Congenital hereditary facial paralysis-variable hearing loss syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital hereditary facial paralysis-variable hearing loss syndrome. ORPHA:306530

Congenital hereditary facial paralysis-variable hearing loss syndrome is an extremely rare autosomal recessive disorder characterized by bilateral facial palsy with masked facies, sensorineural hearing loss, dysmorphic features (midfacial retrusion, low-set ears), and strabismus.