AARSKOG-SYNDROME

INHERITANCE- X-linked recessive

GROWTH

Height
- Short stature, mild to moderate

Other
- Failure to thrive
- Delayed puberty
- Increased upper to lower segment ratio

HEAD & NECK

Face
- Round face
- Maxillary hypoplasia
- Wide philtrum
- Curved linear dimple below the lower lip

Ears
- Fleshy earlobes

Eyes
- Hypertelorism
- Ptosis
- Downslanting palpebral fissures
- Strabismus
- Hyperopia

Nose
- Small, short nose
- Anteverted nostrils
- Broad nasal bridge

Mouth
- Cleft lip

- Cleft palate
Teeth
- Hypodontia
Neck
- Short neck with or without webbing

CHEST
Ribs Sternum Clavicles & Scapulae
- Pectus excavatum

ABDOMEN
External Features
- Prominent umbilicus
- Inguinal hernia

GENITOURINARY
External Genitalia (Male)
- Shawl scrotum
Internal Genitalia (Male)
- Cryptorchidism

SKELETAL
Spine
- Cervical spine hypermobility
- Odontoid hypoplasia
- Scoliosis
Hands
- Short, broad hands
- Brachydactyly
- Clinodactyly
- Mild syndactyly
- Single transverse palmar crease
- Finger joint hyperextensibility
Feet
- Short broad feet

SKIN, NAILS, & HAIR
Skin
- Single transverse palmar crease
Hair
- Widow's peak

NEUROLOGIC

Central Nervous System
- Mental retardation (one-third)
- Attention deficit disorder - ADD
- Hyperactivity

MISCELLANEOUS

- Normal fertility

MOLECULAR BASIS

- Caused by mutation in the faciogenital dysplasia gene (FGD1, 300546.0001)