



SWGDM¹ Considerations for Claims that the CODIS Core Loci are ‘Associated’ with Medical Conditions/Diseases

In a June 2012 ruling, *State v. Abernathy*, No. 3599-9-11, a Vermont Court adopted the testimony from a defense expert on the CODIS core loci and found that “the analogy between DNA testing and fingerprinting is no longer valid, because a DNA profile consisting of the thirteen CODIS loci contains information beyond mere identity.” In reaching that conclusion, the Court found “by a preponderance of the credible scientific evidence... that five of the thirteen CODIS loci contain genetic information associated with at [sic] eight different medical conditions; ...that there are genetic functions or physical traits associated with one or more of the CODIS loci; and ...that the CODIS loci employed in DNA profiling are not “junk DNA” devoid of significant personal information.” In overturning the Vermont DNA sample collection provisions at arraignment, the Court held that the “mandatory seizure and search of genetic material that contains personal genetic information beyond identity implicates the privacy concerns at the core of Article 11 [Vermont Constitution].”

One point that was not addressed in the *Abernathy* discussion is the fact that Vermont, along with many other states, expressly delineates the authorized purposes for the arrestee/offender DNA samples. For example, Vermont law authorizes DNA analysis for “the genetic markers from DNA samples for law enforcement identification purposes” and that “analysis of DNA samples obtained pursuant to this subchapter is **not** authorized for identification of any medical or genetic disorder.” See 20 V.S. A. § 1937 (a)(1), (b) (emphasis added); see also 42 U.S.C. §14133(c).

Assertions that the CODIS core loci could reveal information relating to medical conditions or diseases are not new. In fact, this was the subject of a colloquy between Professors Simon Cole and David Kaye several years ago. [S. A. Cole, *Is the “Junk” DNA Designation Bunk?* 102 NW. U. L. Rev. Colloquy 54 (2007) at <http://www.law.northwestern.edu/lawreview/colloquy/2007/23/>; D.H. Kaye, *Please,*

¹ On January 17, 2013, the SWGDAM membership unanimously approved the Open Letter previously posted by the SWGDAM Executive Board; this approval included technical revisions to update Internet references and publication citations. The original Open Letter approved by the SWGDAM Executive Board on September 17, 2012 is available under SWGDAM Communications at <http://www.swgdam.org/docs.html> .



Let's Bury the Junk: The CODIS Loci and the Revelation of Private Information, 102 NW. U. L. Rev. Colloquy 70 (2007) available at <http://www.law.northwestern.edu/lawreview/colloquy/2007/25/>.] More detailed information relating to the CODIS Core STR Loci are described in Butler, J.M. *Genetics and Genomics of Core STR Loci Used in Human Identity Testing*, J. Forensic Sci. 51(2):253-65 (March 2006) at <http://onlinelibrary.wiley.com/doi/10.1111/j.1556-4029.2006.00046.x/full>.

More recently, news articles have announced significant findings in connection with the Encyclopedia of DNA Elements or ENCODE Project in which areas of the “dark matter” DNA or what has been referred to as “junk DNA” modulate human genes. See G. Kolata, *Bits of Mystery DNA, Far From ‘Junk,’ Play Crucial Role*, N.Y. Times, September 5, 2012. While numerous scientific articles are and will be published on these discoveries, in the interim we offer the following considerations relating specifically to the CODIS core loci:

1. The 13 CODIS core loci were specifically chosen because of their location in regions commonly referred to as “junk DNA” and the assessment that these loci were not causative or indicative of any medical condition or disease status. See Wagner, J.K. (2012), *Out with the “Junk DNA” Phrase*. J. Forensic Sci, January 2013, Vol. 58, No. 1; doi: 10.1111/j.1556-4029.2012.02252.x [noting that “it is appropriate to encourage the discontinued characterization that CODIS loci are ‘junk DNA’” and “it is also appropriate to warn nonscientists that to imply the CODIS loci are each or collectively involved in gene expression and are now important for a wide array of traits and conditions of biomedical relevance is unfounded.”]
2. The 13 CODIS core loci were selected as law enforcement markers to be used for identification purposes only. A numeric designation is given to the two alleles at each locus. This series of numbers at the 26 alleles are what comprise the DNA profile that is stored and searched in the law enforcement DNA databases. For example, the following set of numbers represents the allelic information in a DNA profile:
11,11,12,12,12,13,10,20,29,30,16,18,12,13,09,11,11,15,23,24,06,9,3,08,08,17,19. See generally, *Frequently Asked Questions (FAQs) on the CODIS Program*



and the National DNA Index System, available at <http://www.fbi.gov/about-us/lab/biometric-analysis/codis/codis-and-ndis-fact-sheet>.

