Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Retinitis pigmentosa-intellectual disability-deafness-hypogonadism syndrome. ORPHA:3085

Retinitis pigmentosa - intellectual disability - deafness - hypogenitalism is an extremely rare syndromic retinitis pigmentosa characterized by pigmentary retinopathy, diabetes mellitus with hyperinsulinism, acanthosis nigricans, secondary cataracts, neurogenic deafness, short stature mild hypogonadism in males and polycystic ovaries with oligomenorrhea in females. Inheritance is thought to be autosomal recessive. It can be distinguished from Alstrom syndrome (see this term) by the presence of intellectual disability and the absence of renal insufficiency. There have been no further descriptions in the literature since 1993.