

Title: Typing of 20 Y-chromosome STRs in the Italian population.
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Abstract

The typing of Y chromosome is well known in the forensic field above all for its applications in cases of mixed stains of female/male profiles connected to rape.

Currently, commercial kits are available for the determination of the Y-specific haplotype, as the Powerplex Y (Promega) that allows the amplification of 12 STRs in a single reaction. Moreover, several Y-specific multiplex have been described in the literature as that of Butler et al. (FSI 129(2002):10-24) that describing the simultaneous amplification of 20 Y-specific STRs.

The aim of the present study is to appraise the increase of the discrimination power of the Y-specific haplotype allowed by 20 markers including the "extended haplotype" used in Europe DYS19, DYS385, DYS389I/II, DYS390, DYS391, DYS392, DYS393 and YCAII and in addition the STRs DYS437, DYS438, DYS439, DYS447, DYS448, DYS388, DYS426, GATA A7.1 and GATA H4, in a representative sample of the Italian population.

In the past few years, the typing of Y chromosome markers is well known in the forensic field above all for its application in case of mixed stains of female/male profiles connected to rape.

Currently, commercial kits are available for the determination of the Y-specific haplotype, as the PowerPlex Y by Promega that allows the amplification of 12 STRs in a single reaction. In this study, we have used Y-STR 10plex was initially designed from the primer sets shown by Butler et al. [Forensic Sci. Int. 129 (2002) 10-24], two Y-STR, DY437 and DY438 are overlapped. These two multiplexes were tested on 200 males from Italian population sample.

The aim of this study is to appraise the increase of the discrimination power of the Y-specific haplotype allowed by 20 markers in a representative sample of the Italian population.

Anonymous liquid blood samples from 6 areas (North Ovest, North Est, Central, South, Sicily, Sardinia) of the Italy were collected. All samples were tested with 15 autosomal STRs and amelogenin using PowerPlex 16 to demonstrate uniqueness and gender. Sample were quantified and the PCR amplifications for each multiplex were performed using an amount of the male DNA varying between 1 and 2 ng.

After samples are amplified using the PowerPlex Y System, the separation and detection of Y-STR PCR products was accomplished with the ABI Prism 3100 Genetic Analyzer 16-capillary array system following manufacturer's protocols using the G5 matrix filter set. Samples are analyzed using the GeneScan Analysis Software, the sample files are imported into Genotyper Version 3.5 and analyzed using the PowerTyper Y Macro.

A standard reference material (SRM) the human Y-chromosome DNA profiling standard by NIST SRM 2395, that include five male samples and one female sample was utilized for a standardization and reproducibility of the data.

A complete characterization of the allelic frequencies of each marker and diversity of various combinations of these markers is presented in an effort to evaluate their utility for the human identity testing community.

See Presentation Powerpoint Attached