

Opis choroby *

Definicja

A rare, hereditary amyloidosis with primary renal involvement characterized by renal interstitial and medullary deposition of amyloid, low plasma levels of ApoA-1 and slow disease progression. Main clinical signs and symptoms are hypertension, proteinuria, hematuria and edema due to chronic renal insufficiency leading to end stage renal disease. Hepatosplenomegaly, progressive cardiomyopathy and involvement of skin, testes and adrenals (hypergonadotropic hypogonadism) have also been reported.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

Apolipoprotein A-I amyloidosis

Amyloidoza związana z apolipoproteiną A -
wariant I

Dziedziczna amyloidoza nerek spowodowana
wariantem I apolipoproteiny A

Dziedziczna nefropatia amyloidowa
spowodowana wariantem I apolipoproteiny A

Rodzinna amyloidoza nerek spowodowana
wariantem I apolipoproteiny A

Rodzinna nefropatia amyloidowa spowodowana
wariantem I apolipoproteiny A

Familial amyloid nephropathy due to
apolipoprotein A-I variant

Familial renal amyloidosis due to apolipoprotein
A-I variant

Hereditary amyloid nephropathy due to
apolipoprotein A-I variant

Hereditary renal amyloidosis due to
apolipoprotein A-I variant

Kod ORPHA

93560

Kod OMIM

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Kod ICD10

E85.0

Kod ICD11

5D00.2Y

[*Źródło](#)

orphonet