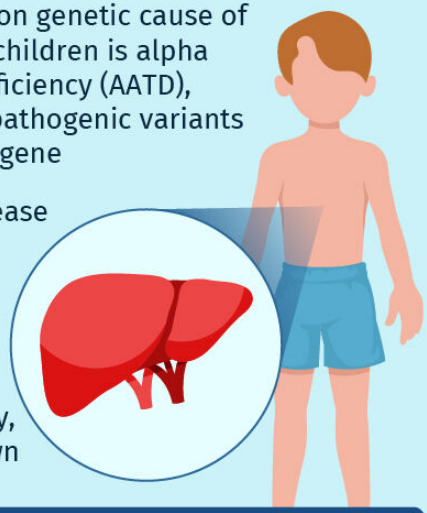
Evaluating Disease Status in Children with Alpha 1-Antitrypsin Deficiency

The most common genetic cause of liver disease in children is alpha 1-antitrypsin deficiency (AATD), attributable to pathogenic variants in the *SERPINA1* gene

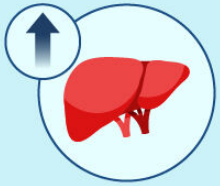
The state of disease at diagnosis in Danish children with AATD, who have a high incidence of ZZ-homozygosity, remains unknown



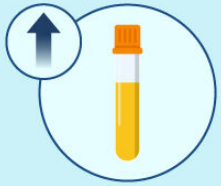
Retrospective cohort study of children with pathogenic variants in *SERPINA1* (N = 183)

Clinical characteristics | SERPINA1-genotype | Blood serum (S) concentrations

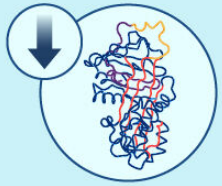
Compared with heterozygous children, ZZ-homozygous children showed:



Serum concentrations of liver enzymes



Conjugated bilirubin



Serum α1-antitrypsin (S-AAT)

Children under 6 months

High serum bilirubin concentrations

Children over 6 months

Normal serum bilirubin concentrations

A low S-AAT concentration and elevated liver markers strongly indicate homozygosity and measuring S-AAT concentration in children with prolonged neonatal jaundice is extremely important for disease diagnosis

Disease Status at Diagnosis in Danish Children with α1-Antitrypsin Deficiency

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