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
Zespół wad wrodzonych

Synonimy
Freire-Maia syndrome
Zespół Freire i Maia

Kod ORPHA
2723

Kod OMIM
273400

Kod ICD10
Q82.4

Kod ICD11
LD27.0Y

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*Źródło

orphanet