3. In designing the CODIS software, a deliberate decision was made not to include any personal identifiers relating to the subject of the DNA. Accordingly, there are no personal identifiers, such as the name or Social Security number of the individual who provided the DNA sample that are stored and searched as part of the DNA record in the law enforcement DNA databases. A specimen identification number similar to an accession number and information identifying the laboratory and analyst responsible for the DNA profile are the other data elements stored with the DNA profile that together make up what we refer to as the DNA record. See *Frequently Asked Questions (FAQs) on the CODIS Program and the National DNA Index System*, available at <http://www.fbi.gov/about-us/lab/biometric-analysis/codis/codis-and-ndis-fact-sheet>.
4. None of the 13 CODIS core STRs is located in a gene or protein coding region. See Wagner, J.K. (2012), *Out with the "Junk DNA" Phrase*. J. Forensic Sci, January 2013, Vol. 58, No. 1; doi: 10.1111/j.1556-4029.2012.02252.x; Katsanis, S.H., et al., *Characterization of the Standard and Recommended CODIS Markers*, J. Forensic Sci, January 2013, Vol. 58, No. S1; doi:10.1111/j.1556-4029.2012.02253.x; Butler, J.M. *Genetics and Genomics of Core STR Loci Used in Human Identity Testing*, J. Forensic Sci. 51(2):253-65 (March 2006) at <http://onlinelibrary.wiley.com/doi/10.1111/j.1556-4029.2006.00046.x/full>; see also Benecke, *Coding or Non-Coding, That is The Question* (2002) 3 EMBO Reports No.6, pp. 498-502 ["Abuse of such information is impossible because it simply has no practical predictive value."].
5. A reported correlation between a CODIS marker and a coding gene for a specific medical condition or disease does not mean that the CODIS marker is now linked to that medical condition. More information concerning the study reporting that correlation would be necessary to determine if this was within a family group and if the marker was being used to track a medical condition within that family. For example, as was reported in one of the scientific articles



referenced in the *Abernathy* case, *Familial Alzheimer's disease (FAD): co-segregation between alleles at the D21S11 DNA marker and the FAD gene in a particular pedigree*, F. David et al., *J. Neurology* (1988) 235:485-486 "in the same family, co-segregation between a polymorphic allele of probe D21S11 and the FAD gene, suggesting that such a linked DNA marker, might be used for pre-symptomatic diagnosis in this particular pedigree." This report concludes that "before these individuals can be offered an informed choice, it will be necessary to confirm the results obtained with additional probes from the same chromosomal region, and to define recombination frequencies between these markers and the FAD gene." See also California Department of Justice, Bureau of Forensic Services, *Proposition 69 (DNA) FAQs*, at <http://oag.ca.gov/bfs/prop69/faqs> under "Searching the CAL-DNA Data Bank and CODIS" [CODIS core loci "may be used to identify locations in the human genome that contain disease-causing genes," but "this use is limited to family lineages, and requires detailed medical information about subjects in conjunction with these STR test results. Specifically, when STR markers are used in this manner, it requires a comparison of the STR types of at least several known family members along with knowledge of their disease status."]; Katsanis, S.H., et al., *Characterization of the Standard and Recommended CODIS Markers*, *J. Forensic Sci.*, January 2013, Vol. 58, No. S1; doi:10.1111/j.1556-4029.2012.02253.x/abstract ["...we found no documentation of individual genotypes for the 24 STRs (current and recommended CODIS loci) to be causative of any documented phenotypes either in the literature or in the interrogated databases."]; Butler, J.M. *Genetics and Genomics of Core STR Loci Used in Human Identity Testing*, *J. Forensic Sci.* 51(2):253-65(March 2006) at <http://onlinelibrary.wiley.com/doi/10.1111/j.1556-4029.2006.00046.x/full> [noting that "in fact, many times these linkage "findings" can later be proven false with further studies, such as with TH01"].

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