

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

Kod ORPHA	Kod OMIM	Kod ICD10
52022	601224	Q93.5

Kod ICD11
LD44.B1

*Źródło

orphanet