

## PROTOCOLS FOR FORENSIC MITOCHONDRIAL DNA ANALYSIS

References		
Status: Published		Document ID: 1167
DATE EFFECTIVE 08/06/2021	APPROVED BY mtDNA Technical Leader	PAGE 1 OF 3

# References

### DNA Sequencing and Analysis

Anderson S, Bankier AT, Barrell BG, de Bruijn MH, Coulson AR, Drouin J, Eperon IC, Nierlich DP, Roe BA, Sanger F, Schreier PH, Smith AJ, Staden R, Young IG. Sequence and organization of the human mitochondrial genome Nature 1981; 290: 457-465. MEDLINE- 81173052 PUBMED-7219534

Andrews RM, Kubacka I, Chinnery PF, Lightowlers RN, Turnbull DM, Howell N. Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. Nat. Genet. 1999; 23 (2): 147. MEDLINE- 99438386 PUBMED- 10508508.

Applied Biosystems. 3130/3130xl Genetic Analyzers Getting Started Guide. 2004. Part Number 4352715 Rev. B. Applied Biosystems, Foster City, CA.

Applied Biosystems. 3130/3130xl Genetic Analyzers Maintenance, Troubleshooting, and Reference Guide. 2004. Part Number 4352716 Rev. B. Applied Biosystems, Foster City, CA.

Applied Biosystems. DNA Sequencing Analysis Software User Guide: Version 5.1 for Windows® XP and 2000 Platforms. 2003. Part Number 4346366 Rev. B. Applied Biosystems, Foster City, CA.

Budowle B, Fisher C, Polanskey D, Hartog BD, Kepler R, Elling J. Standardizing the Nomenclature for mtDNA Haplotypes with an Intuitive Hierarchical Execution Software Program. ISFG Conference. 2007.

Budowle B, Wilson MR, DiZinno JA, Stauffer C, Fasano MA, Holland MM, Monson KL. Mitochondrial regions HVI and HVII population data. Forensic Sci Int 1999; 103: 23-35.

Butler, J.M. Advanced Topics in Forensic DNA Typing: Interpretation. Academic Press. 2015. ISBN: 978-0-12-405213-0.

Carracedo A, Baer W, Lincoln P, Mayr W, Morling N, Olaisen B, Schneider P, Budowle B, Brinkmann B, Gill P, Holland MM, Tully G, Wilson MR. DNA commission of the International Society for Forensic Genetics: Guidelines for mitochondrial DNA typing. Forensic Sci Int 2000; 110: 79-85.

Clopper, CJ, and Pearson, ES. The use of confidence or fiducial limits illustrated in the case of the binomial. Biometrika 1934; 26, 404-413.

Controlled versions of Department of Forensic Biology Manuals only exist in the Forensic Biology Qualtrax software. All printed versions are non-controlled copies.

© NYC OFFICE OF CHIEF MEDICAL EXAMINER

## PROTOCOLS FOR FORENSIC MITOCHONDRIAL DNA ANALYSIS

References		
Status: Published		Document ID: 1167
DATE EFFECTIVE 08/06/2021	APPROVED BY mtDNA Technical Leader	PAGE 2 OF 3

Gene Codes. Sequencher™ 4.0 User Manual. 1999. Gene Codes Corporation. Ann Arbor, MI.

Holland MM, Parsons TJ. Mitochondrial DNA sequence analysis - Validation and use for forensic casework. Forensic Science Review 1999; 11: 21-50.

Parson W, Dur A. EMPOP – A forensic mtDNA database. FSI Genetics. 2007; 1(2): 88-92.

Sanger FS, Nilken S, Coulson AR. Sequencing with chain terminating inhibitors. Proc Nat Acad Sci U.S.A. 1977; 74: 5463-5467.

Scientific Working Group on DNA Analysis Methods (SWGDM). Guidelines for mitochondrial DNA (mtDNA) nucleotide sequence interpretation, vol. 5, no.2. Forensic Science Communications 2003. <http://www.fbi.gov/about-us/lab/forensic-science-communications/fsc/april2003/swgdammitodna.htm/>

SWGDM. Interpretation guidelines for mitochondrial DNA analysis by forensic DNA testing laboratories. 2013. Available at [https://1ecb9588-ea6f-4feb-971a-73265dbf079c.filesusr.com/ugd/4344b0\\_f61de6abf3b94c52b28139bff600ae98.pdf](https://1ecb9588-ea6f-4feb-971a-73265dbf079c.filesusr.com/ugd/4344b0_f61de6abf3b94c52b28139bff600ae98.pdf)

Stewart JEB, Aagaard PJ, Pokorak EG, Polanskey D, Budowle B. Evaluation of a multicapillary electrophoresis instrument for mitochondrial DNA typing. J Forensic Sci 2003; 48: 571-580.

Wilson MR, DiZinno JA, Polanskey D, Replogle J, Budowle B. Validation of mitochondrial DNA sequencing for forensic casework analysis. Int J Leg Med 1995; 108: 68-74.

Wilson MR, Allard MW, Monson KL, Miller KWP, Budowle B. Recommendations for consistent treatment of length variants in the human mitochondrial DNA control region. Forensic Sci Int 2002; 129: 35-42.

Wilson, MR., Allard, MW, Monson, KL, Miller KWP, Budowle B. Further discussion of the consistent treatment of length variants in the human mitochondrial DNA control region, vol. 4, no.4. Forensic Science Communications 2002. <http://www.fbi.gov/about-us/lab/forensic-science-communications/fsc/oct2002/wilson.htm/>

## PROTOCOLS FOR FORENSIC MITOCHONDRIAL DNA ANALYSIS

References		
Status: Published		Document ID: 1167
DATE EFFECTIVE 08/06/2021	APPROVED BY mtDNA Technical Leader	PAGE 3 OF 3

### ExoSAP-IT (PCR Product Clean-up)

Dugan, KA, Lawrence, HS, Hares, DR, Fisher, CL, Budowle, B. An improved method for post-PCR purification for mtDNA sequence analysis. J Forensic Sci 2002; 47:811-18.

### Agilent

Mueller, O, Hahnenberger K, Dittmann M, Yee H, Dubrow R, Nagle R, Ilsley D. A microfluidic system for high-speed reproducible DNA sizing and quantitation. Electrophoresis 2000; 21:128-134.

Bjerketorp J, Chiang ANT, Hjort K, Rosenquist M, Liu W-T, Jansson JK. Rapid lab-on-a-chip profiling of human gut bacteria. J. Microbiol. Methods 2008; 72: 82-90.