

Abstract

Introduction: Genetic determination of human face is clearly visible in family members. The resemblance between monozygotic twins who are genetically identical is especially remarkable. So far the possibilities of reliable prediction of the complex morphology of facial traits on the basis of genome analysis and the ability to capture the variability of human facial morphology through genotype variability are highly limited. Complete genetic basis of the physiological variability of craniofacial traits remains more or less unknown. This master's thesis was created as a pilot study of the shared project of the Laboratory of 3D Imaging and Analytical Methods and the Laboratory of Molecular Anthropology on Department of Anthropology and Human Genetics.

Material and Methods: The specimen collection is composed of DNA samples derived from 30 families (29 with 4 members, 1 with 5 members) who fulfilled required criteria. Nine single nucleotide polymorphisms were chosen based on the available information. Eight of them are linked to normal facial variability and one was chosen based on the assumed function of the gene where the polymorphism is located. There were two methods of genotyping: RFLP method with the use of restriction endonuclease and SNaPshot method. Morphological data were provided by the Laboratory of 3D Imaging and Analytical Methods.

Conclusion: We confirmed the association of two polymorphisms with human facial morphology. SNP rs6569759 influences the eye width, the biocular length and the nasion-chelion dimension, and the rs4648379 polymorphism is linked with pronasale-chelion and nasion-chelion dimensions. We found that the genotype effect does not have to be identical in men and women and in some cases we observed different trends. While looking for links between parents and their children, we discovered a strong correlation between mothers and daughters and their facial height, and stronger general correlation between sons and fathers. The parental contribution to the child's phenotype does not have to be 50% in all parameters and differences between sexes might occur.

Key words:

genetic determination of craniofacial morphology – heredity – SNP – SNaPshot