Brachydactyly-short stature-retinitis pigmentosa syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Brachydactyly-short stature-retinitis pigmentosa syndrome. ORPHA:166035

Brachydactyly-short stature-retinitis pigmentosa syndrome is a rare, genetic, congenital limb malformation syndrome characterized by mild to severe short stature, brachydactyly, and retinal degeneration (usually retinitis pigmentosa), associated with variable intellectual disability, develomental delays, and craniofacial anomalies.