THE GENETIC STRUCTURE OF Y CHROMOSOME IN THE ITALIANS

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The interest of forensic laboratories on Y chromosome SNPs is constantly increasing, since they could be a tool for implementing informativeness in human identification (e.g. mass disasters), paternity tests, and interpretation of mixed male–female samples. Furthermore, their non-random distribution among populations makes them a unique tool for evolutionary and population studies.

As a result of these remarks, the Italian Speaking Group of ISFG, the Ge.F.I. Group, proposed a collaborative exercise with the purpose of verifying the reproducibility and the accuracy of the minisequencing typing method among different laboratories and to collect haplogroup frequencies along the peninsula.

The fifteen participants were asked to analyze the same samples subject of a previous Y-STRs Ge.F.I. collaborative study, close to 1300 regional recruited male subjects. Eighteen markers were selected that allow to explore the basal branches of the phylogenetic tree encompassing all the major clades A-R: M91, M181, M216, M174, M96,M35 M89, M282, M201, M52, M170, M304, M267, M172, M9, M214, M45, M173. Genotyping was accomplished by two multiplex PCRs and subsequent single base extension. Quality assurance comprehended a positive control and two unknown bloodstains.

Spatial frequency distribution of Italian haplogroup-related lineages showed different pattern for the observed clades, and analysis of genetic variability was performed. This project represents the first large local database for both SNPs and STRs. Population and forensic implications of this project were discussed.