

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

*Źródło

orphanet