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CFTR mutations unidentified in CFTR2 database and their phenotypic characteristics: Data from cystic fibrosis registry of Turkey

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Abstract

Background: Cystic Fibrosis Registry of Turkey shows various CFTR mutations due to the geographical location and historical background of our country, and also high prevalence of consanguineous marriages.

Method:All mutations detected in the Cystic Fibrosis Registry of Turkey 2017 (CFRT2017) data were screened in CFTR1 and CFTR2 databases. Mutations which were not found in both were identified and characteristics of these patients were compared with F508del homozygous patients.

Results: Among 1170 registered patients, 978 were genotyped and 200 different mutations were shown in 1270 alleles.29 mutations were not reported in both databases; 58 mutations have been reported in CFTR1 but not in CFTR2. Demographic and phenotypic characteristics of the 112 patients with 87 different alleles those were not previously reported in the CFTR2 database (nonCFTR2 group) were compared with F508del homozygous 103 patients in CFRT2017. In the nonCFTR2 group, mean age was younger (5.81 vs 7.69; p:0.015), mean age at

Conclusion: We suggest that patients in the nonCFTR2 group have a mild clinical course, but in some patients, further investigations and functional studies are required for the exact diagnosis.

Children Cystic fibrosis Genetics

Footnotes

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