European Respiratory Society
Annual Congress 2013

Abstract Number: 800
Publication Number: P415

Abstract Group: 1.1. Clinical Problems
Keyword 1: Orphan disease **Keyword 2:** No keyword **Keyword 3:** No keyword

Title: Hereditary α1-antitrypsin deficiency in children and its clinical consequences

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Body: α1-antitrypsin deficiency (AATD) is a genetic disorder that manifests as pulmonary emphysema, liver cirrhosis and rarely skin panniculitis. The prevalence is estimated at 1: 2500 - 5000 newborns. Most common deficiency alleles are PI Z and PI S, and majority of pts with severe AATD are PI ZZ. Clinical manifestations may widely vary, ranging from asymptomatic to fatal liver or lung disease. The aim of the study is to present clinical observation in children with AATD. Methods: A retrospective review of case reports of AATD children population was undertaken. Age, genotypes, reasons for clinical investigation, hepatic and respiratory symptoms and functions were recorded. Results: 44 children were observed (28/PI ZZ, 12/PI MZ, 2/PI SZ, 1/PI MM and 1/PI FZ). All pts were diagnosed and enrolled because of liver disease. Majority pts presented increased serum transaminase level (96% PI ZZ pts, 50% PI MZ pts). In PI ZZ group 3 pts presented liver cirrhosis, one of them had already undergone liver transplantation. As far as the respiratory system is concerned the incidence of recurrent infections and pneumonia in AATD pts was comparable to healthy population. PFTs and CXRs were within normal range. Asthma was found only in 3 PI MZ and 3 PI ZZ pts.

Pts characteristics

Genotype	No/gender	mean age [yrs]	mean age at diagnosis [yrs]	AAT mg/dL	mean FVC [% pred]	mean FEV1 [% pred]	mean FEV1%VC
PI ZZ	21/M, 7/F	11.3 SD 4.7	1.0 SD 1.1	26.7 SD 7.4	99.3 SD 9.0	96.6 SD 9.2	82.8 SD 4.0
PI MZ	7/M, 5/F	8.6 SD 3.7	2.5 SD 1.8	50.5 SD 13.6	108.3 SD 13.3	106.9 SD 9.3	84.4 SD 5.0

AAT - plasma levels of α1-AT

Conclusions: In AATD in children acute or chronic liver disease development is observed, whereas respiratory system is not involved.