International Conference 'Advances in Pneumology'
Bonn, 17-18 June 2011

ALPHA-1-ANTITRYPSIN DEFICIENCY SCREENING PROGRAM IN POLAND

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Background: In Poland, the overwhelming majority of individuals with alpha-1-antitrypsin (AAT) deficiency still remains undiagnosed. We estimated the AAT gene frequency and prevalence in a large cohort of Polish chronic lung or liver disease patients eligible for AAT testing.
Methods: blood samples were collected prospectively from 500 respiratory patients (COPD, emphysema, bronchiectasis, asthma). AAT serum concentration was measured by turbidimetry and PI-phenotype identified by isoelectrofocusing. The PI*S and PI*Z alleles were confirmed by real-time PCR; rare phenotypes were characterized by sequencing.
Results: 63 (12.6%) lung disease patients demonstrated AAT deficiency phenotypes. Calculated frequencies expressed per 1000 were for PI*Z 46.6 (95% CI: 32.3-60.8), PI*S 20,3 (95% CI: 10.8-29.8). The AAT gene prevalence calculated by Hardy-Weinberg equilibrium were: 1/1.16 for MM, 1/26 for MS, 1/2429 for SS, 1/11 for MZ, 1/530 for SZ and 1/462 for ZZ.
Conclusion: Our results show relatively high frequency of AAT deficiency among Polish patients with chronic obstructive respiratory disorders. Estimated frequency for PI*Z and PI*S allele in respiratory group was about four-fold higher than estimated prevalence in healthy Polish population.