33* *CFTR* gene mutations in IRT/DNA cystic fibrosis newborn screening in Poland

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Screening of newborn infants for Cystic Fibrosis (CF NBS), one of the most common monogenic diseases among Caucasians, has already been introduced in many countries. One of the most frequent CF symptoms is malnutrition, which can easily be overcome after rapid diagnosis. Due to healthcare system organization in Poland prompt specialists availability is rather difficult resulting in late CF diagnosis. Therefore CF NBS seems to be here of additional advantage.

CF NBS covers 8 Polish regions and is organized according to current guidelines including IRT determination in dried blood spots, identification of the *CFTR* gene mutations and sweat chloride test. Our strategy of molecular analysis is based on sequencing of five *CFTR* exons (7, 10, 11, 13, 21). This allows to identify 15 most frequent mutations in Poland and about 300 rare variants. Analysis based on commercial tests (INNOLiPA 12/19/17+Tn, Innogenetics; OLA Assay v3, Abbott) does not seem to be sensitive enough as only about 50% of mutations covered by them have ever been identified in Polish population.

CF NBS was started in September 2006. Until December 2007, 1005 samples, selected on the basis of IRT level over cut-off, underwent DNA analysis. Molecular confirmation of CF diagnosis was achieved in 30 patients with genotypes *F508del/F508del (15 cases), *F508del/other mutation (7), *other mutation/other mutation (8).

In 3 patients with CF confirmed clinically only 1 mutation (F508del) was identified. The frequency of F508del among CF alleles in tested population was evaluated as 60.6%.

We also identified 80 symptomless carriers. Their follow-up observation is necessary. Further study will result in recognition of CF epidemiology and heterogeneity of *CFTR* mutations in Poland.