

VALIDATION OF CANDIDATE SNPS AND CPGS FOR HUMAN FACE PREDICTION

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Background: Human facial genetics remains challenging due to the complexity of facial morphology, its highly polygenic basis, and the combined influence of genetics, environment, age, and lifestyle on facial appearance.

Objective: To develop predictors of human facial appearance that could aid facial reconstruction in forensic anthropology and predict facial appearance using DNA-based methods alone.

Subjects and methods: Data from ~800 individuals, including Illumina GSA genotypes, EPIC methylation profiles, metadata, and 3D facial scans, were analyzed. Candidate facial SNPs were evaluated using MeshMonk with facial segmentation and landmark-based phenotyping. Associations were tested between 747 DNA variants and facial traits in 658 individuals. In addition, BMI-predictive CpG sites were validated, and a compact BMI prediction model was developed using Elastic Net regression on methylation data from 624 individuals.

Results: Using global-to-local segmentation approach we found 29 SNPs to be significantly associated with 22 segments of the faces. The facial landmarking approach identified 27 candidate SNPs to be significantly associated with 60 facial distances. Elastic Net regression yielded 30 CpG BMI predictors. In the validation set, the model achieved a mean absolute error of 2.9 kg/m². The current results represent a step forward in our research on facial prediction.

Keywords: Human face morphology, BMI, Phenotyping, DNA-based prediction.

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