

PROTOTYPE Y CHROMOSOME STANDARD (NIST SRM 2395)

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Y chromosome short tandem repeat markers have a number of applications in human identity testing including typing the perpetrator of sexual assault cases without differential extraction and tracing paternal lineages for missing persons investigations. In an effort to support the growing demand for Y chromosome testing, the National Institute of Standards and Technology (NIST) is developing a standard reference material (SRM) that can be used to calibrate instrumentation and verify assay performance with Y STR and Y SNP markers. Over 150 candidate DNA materials for SRM 2395 have been screened. From these samples, five male and one female DNA samples have been selected and extensively characterized. These candidate materials reflect multiple alleles at each Y STR and SNP locus so that a variety of haplotypes are present. We plan to include 100 ng of each reference sample in the final SRM package and have certified values for ~20 Y STRs and ~10 Y SNPs that have been confirmed by DNA sequencing.

We have performed testing of these candidate reference samples with commercially available and research multiplex assays including a new Y STR 20plex. In addition, all of the DNA samples have been or will be sequenced to confirm exact repeat compositions of the Y STR markers. Five Y SNP markers have been analyzed with multiple technologies to confirm the polymorphic nucleotide present in each sample. These candidate SRM materials have been tested at the following Y chromosome markers: DYS19, DYS385, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS426, DYS435, DYS436, DYS437, DYS438, DYS439, DYS447, DYS448, GATA A7.1, GATA H4, and YCAII.