HUMAN MITOCHONDRIAL DNA - AMPLIFICATION AND SEQUENCING STANDARD REFERENCE MATERIALS – SRM 2392 (UPDATED) AND SRM 2392-I (NEW) – FOR FORENSIC AND MEDICAL USE

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Forensic and clinical laboratories benefit from NIST DNA Standard Reference Materials (SRMs) that provide the quality control and assurance that laboratories are performing their DNA amplification and sequencing accurately and that the DNA sequences obtained from unknown samples are correct. The identification of unknown human remains by mitochondrial DNA (mtDNA) testing is an important program within the law enforcement community. MtDNA is especially important when the amount of nuclear DNA is insufficient or degraded (e.g., in burn victims following mass disasters such as the attack on the World Trade Center). MtDNA has the advantages that less material is needed for human identification and that maternal relatives can be used to verify the identification. The medical community also needs mtDNA SRMs to serve as controls in the verification of the genetic basis of mtDNA diseases. To fulfill the needs of these two communities, NIST has developed and recently released the human mtDNA amplification and sequencing SRM 2392-I to complement SRM 2392 that has been available since 1999 (1,2). Both SRM 2392-I and SRM 2392 are currently available from the Office of Standard Reference Materials at NIST and provide quality control for forensic identification, medical diagnosis, and mutation detection. They contain all the information (e.g., the sequences of 58 unique primer sets) needed to use these SRMs as positive controls for the amplification and sequencing of any DNA. SRM 2392-I contains the DNA from HL-60, a promyelocytic leukemia cell line, whereas, SRM 2392 includes the DNA from two apparently healthy individuals (CHR and 9947A). Three laboratories participating in an interlaboratory evaluation to determine the sequence of the entire HL-60 mtDNA (16,569 bp) found the identical sequence as that determined by NIST. The Office of Standard Reference Materials decided that the HL-60 DNA should be provided as a separate SRM (2392-I) rather than as part of SRM 2392. The certificate of analysis (which accompanies SRM 2392-I) (3), a journal article (4) and a NIST special publication (5) have been written. Both these mtDNA SRMs will provide enhanced quality control when examining mtDNA for forensic identification, medical diagnosis, and single nucleotide polymorphism (SNP) detection.

1. Levin, B.C., Cheng, H., and Reeder, D.J. 1999. A human mitochondrial DNA Standard Reference Material for quality control in forensic identification, medical diagnosis, and mutation detection. Genomics 55:135-146.
2. Levin, B.C., Cheng, H., Kline, M.C., Redman, J.C., and Richie, K.L. 2001. A review of the DNA Standard Reference Materials developed by the National Institute of Standards and Technology. Fresenius J. Anal. Chem. 370:213-219.
3. http://ts.nist.gov/ts/htdocs/230/232/232.htm
4. Levin, B.C., Holland, K.A., Hancock, D.K., Coble, M., Parsons, T.J., Kienker, L.J., Williams, D.W., Jones, MP, and Richie, K.L. 2003. Comparison of the complete mtDNA genome sequences of human cell lines - HL-60 and GM10742A - from individuals with Pro-Myelocytic Leukemia and Leber Hereditary Optic Neuropathy, respectively, and the inclusion of HL-60 in the NIST human mitochondrial DNA Standard Reference Material - SRM 2392-I. Mitochondrion 2:386-399.
5. Levin, B.C., Hancock, D.K., Holland, K.A., Cheng, H., and Richie, K.L. 2003. Human mitochondrial DNA - amplification and sequencing Standard Reference Materials – SRM 2392 and SRM 2392-I. NIST SP 260-155, National Institute of Standards & Technology, Gaithersburg, MD.

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