Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome. ORPHA:397623

Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome is a rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by short stature, conductive hearing loss due to bilateral auditory canal atresia, mandibular hypoplasia and multiple skeletal abnormalities, including bilateral humeral hypoplasia, humeroscapular synostosis, delayed pubis rami ossification, central dislocation of the hips, and proximal femora defects, as well as bilateral talipes equinovarus, proximally implanted thumbs and lumbar hyperlordosis. Associated craniofacial dysmorphism includes micro/scaphocephaly, malar hypoplasia, high-arched palate, and simple, dysplastic pinnae with prearicular pits/tags.