

Abstract:

Cystic fibrosis (CF) is genetically conditioned, autosomal recessive disease that occurs in the European population with a prevalence of about 1:2500 – 1:1800. In this disease we observe a mutation of the *CFTR* gene with subsequent fault in chloride channels. Such afflicted individuals usually suffer from chronic respiratory problems, pancreatic insufficiency, high concentration of chloride ions in sweat and obstructive azoospermia. Genetic testing of *CFTR* gene is indicated in individuals who meet the CF clinical picture and a positive sweat test (increased concentration of chlorides in the sweat). Genetic testing of the *CFTR* gene is usually done by using commercial kits detecting the most common mutations of the *CFTR* gene in the Czech Republic. If the testing results are negative, it is further performed an MLPA method that captures the larger deletions and duplications of gene, eventually a sequencing of all exons is. Despite the well-established algorithm of the testing, some patients suffering from symptoms of CF are left without genetic findings. Thanks to development of next generation sequencing, it is possible to make the diagnosis of CF more effective and uncover the variants that were not captured by previous methods.