

Opis choroby *

Definicja

X-linked intellectual disability, Nascimento type is a rare X-linked intellectual disability syndrome characterized by intellectual disability (with severe speech impairment), a myxedematous appearance, dysmorphic facial features (including large head, synophrys, prominent supraorbital ridges, almond-shaped and deep-set eyes, large ears, wide mouth with everted lower lip and downturned lip corners), low posterior hairline, short, broad neck, marked general hirsutism and abnormal hair whorls, skin changes (e.g. dry skin or hypopigmented spots), widely spaced nipples, obesity, micropenis, onychodystrophy and seizures.

Dane

Klasyfikacja

Choroba

Synonimy

X-linked intellectual disability-nail dystrophy-seizures syndrome
Niepełnosprawność intelektualna sprzężona z chromosomem X - dystrofia paznokci - napady padaczkowe

Kod ORPHA

163956

Kod OMIM

300860

Kod ICD10

Q87.8

Kod ICD11

LD90

[*Źródło](#)

orphanet