UMD-CFTR-France: a model of national database for collection and analysis of extensive molecular data in CF and CFTR-related diseases (CFTR-RD)

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CF and particularly CFTR-RD provide a challenge for molecular diagnosis because of many unclassified variants (UV) and identical genotypes associated with different phenotypes. There is a need for an accurate and exhaustive collection of sequence variations identified in patients suffering from disease related with the CFTR gene as most of these variants reside in genetic testing laboratories and remain unpublished. Since 2009, we have collected and curated molecular and minimal clinical data from 9 French expert laboratories. Using the Universal Mutation Database (UMD®) software, we have constructed the UMD-CFTR-France, an exhaustive and manually-curated database, dedicated to sequence variations of the CFTR gene identified in CF and CBAVD patients, in patients analysed in the context of newborn screening, chronic rhinosinusitis, bronchiectasy, pancreatitis, nasal polyposis, fetal cp. aeruginosa.

Cystic fibrosis in Republic of Moldova

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Actuality: 57 patients with cystic fibrosis (CF) are evaluated and treated in Clinic of Pneumology, Republic of Moldova.

Aim: In this work we assessed the peculiarities of genetic findings, clinical symptoms and explorative picture in children with CF from Republic of Moldova.

Materials and Methods: This study included 29 boys, 28 girls, mean age 8.91±0.45 years. Genetic diagnosis was realized in children and their parents for 4–8 CFTR mutations (F508del, R334W, N1303, G551D, R347P, R553X, R117H). More rare mutations were revealed in Center of Bordeaux. The evaluation program for CF patients include: anthropometry, chest radiography, computed tomography (CT), spirometry, sputum bacteriology, serum biochemistry, coprology.

Results: F508del mutation was revealed in 57.8% cases (inclusive 24.5% – homozygotes), 1 child – N1303K, 1 case – R347P. In Bordeaux were discovered mutation in 4 children (F508del/296+1G→C, F508del/185+1G→T, G542X/N1303K, 128+1G→A/1677delTA). The mean age of CF diagnosis was 2.65±0.03 years: at the age of <1 year in 52.6% children, 1–5 year – 38.5% children, 5–10 years – 7.1% cases; 10–18 years – 8.7% children, 19 years – 1.7% cases. Ps. aeruginosa was isolated in 43.7% cases. CT has revealed bronchiectasis (34.3% cases), pulmonary fibrosis (23.8% cases) and chronic bronchitis (68.8% cases). Spirography showed pulmonary function impairments (FVC – 65.8±1.2%, FEV1 – 68.8±2.6%). Nutrition impairment was established in 71.5% cases.

Conclusion: Lung affecting and nutrition problems are present in most children with CF from Moldova. There is a prevalence F508del CFTR mutation in CF patients in Moldova.