

**POLYMORPHIC VARIATION BETWEEN FULL SIBLINGS IN THE MITOCHONDRIAL  
DNA HV2 REGION C-STRETCH**

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At the Armed Forces DNA Identification Laboratory (AFDIL) our mission is to support the Central Identification Laboratory, Hawaii (CILHI) in identifying the skeletal remains from missing and unknown personnel from the Vietnam War, Korean War, Cold War, and World War II. We also support the mission of the Office of the Armed Forces Medical Examiner (OAFME) in identifying remains from mass disasters and recent military deaths. To this end we use DNA testing.

There are two different types of DNA that can be used for the identification of individuals: nuclear and mitochondrial. We primarily use the sequencing of mitochondrial DNA (mtDNA) for our work with CILHI. This is due to the low quality of the nuclear DNA in the source material, and the advantage of mtDNA that it has maternally inherited. When we are setting out to identify an individual, we receive a fragment of skeletal remains from CILHI. Analysts extract, amplify, and sequence the mtDNA obtained from this bone. This sequence is compared to a published standard sequence (Anderson) in order to determine the number of differences, or polymorphisms, from this standard sequence. In conjunction with the processing of the unknown sample, we also receive numerous blood reference samples from family members of missing service members. These blood samples are extracted, amplified and sequenced in a manner similar to the treatment of the skeletal remains, but by a set of scientists dedicated specifically to the treatment of such family references. Once a sequence is obtained from a reference, it too is compared to polymorphism from skeletal remains to identify the missing individuals. If the reference was obtained from a maternal relative of the questioned sample, the sequence from the bones should be consistent with the sequence from the blood.

In the analysis of the mtDNA, we look at two specific regions: the HV1 region (positions 16024-16365) and the HV2 region (positions 73-340). For this poster we will be focusing on the HV2 region. This region contains a sequence of base pairs from positions 303-315 that is a series of seven C's interrupted by a single T and then followed by another 4 C's. This is typically called the C-stretch region and is the cause of much difficulty in sequencing since it tends to be highly polymorphic. There are often one or more C's inserted before and after the T.

At AFDIL, we do not use polymorphism within the C-stretch to differentiate between individuals. This poster will present three family references from full-siblings that share the same polymorphisms in both the HV1 and HV2 regions, except with the C-stretch, where they vary significantly; thus indicating that the C-stretch region may not be a rigorous enough diagnostic tool with which to identify individuals.