

Opis choroby *

Definicja

Macrocephaly-spastic paraplegia-dysmorphism syndrome is a rare syndrome of multiple congenital anomalies characterized by macrocephaly (of post-natal onset) with large anterior fontanelle, progressive complex spastic paraplegia, dysmorphic facial features (broad and high forehead, deeply set eyes, short philtrum with thin upper lip, large mouth and prominent incisors), seizures, and intellectual deficit of varying severity. Inheritance appears to be autosomal recessive.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Fryns macrocephaly Makrocefalia Frynsa

Kod ORPHA	Kod OMIM	Kod ICD10
2429	600302	Q87.8

Kod ICD11
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*Źródło

orphanet