Diagnostic test results are crucial for treatment management and family planning of an individual. Considering that around 80% of medical decisions are based on diagnostic tests and that genotyping is usually concluded only once in a lifetime, it is of a great importance to assure highly accurate test results and provided under high quality standards.

Cystic fibrosis (CF) is one of the most common and life-threatening autosomal recessive genetic disease affecting mainly Caucasian populations. CF is caused by mutations in the CFTR gene and until this date, more than 1900 mutations have been detected, while only few of them have frequencies higher than 1% worldwide. Thus, to confirm the diagnosis of cystic fibrosis in patients where only one disease-causing mutation has been found, it is necessary to apply a sensitive test to search for uncommon CFTR gene mutations/variants. In this work, we have successfully used HRM for gene scanning of certain exons of the CFTR gene. We have confirmed the numerous advantages of the HRM method for gene scanning and also detect some limitations that must be considered through an implementation process in a DNA diagnostic laboratory.

Hyperhomocysteinemia has been proposed as a risk factor for several diseases such as recurrent pregnancy loss and inherited thrombophilia and might be caused from acquired or genetic factors. One of the genetic factors are the c. 677 C>T and c. 1298 A>C variants in the MTHFR gene which alter the enzymatic activity of the Methylenetetrahydrofolate reductase enzyme (MTHFR) and participate in the homocysteine metabolism. Hence, the genotyping of these variations has become of importance to establish the diagnosis of disorders related to hyperhomocysteinemia in affected patients. Using a variation of HRM reducing amplicon sizes we were able to successfully genotype the c. 677 C>T and c. 1298 A>C variations.