

Open Peer Review on Qeios

Dysmorphism-short stature-deafnessdisorder of sex development syndrome

INSFRM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

<u>Dysmorphism-short stature-deafness-disorder of sex development syndrome.</u>

ORPHA:2282

Dysmorphism-short stature-deafness-disorder of sex development syndrome is characterized by dysmorphism (including facial asymmetry, arched eyebrows, hypertelorism, broad and flat nasal bridge, microtia, small nose with anteverted nostrils, micrognathia), deafness, cleft palate, male pseudohermaphroditism, and growth and psychomotor retardation. It has been described in two siblings. It is transmitted as an autosomal recessive trait.

Qeios ID: U523EA · https://doi.org/10.32388/U523EA