Summary

Cystic fibrosis (CF) is the most frequent serious autosomal recessive genetic disease. CF is caused by mutations in the gene coding for Chloride transporter on the cell membrane (66). According to the statistics, this disease has prevalence 1 in 2500 to 3500 live newborns in European populations, where in Czech republic about 40 affected children a year are born (63). With respect to very serious clinical symptoms of this disorder (which substantially shortens the life expectancy of affected individual) in all newborns born in Czech Republic since October 1, 2009 the newborn screening from the dry blot spot is being performed. Blood samples from newborns who have in the primary test increased levels of IRT over given cut off are being sent to molecular genetic laboratory for analysis of the most frequent, pathogenic and population-wise important mutations in the *CFTR* gene.

DNA isolation is being performed from the same dry blood spots on the screening card, which correspond to those where the increased IRT were found. It is important to choose the most suitable isolation method with respect to DNA yield and concentration. Those are crucial for quality and turn around time of molecular genetic analysis results and influence the following information retrieval . There are 2 mutations found in affected individuals, sometimes 1 homozygous mutation in trans position, in the CFTR gene. Until today there was described more then 1500 mutations in this gene and their rapid identification plays important role in the patient's early therapy. Currently, the patients with CF have average life expectancy 30-35 years. This is improvement accomplished by new approaches in the patient's care and as well as by the possibility of early diagnosis through the newborn screening. Currently newborn screening crucially influences survival and the quality of live of the affected children thanks to the new approaches introducing targeted gene therapy. Important criterion for this therapy is absence of irreversible tissue damage especially in sinopulmonal tract (20). Spectrum of offered analytical methods is very broad and so it is important the employ the correct algorithm while choosing particular analytical protocol for *CFTR* gene mutation confirmation or exclusion.

Key words: DNA, cystic fibrosis, newborn screening, *CFTR* gene, IRT, genetic analysis, molecular diagnostic, mutation testing