This bachelor thesis deals with mosaicism and its detection. Mosaicism is defined as the presence of two or more cell lines with different karyotype in the patient's body. An aspect of particular importance for the resulting phenotype is the overall ratio in which the individual cell lines are represented. It is often found in Turner and Klinefelter syndrome. Turner syndrome is caused by the loss of a part of, or the whole of the X chromosome in females and it belongs among the most common chromosomal constitutions, found in spontaneous abortions. However, it is estimated that only about half of the women with Turner syndrome have karyotype 45,X; the others are mosaics or have other abnormalities of the chromosome X.

The main aim of the practical part of this thesis was to evaluate the possibilities of detecting mosaicism using fluorescence in situ hybridization (FISH) and to consider the possible factors affecting the accuracy of the examination. The objectives of the assessment were the individual variability between the evaluators and also the differences caused by using two differently labelled probes. As a result, the thesis confirms that the FISH method enhances the accuracy of the results of conventional cytogenetic examination and it is suitable for analysis of mosaicism thanks to the possibility of analyzing a large number of cells.