The primary focus of this diploma thesis is on marker chromosomes and phenotypically similar human karyotype polymorphisms, variants of short acrocentric arms in particular. The first half provides a very useful review of literature concerning different aspects of both sSMC and human polymorphisms such as their origin, inheritance, associated phenotype, formation and molecular cytogenetic methods that are applied in the process of identification of these aberrations. The methodical emphasis is on FISH and its modifications (e.g. M-FISH, acro M FISH, cen M-FISH) as well as on the CGH methods. The main objective was to test the above-mentioned methods and state their limitations and applications. Thus, in the other half we provide evaluations of commonly used methods and introduce new strategies that could be implemented to make the identification of these additional chromosomes or satellite translocations more effective. All the conclusions are based on the analysis of 7 patients with sSMC and 4 patients with variants involving acrocentric NOR regions. The results of our thorough research into their karyotypes have been compared with similar findings in the literature. Last but not least, we tried to establish a link between observed abnormalities and the type of a chromosomal aberration at hand.