

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
Amylidoza związana z apolipoproteiną A - wariant I
Dziedziczna amylidoza nerek spowodowana wariantem I apolipoproteiny A
Dziedziczna nefropatia amyloidowa spowodowana wariantem I apolipoproteiny A
Rodzinna amylidoza 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](#)

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