Bangstad syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Bangstad syndrome. ORPHA:1227

Bangstad syndrome is a rare endocrine disease characterized by the association of primordial birdheaded nanism, progressive ataxia, goiter, primary gonadal insufficiency and insulin resistant diabetes mellitus. Plasma concentrations of TSH, PTH, LH, FSH, ACTH, glucagon, and insulin are usually elevated. A generalized cell membrane defect was suggested to be the pathophysiological abnormality in these patients. The mode of inheritance was thought to be autosomal recessive. There have been no further descriptions in the literature since 1989.