

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Ataxia-diabetes-goiter-gonadal insufficiency syndrome
	Ataksja - cukrzyca - wole - niedoczynność gonad

Kod ORPHA	Kod OMIM	Kod ICD10
1227	210740	E31.8

Kod ICD11

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*Źródło

orphanet