Phenotyping the 711+1 G→T mutation in cystic fibrosis patients
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Objectives: To describe the phenotype associated with the 711+1 G→T mutation in cystic fibrosis patients in Serbian patients with classical clinical diagnosis, pancreatic status) were similar (p>0.05) between patients with a 711+1G→T mutation and a DF508 mutation when other initial factors were taken into account including the mutation on the second allele.

Results: All measured variables for respiratory (FEV1), nutritional (BMI and BMI%), infectious (age at first Pseudomonas colonization, presence of other pathogens) status and presence of complications (diabetes, significant liver disease, nasal polyps) as well as initial characteristics at diagnosis (sweat test value, age at clinical diagnosis, pancreatic status) were similar (p>0.05) between patients with a 711+1G→T mutation and a DF508 mutation when other initial factors were taken into account including the mutation on the second allele.

Conclusion: Patients with a X/711+1G→T genotype are expected to have a similar clinical course as patients with a X/DF508 genotype. Genetic counseling for these patients can therefore be done in a similar way.

CFTR rearrangements in Serbian patients with classical cystic fibrosis
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Objectives: Although majority of over 1600 mutations identified in CFTR gene are point mutations or small deletions/insertions, large gene rearrangements have been reported with a relatively high frequency in various ethnic groups. The aim of our study was to define the incidence of CFTR gene rearrangements in Serbian CF patients in which one or no other mutation was identified after the detection of 7 most common mutations (c.1521_1523delCTT, c.489+1G→T, c.1624G→T, c.1652G→A, c.1657C>T, c.1585–1G→A; c.3909C>G) or after the sequencing of the whole CFTR gene.

Methods: Out of 154 patients referred to our Laboratory for molecular analysis, the clinical diagnosis was not confirmed in 27 patients. Seven CFTR rearrangements (dele1: c.4_53+69del53+c.4489G→insG; dele2: c.54–5811_164+2186delT73+6780_237+6961insG; dele2_3: c.54–5490_273+10250del; dele14_17b: c.2620–674_3367+198del; dele17a_18: c.2988–173_3468+2111del; dele22_23: c.3964_78_4242+577del; dele22_24: c.3964–3890_3143delinsTACTG) were detected by duplex PCR assay. The breakpoints were confirmed by sequencing.

Results: We identified CFTR gene rearrangements in 3/27 patients. In one patient dele2 was identified (genotype c.1521_1523delCTT/dele2), and in two dele2_3 was identified (genotypes: c.1521_1523delCTT/dele2_3 and c.1753G→T/dele2_3) thus confirming clinical CF diagnosis.

Conclusions: These results are in concordance with our previous findings that the molecular basis of CF in Serbia is highly heterogeneous. It would be beneficial if the detection of these rearrangements would be incorporated in routine diagnostic analysis of CF since their incidence was relatively high and the method was reliable and inexpensive.