

Opis choroby *

Definicja

Megalocornea-intellectual disability syndrome is a rare intellectual disability syndrome most commonly characterized by megalocornea, congenital hypotonia, varying degrees of intellectual disability, psychomotor/developmental delay, seizures, and mild facial dysmorphism (including round face, frontal bossing, antimongoloid slant of the eyes, epicanthal folds, large low set ears, broad nasal base, anteverted nostrils, and long upper lip). Interfamilial and intrafamilial clinical variability has been reported.

Dane

Klasyfikacja

Zespół wad wrodzonych MMR syndrome

Synonimy

Zespół MMR

Zespół Neuhäusera

Neuhäuser syndrome

Kod ORPHA

2479

Kod OMIM

249310

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet