Opis choroby *

Definicja

A rare partial autosomal monosomy characterized by global developmental delay, intellectual disability, multiple cartilaginous exostoses, and craniofacial anomalies (such as brachycephaly, biparietal foramina, large fontanels, craniosynostosis, ptosis, epicanthic folds, prominent nasal bridge with broad, depressed nasal tip, hypoplastic nares, short philtrum, downturned upper lip, and micrognathia). Additional reported features include behavioral abnormalities, myopia, strabismus, and sensorineural hearing loss, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
11p11.2 deletion
Delecja 11p11.2
Zespół proksymalnej delecji 11p
Proximal 11p deletion syndrome

Kod ORPHA
52022

Kod OMIM
601224

Kod ICD10
Q93.5

Kod ICD11
LD44.B1

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*Źródło

orphanet