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Title: The incidence of alpha-1-antitrypsin (A1AT) deficiency alleles in Polish population – Results from the ongoing newborn screening

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Body: Data concerning the prevalence of A1AT deficiency in Poland are very limited due either to small groups analyzed or unreliable methodology. Here, we present the up-date from the large scale newborn screening ongoing in Central Poland. Methods: DBS samples were collected prospectively from 2423 newborns between September 2011 – December 2012. AAT serum concentration was measured by nephelometry and PI-phenotype identified by real-time PCR. The PI*S and PI*Z alleles were confirmed by isoelectrofocusing. Results: Deficiency S- or Z allele was observed respectively in 54 (2,23%) and 55 (2,27%) DBS samples (in total 109 (4,5%) samples). Calculated frequencies expressed per 1000 were for PI*Z 11,3 (95% CI: 8,4-14,3), PI*S 11,1 (95% CI: 8,2-14,1). The AAT gene prevalence calculated by Hardy-Weinberg equilibrium were: 1/1.04 for MM, 1/46 for MS, 1/8053 for SS, 1/45 for MZ, 1/3953 for SZ and 1/7763 for ZZ. The mean A1AT concentration was 177±39 mg/dl, in non-S non-Z individuals 179±38 mg/dl, in PI*S carriers 154±36 mg/dl, in PI*Z 116±17 mg/dl. Conclusion: Our results suggest considerably higher prevalence of deficiency alleles in Polish population than currently available data.