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Title: Screening for alpha-1 antitrypsin deficiency in Germany – Update 2012

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Body: Background: Alpha-1 antitrypsin deficiency (AATD) is characterized by decreased serum levels of alpha-1 antitrypsin (AAT). The most common clinical manifestations are pulmonary emphysema and liver cirrhosis. Epidemiological estimates postulate around 8000 people in Germany with a severe AATD. Although current guidelines stress the importance of screening for AATD, the majority of patients remains undetected. Aim: To provide recent screening data from the German central laboratory for AATD in Marburg, Germany. Methods: From dried blood spot (DBS) samples we performed AAT measurements (for internal use only) and genotyping for S and Z alleles. When either of both tests was suggestive for AATD we went on to perform phenotyping by IEF. When phenotyping resulted in bands suggestive for rare deficiency alleles we conducted complete sequencing of the AAT gene. Results: In the period from August 2003 to February 2012 more than 50.000 test kits had been requested of which 13.010 kits have been returned. Of these, 75 were not evaluable, and 185 samples had already been submitted before. Our results are based on 12.750 analyzed samples. In descending order of frequency, we have diagnosed the following phenotypes: PIMM (8577, 67.27%), PiMZ (2383, 18.69%), PiZZ (846, 6.64%), PIMS (637, 4.99%), PiSZ (192, 1.51%), PiSS (38, 0.29%). 140 samples were submitted to gene sequencing. Here we found 75 rare (R) genotypes (PiZR 59; PiMR 10; PiSR 4; PiRR 2). Conclusion: Almost a third (32.73%) of the submitted samples was found to represent at least a carrier status, and over 8% carried a genotype that is associated with a severe AATD. We conclude that screening is useful to detect AATD and should be expanded in Germany.