

## 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

Rimbaud-Gallian syndrome

Zespół Rimbauda i Galliana

Zespół Rimbauda, Galliana i Toucharda

Rimbaud-Gallian-Touchard syndrome

#### Kod ORPHA

3018

#### Kod OMIM

277175

#### Kod ICD10

E78.8

#### Kod ICD11

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#### \*Źródło

orphanet