

Opis choroby *

Definicja

A rare genetic disease characterized by intellectual disability, growth delay, absence deformities of upper and lower limbs, hypotrichosis, hypoplastic nails, abnormal dentition, abnormal auricles, hypoplastic nipples, thyroid enlargement, and abnormalities of tyrosine and/or tryptophane metabolism. Hypogonadism and cleft lip have also been reported. No new cases have been confirmed since 1970.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Freire-Maia syndrome
	Zespół Freire i Maia

Kod ORPHA	Kod OMIM	Kod ICD10
2723	273400	Q82.4

Kod ICD11
LD27.0Y

*Źródło

orphanet