Scientific Working Group on DNA Analysis Methods

Mitochondrial DNA Nomenclature Examples Document

Additional Guidance for the Application of the SWGDAM Interpretation Guidelines for Mitochondrial DNA Analysis for Forensic DNA Testing Laboratories.

The Scientific Working Group on DNA Analysis Methods, better known by its acronym of SWGDAM, is a group of approximately 50 scientists representing federal, state, and local forensic DNA laboratories in the United States and Canada. During meetings, which are held twice a year, subcommittees discuss topics of interest to the forensic DNA community and often develop documents to provide direction and guidance for the community. This version was presented to the Executive Board of SWGDAM and received approval on May 5, 2014.

This guidance document contains examples of the application of the SWGDAM Nomenclature Rules described in Section 2.3 of the SWGDAM Interpretation Guidelines for Mitochondrial DNA (mtDNA) Analysis for Forensic DNA Testing Laboratories. The list of examples is not intended to be all inclusive, and focuses on the application of the nomenclature rules to challenging mtDNA sequences obtained from forensic and population database samples. If the listed examples do not address a particular sequence of interest, practitioners are encouraged to consult Section 2.3 of the aforementioned

guidelines document and the public EDNAP mtDNA Population Database (EMPOP) (http://empop.org/) website to help determine the proper mtDNA nomenclature.

The listed examples are organized by the range of the nucleotide positions for the observed DNA sequence, the corresponding sequence of the revised Cambridge Reference Sequence (rCRS) standard for the given range, the determined mtDNA haplotype by applying the SWGDAM Nomenclature Rules, and the determined mtDNA haplotype from the superseded Rule Based approach.

1.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCC-CCATG		
Sample	AAAACCCCCCCCCTCATG	16189C 16191.1C 16192T	16189C 16192.1T

2.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCC-CCATG		
Sample	AAAACCCCTCCCCTCATG	16188T 16189C 16191.1C 16192T	16187.1T 16189C 16192T

3.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCCATG		
Sample	AACCCCCCTCCCCCATG	16182C 16183C 16188T 16189C	16182DEL 16183C 16193.1C

4.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCCATG		
Sample	AAAACTCCCCCCC-ATG	16185T 16189C 16193DEL	16185T 16189DEL

5.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCCATG		
Sample	AAAACCTCCCCCC-ATG	16186T 16189C 16193DEL	16186T 16189DEL

6.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCC-ATG		
Sample	AAACACCCCCCCCCCATG	16183C 16184A 16189C 16193.1C	16182.1C 16189C

7.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCCATG		
Sample	AAACACCCCCCCCCATG	16183C 16184A 16189C	16182.1C 16189DEL

8.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCC--ATG		
Sample	AAAACCCCCCTCCCCCATG	16189C 16190T 16193.1C 16193.2C	16188.1C 16193.1C

9.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCC-ATG		
Sample	AAACCCCCTCCCCCCATG	16183C 16188T 16189C 16193.1C	16183DEL 16193.1C 16193.2C

10.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCC-ATG		
Sample	AAAACCCCCCCTCCCATG	16189C 16191T 16193.1C	16189C 16190.1T

11.	Range: np 16180-16196	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AAAACCCCCTCCCCATG		
Sample	AAAACCCCCCCCC-ATG	16189C 16193DEL	16189DEL

12.	Range: np 55-68	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	TATTTT-CGTCTGGG		
Sample	CACTTTTCGTCTGGG	55C 57C 60.1T	55C 56.1C

13.	Range: np 55-68	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	TATTTT-CGTCTGGG		
Sample	TATCTTTCGTTTGGG	58C 60.1T 64T	57.1C 64T

14.	Range: np 55-72	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	T-ATTTTCGTCT-GGGGGGT		
Sample	TTATT--CGTCTTTGGGGGT	55.1T 59DEL 60DEL 65.1T 66T	56DEL 58A 65.1T 66T

15.	Range: np 240-255	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	AACAATTGAATGTCTG		
Sample	AACAATTAA-TGTCTG	247A 249DEL	247DEL

16.	Range: np 302-317	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	ACCCCCCCTCCCCCGC		
Sample	ACCCCCCCCCCCC-GC	310C 315DEL	310DEL

17.	Range: np 453-465	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	TTTCCCCTCCCAC		
Sample	TCCCCCCCC--AC	454C 455C 460C 462- 463-	454DEL 455DEL 460C

18.	Range: np 570-580	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	CCCC-ACAGTTT		
Sample	CCCCCCCAGTTT	573.1C 574C	574C 575.1C

19.	Range: np 570-580	SWGDAM Nomenclature Rules	Superseded Rule Based Approach
rCRS	CCCCACAGTTT		
Sample	CCC-CCAGTTT	573DEL 574C	574DEL

Document Version	Revision History
August 2013	Approved by the SWGDAM Executive Board on August 7, 2013.
May 2014	Revisions approved by the SWGDAM Executive Board on May 5, 2014, that included the following: correction of typographical errors in the sequences and provided nomenclature, elimination of a duplicate example, change of font for sequences to align, and correction of the cited revised Cambridge Reference Sequence (rCRS).