Molecular genetic diagnostic strategies of cystic fibrosis (CF) are dependent on the mutation distribution of the analyzed population. The goal of our study was to establish the mutational spectrum of 39 clinically diagnosed severe CF patients in Eastern Hungary.

For the mutation testing, multiplex commercially available diagnostic assays were used and in patients having mutation not included in the panel, the entire coding region of the CFTR gene was sequenced.

Using the mutation detection kits only, the detection rate was 81%. When DNA sequencing was used, disease-causing mutations could be detected in 77 out of the 78 CFTR alleles (99% sensitivity). Presence of six mutations (delta F508, CFTRdele2.3(21kb), 2184insA, N1303K, G542X and L101X) was shown in 31% of CF chromosomes.

Our results suggest that in Eastern Hungary the majority of CF-causing mutations are small-scale. Two mutations (CFTRdele2.3(21kb), 2184insA) were found in surprisingly high frequency. Based on our data, the Eastern Hungarian CF mutation testing panel can be established.