

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

Zespół wad wrodzonych Frys macrocephaly
Makrocefalia Frynsa

Kod ORPHA

2429

Kod OMIM

600302

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet