The focus of this diploma thesis is on mosaic numerical and structural chromosomal aberrations. In its theoretical part, general problems of mosaicism, its phenotypic effect, mechanisms of origin, related epigenetic modifications, and diagnostic options are described. The methodical part of the thesis then primarily refers to fluorescence *in situ* hybridization (FISH) and its application in the diagnostics of mosaicism. This method was used in the examination of 29 patients with numerical as well as structural abnormalities of autosomes or gonosomes with proven or suspected mosaicism. On the basis of this analysis, possible errors of measurement were determined and data for statistic evaluation were retrieved. For the examinations of three patients an alternative of the comparative genomic hybridization, the array CGH technique, was applied. The FISH method, although being based on random selection and human factor, proved sufficient sensitivity as well as specificity in the field of low-frequency mosaicism diagnostics. The main critical factors responsible for potential misinterpretation of the data arose from inherent characteristics of the biological material, incorrect targeting of the analysis, probe instability, bleed through effect and absence of mitosis during the structural aberrations analysis.