Opis choroby *

Definicja

A rare inborn error of metabolism characterized by a deficiency in erythrocyte catalase, an enzyme responsible for the breakdown of hydrogen peroxide. The disorder is usually asymptomatic but may be associated with oral ulcerations and gangrene, or diabetes mellitus and atherosclerosis in certain populations.

Dane

Klasyfikacja
Choroba

Synonimy
Catalase deficiency
Niedobór katalazy

Kod ORPHA
926

Kod OMIM
614097

Kod ICD10
E80.3

Kod ICD11
5C57.1

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*Źródło

orphanet