DEEP-ROOTING AFRICAN INFLUENCE IN SOUTHWESTERN EUROPE: A VIEW FROM MTDNA IN THE IBERIAN PENINSULA

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In the past few decades, the gene flow between North Africa and Europe has been a topic of interest for population genetic studies. Key unanswered questions include the timing of migration episodes and their real impact on the European gene pool, the role of physical barriers – as the Mediterranean Sea – and the most plausible dispersal routes for transcontinental crossings. In order to address these topics, different genetic markers have been used. Mitochondrial DNA has shown substantial traces of bidirectional genetic interchange between Africa and Europe, and in a more detailed view between the North of Africa and Iberia. We approached this by analyzing the maternal heritage of the closest Iberian population to North Africa: Andalusia. mtDNA profiles were characterized in two southern Iberian subpopulations, represented by 158 samples from Huelva province and 121 individuals from Granada province. Our results reveal distinctive local histories among Andalusians regarding their maternal legacy, suggesting a role of the westernmost Iberian territory as a noticeable recipient of multiple and diverse human migrations, including relevant African contribution. The African component in western Andalusia was characterized by a high prevalence and diversity of U6 lineage. This finding leads us to further investigate U6 in southern Iberia and northern Africa, by sequencing mitogenomes of 16 U6 Andalusian samples. In order to acquire insight into the other side of the Strait of Gibraltar, we also obtained complete mtDNA sequences from Moroccan Berbers. We then compared these profiles with around 250 U6-genomes reported from literature. The resulting phylogenetic and phylogeographic analyses show that U6 lineage has deep temporal roots in Iberia, revealing old transcontinental human crossings from North Africa to Iberia – and therefore, ancient African traces in Europe.

Key words: *gene flow, migration, haplogroup U6, Mediterranean space, phylogeography, phylogeny*

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NOVEL MODEL OF BIVARIATE ANALYSIS FOR HUMAN GENETIC STUDY. APPLICATION TO MUSCULAR MASS AND METABOLITE LEVEL VARIATION

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We propose a statistical method of bivariate genetic analysis, designed to evaluate contribution of the DNA polymorphisms such as SNP and familial effects (additive genetic, common environment) to variation of two interrelated traits without predefined distribution (i.e. could be quantitative and/or qualitative phenotypes). Our approach is an alternative of the liability-threshold concept (Falconer, 1965), and it is based on the discrete models of genetic and familial effects. In order to take into account additive effect of the other genes on the traits', we introduce three independent binary factors ZX, ZY, and ZXY. In our model they represent genetic factors affecting variation of each trait separately (ZX and ZY) and both traits simultaneously (ZXY), pleiotropic effect. Gene-independent effects, caused by random or common familial effects on the phenotype variation are also taken into account in the model. The model application to analysis muscular mass, metabolomics and genotyping data in a large sample of middle-aged UK female twins is exemplified.

Key words: *muscular mass, metabolomics, GWAS, bivariate analysis, additive genetic and environmental factors*

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