GENOMIC VARIABILITY ACROSS THE ITALIAN POPULATION AND IMPACT ON COMPLEX DISEASES

GIOVANNI FIORITO

ABSTRACT. Population genetics is traditionally a mathematical discipline studying the genetic variation within individuals that involve the examination and modelling of allele frequency changes in populations over space and time. Recent advances in high-throughput technologies allowed the collection of huge genetic datasets for hundreds of individuals worldwide, providing a considerable amount of information useful both to test the robustness of theoretical models and to develop novel hypotheses about human population evolutionary history. In this study, the genetic variability within Italians was investigated. Fine-scale genetic differences were evaluated within the Italian population using a large set of common genetic variants analyzed on a well-selected Italian sample. A continuous pattern of genetic differentiation across the Italian peninsula was shown, the major historical events leading to such variability were traced, and the relationship between genetic and epigenetic profiles in Italians was described. Finally, the main implications for further genetic and epidemiological studies in Italy were discussed.

DIPARTIMENTO DI SCIENZE MEDICHE, UNIVERSITÃ DI TORINO E HUMAN GENETICS FOUNDATION
E-mail address: giovanni.fiorito85@gmail.com