Opis choroby *

Definicja

An extremely rare genetic syndrome characterized by the association of microcephaly, intellectual deficit and achalasia (with symptoms of coughing, dysphagia, vomiting, failure to thrive and aspiration appearing in infancy/early-childhood). Antenatal exposure to Mefloquine was reported in one simplex case.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
929

Kod OMIM
200450

Kod ICD10
Q39.5

Kod ICD11
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*Źródło

orphanet