Aarskog syndrome

Genetics

-Gene: FGD1 (Rho/Rac guanine nucleotide exchange factor; Xp11.22)

-XLR (some cases AR or AD)

Clinical findings/Dysmorphic features

-Shawl scrotum (scrotum surrounds penis); cryptorchidism; brachydactyly (short fingers); short stature; cervical vertebral abnormalities; ID in 30%

-High anterior hairline, frontal bossing, hypertelorism, anteverted nares

-Milder manifestations in females: hypertelorism, short stature, widow's peak hairline

Etiology

-Not known

Pathogenesis

-Unclear: FGD1/Rho GTPase Cdc42 implicated in cytoskeletal organization, potentially in skeletal formation and morphogenesis

Genetic testing/diagnosis

-FGD1 sequencing (20%)

Others

-Orchiopexy (surgery to move undescended testicle into scrotum and permanently fix it there)