Microdeletion syndromes occur in high incidence in the population, the most common syndrome is DiGeorge syndrome. Its incidence is 1 : 4 000. Another relatively frequent syndromes are Williams-Beuren syndrome, Miller-Dieker syndrome, 1p36 deletion syndrome and others. Most of these syndromes cause the serious clinical disorder to their carriers. Early and reliable diagnosis can help in treatment of the affected patient (education, surgical correction, physiotherapy, pharmacotherapy) or in prenatal diagnostics. We use FISH (fluorescent in situ hybridization) or PCR (polymerase chain reaction) based methods to analyse cryptic chromosomal rearrangements, which cause these syndromes.