Opis choroby *

Definicja

A rare systemic disease characterized by progressive hyalinosis involving capillaries, arterioles and small veins of the digestive tract, kidneys, and retina, associated with idiopathic cerebral calcifications, manifesting with severe diarrhea (with rectal bleeding and malabsorption), nephropathy (with renal failure and systemic hypertension), chorioretinal scarring, and subarachnoid hemorrhage. Poikiloderma and premature greying of the hair may be additionally observed.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Rambaud-Gallian syndrome
Zespół Rambauda i Galliana
Zespół Rambauda, Galliana i Toucharda
Rambaud-Gallian-Touchard syndrome

Kod ORPHA
3018

Kod OMIM
277175

Kod ICD10
E78.8

Kod ICD11
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*Źródło

orphanet