

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