Molecular Genetic Diagnostic Difficulties in Two Hungarian Gypsy Samples with Cystic Fibrosis

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ABSTRACT The frequency of DF508 mutation in the CFTR gene was compared in Gypsy and European samples from 3 different geographical regions of Hungary. The frequency of DF508 mutation in a total of 21 Gypsy patients was 43%, with 0.144 homozygosity index. This frequency was 50% with 0.127 homozygosity index in a total of 531 European Hungarian patients. Among the Gypsy patients 52 % had unknown mutations, but not the G542X, G551D, R553X and N1303K ones. However, there was a *geographical difference* in the distribution of homozygous DF508 mutations. In the two Gypsy samples of 13 Gipsy patients from *North-East* Hungary, only one possessed DF508 homozygote genotype, while all 7 Gypsy patients harboured this genotype from *South-West* Hungary. The difference in the occurrence of this mutation between the two geographically different Hungarian Gypsy samples can be explained by their different gene pools connected with their previous and present location, genetic drift and their isolation from each other. These findings need to be considered when planning any population screening programme for CF.

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