diagnosis

-Karyotype/FISH