

Antitrypsin deficiency

- incidence: 1: 1,600–2,000;
- normal antitrypsin level is 1.5-3 g / l;
- It can manifest as neonatal hepatitis with cholestatic jaundice, chronic hepatitis or it can be asymptomatic;
- the most common metabolic cause of liver disease in children and early pulmonary emphysema in adult patients;
- there is no effective treatment, some patients require liver transplantation;
- prognosis - some are asymptomatic, others develop liver cirrhosis.



Alpha 1 antitrypsin deficiency (video).

α -1-antitrypsin.

Játra u pacienta s deficitem α -1-antitrypsinu.

Links

Reference

Alpha-1 Antitrypsin Deficiency - NORD (National Organization for Rare Disorders). *Home - NORD (National Organization for Rare Disorders)* [online]. Copyright ©2021 NORD [cit. 26.03.2022]. Available from <<https://rarediseases.org/rare-diseases/alpha-1-antitrypsin-deficiency/>>

Alpha-1 antitrypsin deficiency: MedlinePlus Genetics. *MedlinePlus - Health Information from the National Library of Medicine* [online]. Copyright © 2009 Terese Winslow LLC for the National Cancer Institute [cit. 26.03.2022]. Available from <<https://medlineplus.gov/genetics/condition/alpha-1-antitrypsin-deficiency/>>