Opis choroby *

Definicja

Ring chromosome 17 syndrome is a rare chromosomal anomaly syndrome, resulting from partial deletion of chromosome 17, characterized by highly variable manifestations, ranging from a severe phenotype which presents with lissencephaly and severe intellectual disability to a milder phenotype that includes short stature, microcephaly, intellectual disability, seizures (that may be pharmacoresistant), café-au-lait spots, retinal flecks and minor facial dysmorphism, depending on the presence or absence of the Miller-Dieker critical region.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ring 17
Ring chromosome 17

Kod ORPHA
1441

Kod OMIM
-

Kod ICD10
Q93.2

Kod ICD11
-

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*Źródło

orphanet