Opis choroby *

Definicja

Microbrachycephaly-ptosis-cleft lip syndrome is characterised by the association of intellectual deficit, microbrachycephaly, hypotelorism, palpebral ptosis, a thin/long face, cleft lip, and anomalies of the lumbar vertebra, sacrum and pelvis. It has been described in two Brazilian sisters. Transmission appears to be autosomal recessive.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Richieri Costa-Guion Almeida-Ramos syndrome
Zespół Richieri Costa, Guion Almeida i Ramosa

Kod ORPHA
2511

Kod OMIM
268850

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet