Acrocraniofacial dysostosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acrocraniofacial dysostosis. ORPHA:949

Acrocraniofacialdysostosis is a very rare form of acrofacialdyosotosis, reported in two sisters to date, characterized by short stature, acrocephaly, ocular hypertelorism, ptosis of eyelids, ocular proptosis, downslanting palpebral fissures, high nasal bridge, anteverted nostrils, short philtrum, cleft palate, micrognathia, abnormal external ears, preauricular pits, mixed hearing loss, bulbous digits, metatarsus varus, pectus excavatum and various radiological abnormalities. Features of this syndrome were reported to overlap with otopalatodigital syndrome types 1 and 2. There have been no further descriptions in the literature since 1988.