Datamining in the Alpha1 International Registry (AIR) Database

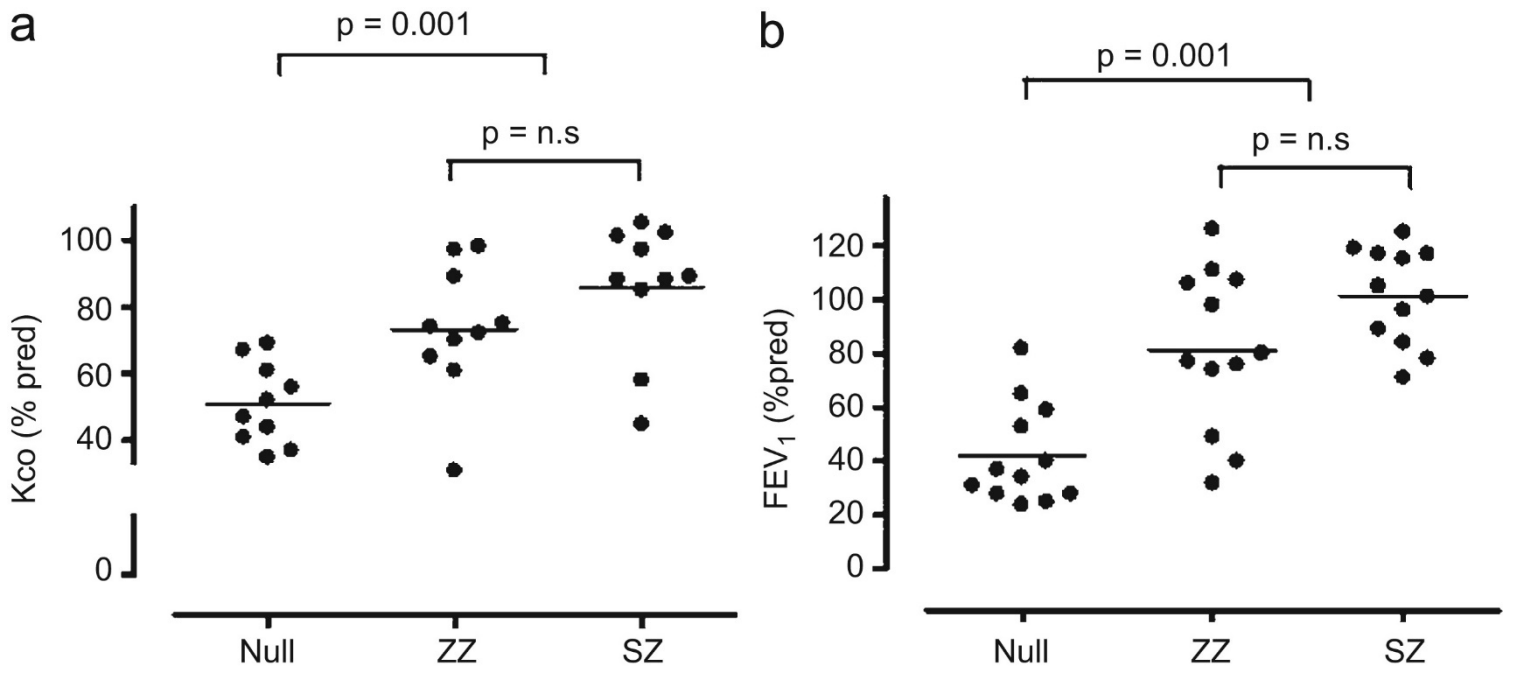
- AAT Null mutations are extremely rare
- There are 21 mutations known, its name is connected to the town name of first identified patient
- Mutation results in absence of AAT in serum
- A matched analysis of AIR data revealed worse clinical course

ISBN: 978-0-12-803942-7

Conflict of Interest

None

Matched analysis of Null AATD patients



Respir Med. 2008 ;102:876-84

Selection of patients with Null mutations

- a minimum of 6 years of follow-up registered in the database.
- Patients on iv AAT augmentation in It + Sp + Fr.
- Patients **not** on AAT augmentation in NL.
- Annual post bronchodilator FEV1 available
- No or ex-smokers during the period of follow up.

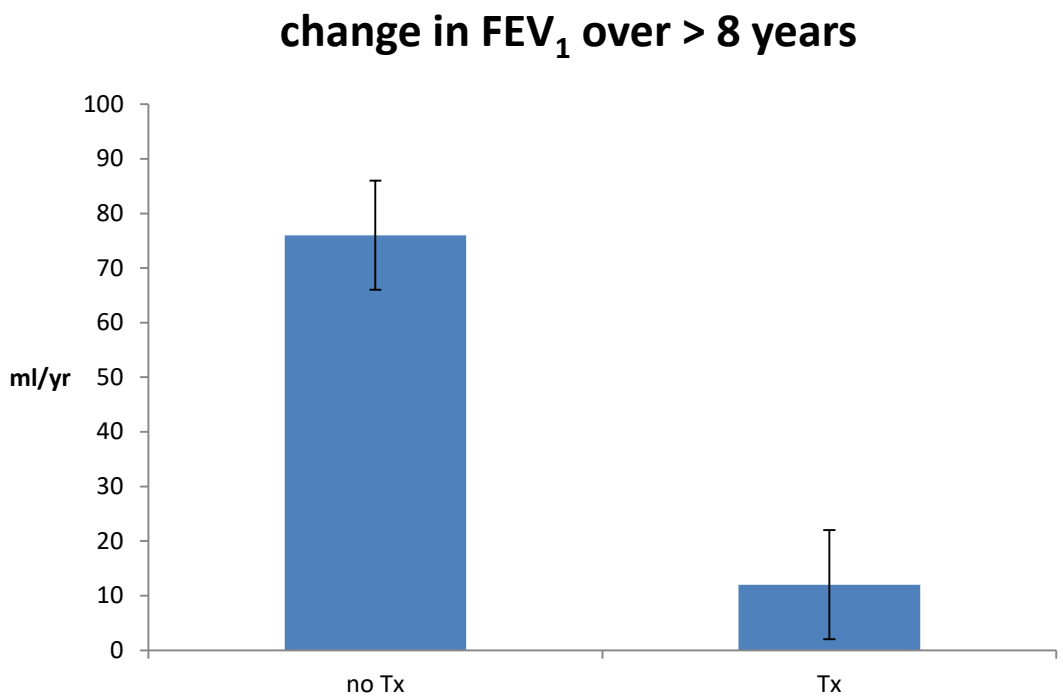
Patient characteristics

- Dutch patients (n = 12): Bellingham (2); Bredevoort (5), Soest (1); Granite Falls (2); Amsersfoort (1); Mattawa (1)
- It, Sp, Fr patients (n = 10): Brescia (3); Procida (1); Bellingham (2); Granite Falls (1); Lisbon (1); Perugia (2).
- Age at baseline between groups: $p > 0.05$
- Baseline FEV1 between groups: $p > 0.05$

Data analysis

- Mean follow up time Dutch group: 8.2 years (SD=2.7)
- Mean follow up time AAT Tx group: 9.0 years (SD=3.2)
- Data at baseline showed normal distribution
- FEV1 slope analysis
- Statistical analysis of FEV1 slopes by t-test.

Alpha-1-antitrypsin i.v. treatment effect in homozygous Null AATD



Conclusions

- Alpha1 International Registry (AIR) is a rich source for analysis of clinical characteristics of pulmonary disease in AATD
- AIR data quality control is a challenge
- In a selected population of ultra-rare AAT mutations, iv AAT augmentation seems to protect against decline in FEV1