Cooper-Jabs syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cooper-Jabs syndrome. ORPHA:1488

A rare multiple congenital anomalies/dysmorphic syndrome characterized by auditory canal atresia (resulting in moderate conductive hearing loss) associated with intellectual disability, ventricular septal defect, umbilical hernia, anteriorly displaced anus, various skeletal anomalies (such as mild clubfoot, long fifth fingers, proximally placed thumbs), and craniofacial dysmorphism which includes brachycephaly, prominent forehead, flattened occiput, midface hypoplasia, anteverted nares, and low set, posteriorly rotated ears with overlapping superior helix. There have been no further descriptions in the literature since 1987.