Opis choroby *

Definicja

A rare constitutional hemolytic anemia that is characterised by the association of Alport syndrome, midface hypoplasia, intellectual deficit and elliptocytosis. It has been described in two families. The syndrome is transmitted as an X-linked trait is caused by a contiguous gene deletion in Xq22.3 involving several genes including COL4A5, FACL4 and AMMECR1.

Dane

Klasyfikacja
Choroba

Synonimy
AMME complex
ATS-MR
Kompleks AMME
Zespół AMME
AMME syndrome
ATS-MR

Kod ORPHA
86818

Kod OMIM
300990

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet