Opis choroby *

Definicja

A rare hereditary, metabolic disease characterized by serum levels of alpha-1-antitrypsin (AAT) that are well below the normal range. In the most severe form, the disease can clinically manifest with chronic liver disorders (cirrhosis, fibrosis), respiratory disorders (emphysema, bronchiectasis), and rarely panniculitis or vasculitis.

Dane

Klasyfikacja
Choroba

Synonimy
Alpha-1-proteinase inhibitor deficiency
Niedobór inhibitora Alfa-1-proteinazy
Alpha1-antitrypsin deficiency

Kod ORPHA
60

Kod OMIM
613490

Kod ICD10
E88.0

Kod ICD11
5C5A

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*Źródło

orphanet