

Facio-Digito-Genital Dysplasia Syndrome (Aarskog Syndrome)

Main Features

- Short stature
- brachydactyly
- shawl scrotum

Eye Findings

- Hypertelorism (90%)
- Ptosis (50%)
- Downslanting palpebral fissures
- Strabismus
- Hyperopic astigmatism
- latent nystagmus
- blue sclera
- posterior embryotoxin
- corneal enlargement

Etiology

- X-linked recessive, linked to Xp11.21
- Female carriers have minor manifestations
- 100 cases reported

Other Findings

- Broad upper lip, anteverted nostrils
- Osteocondritis dessicans
- Stretchable skin

References

- [OMIM 100050](#)
- [OMIM #305400](#)
- *Pediatric Ophthalmology and Strabismus* eds. Wright KW, Spiegel PH. 2nd ed. p.1029

[syndrome](#)

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