

Opis choroby *

Definicja

Microcephaly-cervical spine fusion anomalies syndrome is characterized by microcephaly, facial dysmorphism (beaked nose, low-set ears, downslanting palpebral fissures, micrognathia), mild intellectual deficit, short stature, and cervical spine fusion anomalies producing spinal cord compression. It has been described in two brothers born to consanguineous parents. Transmission is likely to be autosomal recessive.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2522

Kod OMIM

251250

Kod ICD10

Q87.8

Kod ICD11

LD20.2

*Źródło

orphanet