Abstract

Nonallelic homologous recombination (NAHR) mediated by LCRs (low-copy repeats) produces chromosomal rearrangements in the human genome. Those rearrangements include microdeletion and microduplication. Those mutations cause a great number of syndromes and thus are studied along with its genesis. Studies are enabled by the development of methods, which are able to detect those cryptic aberrations, e.g. comparative genomic hybridisation (CGH). Nowadays scientists often come across the mirror phenotype of the already described microdeletion (microduplication) syndromes. The presence of the reciprocal microduplication (microdeletion), which afflicted a gene sensitive to gene dosage or other important region of the human genome, is discovered by a genomic analysis. The examples of those affected chromosomal regions (and associated diseases) are areas 1q21.1; 5q35.2-3 (Sotos syndrome); 7q11.23 (Williams-Beuren syndrome); 16p11.2 až 12.2 a 16p13.11; 17q11.2 (Neurofibromatosis type 1); 17p11.2-12 (CMT1A/HNPP) a 22q11.2 (DiGeorge syndrome and VCFS).

Key words: microduplication; microdeletion; nonallelic homologous recombination (NAHR); comparative genomic hybridisation (CGH); mirror phenotype; reciprocal rearrangements.