

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

*Źródło

orphanet