Opis choroby *

Definicja

A rare constitutional hemolytic anemia due to a red cell membrane anomaly characterized by lack or severe reduction of Rh blood group antigens, resulting in increased osmotic fragility of red blood cells and chronic hemolytic anemia of varying severity with stomatocytosis and spherocytosis. Two types of the syndrome arising from independent genetic mechanisms have been distinguished: the regulator type is caused by defects of the Rh associated glycoprotein (encoded by the RHAG gene), while the amorph type is due to mutations at the RH locus itself.

Dane

Klasyfikacja
Choroba

Synonimy
Rh-null syndrome
Rh-null syndrome

Kod ORPHA
71275

Kod OMIM
268150

Kod ICD10
D58.8

Kod ICD11
3A10.Y

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*Źródło

orphanet