Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome. ORPHA:293967

Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome is a rare, non-acquired pituitary hormone deficiency syndrome characterized by severe, congenital microcephaly, facial dysmorphism (highly arched eyebrows, hypertelorism, convex nasal ridge, protruding ears with underdeveloped superior antihelix crus, micrognathia), bilateral sensorineural deafness and hypogonadotropic hypogonadism, in association with early feeding problems, myopia, moderate intellectual disability and moderate short stature.