Abstract

DNA analysis aimed at individual identification of persons is based on the examination of a panel of STRs (Short Tandem Repeats). These polymorphic loci were selected on the basis of broadly conceived international testing and the frequencies of individual alleles across different ethnicities are known.

Currently, attention is also paid to polymorphisms involving larger regions of DNA - the so-called Copy Number Variants (CNV). According to the literature (Repnikova et al., 2013), it turns out that these CNVs can also affect areas of forensically significant STR loci, resulting in, for example, the deletion of one of the alleles. The examined person, then in such an affected locus is not a homozygote, but a hemizygote. Otherwise, when duplication event of one of the alleles occurs, a tri-allelic STR arises. Such situations can then distort the result of individual identification, or even the correct evaluation of kinship relations.

In the scope of master's thesis, a methodology based on digital droplet PCR (ddPCR) will be established, which will allow verification in cases where routinely obtained DNA profiling results in the suspicion of the presence of a third allele in one of the STR loci. Samples for testing will be obtained in cooperation with the Institute of Criminology in Prague on the basis of a jointly designed research project. 200 000 DNA profiles obtained from control samples - buccal swabs - will be screened. The result of the screening will be the selection of suspicious samples for testing using the ddPCR method. We assume that we will find rare CNVs and validate their presence using the ddPCR method.

Key words: polymorphisms, Short Tandem Repeats (STR), Copy Number Variants (CNV), droplet digital PCR (ddPCR), tri-allelic, profiling