Curry-Jones syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Curry-Jones syndrome. ORPHA:1553

Curry-Jones syndrome is a form of syndromic craniosynostosis characterized by unilateral coronal craniosynostosis or multiple suture synostosis associated with complete or partial agenesis of the corpus callosum, preaxial polysyndactyly and syndactyly of hands and/or feet, along with anomalies of the skin (characteristic pearly white areas that become scarred and atrophic, abnormal hair growth around the eyes and/or cheeks, and on the limbs), eyes (iris colobomas, microphthalmia,) and intestine (congenital short gut, malrotation, dysmotility, chronic constipation, bleeding and myofibromas). Developmental delay and variable degrees of intellectual disability may also be observed. Multiple intra-abdominal smooth muscle hamartomas, trichoblastoma of the skin, occipital meningoceles and development of desmoplastic medulloblastoma have been reported.