Aarskog Syndrome

National Cancer Institute

Source

National Cancer Institute. Aarskog Syndrome. NCI Thesaurus. Code C129720.

An X-linked condition associated in a subset of cases with mutation(s) in the FGD1 gene, encoding a complex signaling protein containing FYVE, RhoGEF, and PH domains. The condition is usually characterized by distinctive facial features, short stature, skeletal anomalies, shawl scrotum (altered anatomical relationship between the penis and the scrotum) cryptorchidism, and developmental delay.