

Open Peer Review on Qeios

Arachnodactyly-abnormal ossificationintellectual disability syndrome

INSFRM

Source

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

<u>Arachnodactyly-abnormal ossification-intellectual disability syndrome</u>. ORPHA:1129

Arachnodactyly - abnormal ossification - intellectual disability is a multiple congenital developmental anomalies syndrome characterized by arachnodactyly of fingers and toes associated with craniofacial dysmorphism (including abnormal cranial ossification, frontal bossing, flat calvaria, shallow deformed orbits resulting in exophtalmos, midface hypoplasia and micrognathia), feeding difficulties in infancy, infantile muscular hypotonia, and developmental delay leading to intellectual disability.

Qeios ID: GA1R8P · https://doi.org/10.32388/GA1R8P