B156 Case Study of 'Deleted-Amelogenin' Males Within the WTC DNA Identification Project

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After attending this presentation, attendees will be informed of the "amelogenin deletion" phenomenon (known to occur in certain Asian populations) which has been confirmed in the World Trade Center victim population.

This presentation will impact the forensic community and/or humanity by raising awareness in the forensic DNA community of "amelogenin deletion," and explain how this phenomenon was dealt with during the WTC project in order to make a positive identification of at least one victim.

Due to a known genetic mutation on the Y chromosome in which the amelogenin locus is deleted (1, 2), STR-typing kits such as Cofiler, Profiler Plus, and PowerPlex™ 16 only detect the copy of the allele which originates on the X chromosome. As a result, the genotypes appear to be female, unless shown to be otherwise by use of Y-STR typing. This rare "Y-deletion" is found most often in males from the areas between Southeast Asia and the Indian subcontinent.

The World Trade Center disaster included many persons whose ancestry can be traced to that region of the world, and the presence of Ychromosome anomaly has been confirmed during the identification process for one victim, and is suspected in another. The latter was identified by dental records (his body was intact enough at recovery to still be wearing masculine clothing, but gave a "female" profile in autosomal STRs. Thus, for the ~ 2800 reported missing (about three quarters of which are male), the prospect of observing this twice is somewhat remarkable.

During DNA testing for the World Trade Center identification project, two human remains fragments (both are pieces of bones) found at the site matched a personal effect submitted by a male victim's family. The match was initially thought to be a "problem case" such as comingling (two persons' remains, and therefore DNA as well) or a sample mixup (for example, another family member's toothbrush submitted by mistake as an exemplar for their missing loved one), because the personal sample for this missing man came up with a female profile during initial testing (Cofiler and Profiler Plus). In addition, several members of his family submitted buccal swabs for use in kinship analysis, including his father and a full brother. It was found that all of this family's samples appeared to be female, but in all other loci tested, the correct allele inheritance patterns indicated that there likely wasn't a mixup of the swabs. These results bolstered our suspicion that the Y chromosome in that paternal line was "deleted-amelogenin."

Additional testing using Y-STRs (OCME's YM1 panel, consisting of DYS 19, DYS 389 I and II, DYS 390) confirmed an identical Y haplotype in the father's and brother's buccal swabs and the victim's toothbrush. That result confirmed our suspicion of the Y-chromosome mutation's presence in this family; and taken together, all results were enough to declare a positive DNA identification of this WTC victim.

Laboratories performing DNA typing involving victims, suspects, or missing persons (specifically, males) from southern and eastern Asia must be aware of the possibility of a situation like the above arising, not only in identifications when kinship samples are also available to confirm the phenomenon, but also during criminal casework such as homicides and sexual assaults when an unexpected additional female profile is obtained. Additional, Y-chromosome-specific, testing can be performed to help solve these cases.

1. Santos FR, Pandya A, Tyler-Smith C. Reliability of DNA based sex tests. Nat. Genet. 1998; 18:103.
2. Roffey PE, Eckhoff CI, Kuhl JL. A rare mutation in the amelogenin gene and its potential investigative ramnifications. J. Forensic Sci. 2000; 45:1016-1019.

Y-Deletion, World Trade Center, Amelogenin

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