Abstract

Trichorhinophalangeal syndrome is a malformation syndrome characterized by craniofacial and skeletal abnormalities and is inherited in an autosomal dominant manner. We distinguish free subtypes on clinical and molecular level – TRPS I, TRPS II, TRPS III. All TRPS patients have sparse hair, a pear-shaped nose, a long flat philtrum, a thin upper lip and protruding ears. Skeletal abnormalities include cone-shaped epiphyses at the phalanges, hip malformations and short stature are present. The subgroups TRPS I and TRPS III are result of the mutated *TRPS1* gene, which is maped into the 8q24 region. This gene is situated proximal of the *EXT1* gene, both genes are affected in a subgroup of patients with TRPS II. These patients suffer more from multiple (cartilaginous) exostoses and mental retardation.
In this work we performed molecular genetic analysis of a sample of 16 patients, 8 probands showed a TRPS phenotype and 8 probands had only isolated exostoses. The peripheral venous blood of patients was used to gain purified DNA, which was subsequently used to investigate the chromosome 8q24 region using MLPA („multiplex ligation-dependent probe amplification"). This analysis revealed a deletion in 1 TRPS patient and 1 patient with exostoses. Sequencing of the *TRPS1* gene coding exons in remaining 7 TRPS patients revealed mutations in another 2 of them. Overall it was revealed probable molecular cause of the disease in 3 TRPS patients and 1 patient with exostosis.