

Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by infantile-onset non-progressive intellectual deficit (with psychomotor developmental delay, cognitive impairment and macrocephaly) and early-onset parkinsonism (before 45 years of age), in male patients.

Dane

Klasyfikacja

Choroba

Synonimy

Laxova-Opitz syndrome

Zespół Laxova i Opitza

Zespół Waismana

Waisman syndrome

Kod ORPHA

2379

Kod OMIM

311510

Kod ICD10

G20

Kod ICD11

LD90.1

*Źródło

orphanet