B50 Implementation of CODIS^{mt}: The National Mitochondrial DNA Database

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The goal of this presentation is to introduce CODIS^{mt}, the first national mitochondrial DNA database, to the forensic DNA community Databanks that contain DNA profiles of convicted felons, missing persons, and/or profiles from evidence from cases are useful for providing investigative leads for resolving certain crimes. Use of the COmbined DNA Index System (CODIS) can assist in resolving crimes, particularly violent crimes, and prevent further crimes by quickly identifying the perpetrator. CODIS^{mt} enables federal, state, and local crime laboratories to exchange and compare DNA profiles electronically, thereby linking crimes to each other and to convicted offenders.

The use of CODIS^{mt} was expanded to include mitochondrial DNA (mtDNA) profile searching software, a missing persons index, and a relatives of missing persons index once authorized by federal missing persons legislation. The new software, known as CODIS^{mt}, facilitates the searching of mtDNA nucleotide sequences developed from evidentiary samples against one or more sequence database(s) and provides a population database index for statistical applications.

In order to ensure an effective system, all profile identification numbers and the designation of genetic variants have been standardized across participating laboratories in order to facilitate profile searching at the national level. In addition, minimum requirements for the processing of controls, reporting of polymorphisms, and submitting of sequence data have been established.

Laboratories participating in the National DNA Index System (NDIS) must establish evaluation criteria for the use of controls in their analyses, including but not limited to a positive control, a negative control, and a reagent blank control. These control samples must be processed through nucleotide sequencing as is done with corresponding questioned or known samples. DNA purified from the HL60 cell line is the NDIS-accepted positive control. If the reagent blank and/or the negative control of a particular amplification yield a sequence that is the same as that of the sample, or if contamination is present above the threshold set by laboratory, then the data will not be acceptable at NDIS. CODIS^{mt} archives profiles as differences from the revised Cambridge Reference Sequence (rCRS) according to the nucleotide position and the DNA base difference from the reference (e.g., 16089 C). There are no minimum length requirements for nucleotide sequence data obtained from questioned (Q) samples, but nucleotide sequence from known (K) samples should include both hypervariable region 1 (HV1; nucleotide positions 16024-16365) and hypervariable region 2 (HV2; nucleotide positions 73-340) whenever possible. Nucleotide sequence obtained from population database samples must include a minimum of HV1 and HV2, and must not contain over 1% ambiguity over the length of the sequence. Both strands of the amplified product must be
sequenced for one to reduce ambiguities in a sequence region.

Laboratories must develop guidelines for evaluation of cases and for the presence of heteroplasmy. At NDIS, mtDNA profiles are considered candidate matches if there are two or fewer differences between the profiles in question. Differences between sequences caused by length heteroplasmy are not considered when determining a candidate match. The mtDNA committee of the Scientific Working Group on DNA Analysis Methods (SWGDAM) has proposed the following interpretation guidelines that can be used in most cases:

Exclusion: If there are two or more nucleotide differences between the questioned and known samples, the samples can be excluded as originating from the same person or maternal lineage.

Inconclusive: If there is one nucleotide difference between the questioned and known samples, and no additional data are available (i.e., no other samples have been typed), the result will be inconclusive.

Cannot Exclude: If the sequences from questioned and known samples under comparison have a common base at each position or a common length variant in the HV2 C-stretch, the samples cannot be excluded as originating from the same person or maternal lineage.

Mitochondrial DNA, CODIS, Database

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