

DNA TESTING

FACT SHEET

DNA testing is one of the primary scientific techniques used for human identification. The National DNA Program for Unidentified and Missing Persons will use DNA testing to link unidentified and missing persons cases. The success of this DNA-led identification approach will be dependent on:

- ▶ **The use of contemporary DNA techniques to aid the recovery and profiling of DNA from old and degraded unidentified human remains (UHR)**
- ▶ **The profiling of suitable reference DNA samples collected for each long-term missing person or from their relatives for comparison to the DNA recovered from the UHR**

DNA Testing

The Program will utilise a number of techniques for DNA identification. A description of the DNA testing process is provided below.

DNA:

Deoxyribonucleic acid (DNA) is a chemical substance present in almost every cell of the human body. It is considered our genetic blueprint as it contains all of the instructions necessary for life. This genetic code is inherited from one generation to the next and is unique to each person (except identical twins). There are two different types of DNA in a cell: nuclear DNA (nDNA) found in the nucleus of the cell, which contains both autosomal DNA (atDNA) and Y-chromosome DNA (Y-DNA), and mitochondrial DNA (mtDNA) found in the mitochondria of the cell.

Nuclear DNA:

atDNA provides information about both the paternal and maternal lineage as it is inherited equally from both parents, with atDNA profiles of close relatives being more similar than distant relatives. In male or female missing person cases, atDNA is used to match UHR against close relatives such as a mother, father and/or children. Y-DNA provides information about the paternal lineage and is transferred directly from father to son. Therefore, paternal relatives will have the same Y-DNA profile. In male missing person cases, Y-DNA is used to match UHR against any paternal relative such as a father, grandfather, brother or son.

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Mitochondrial DNA:

mtDNA provides information about the maternal lineage and is transferred directly from mother to child (both male and female children). Therefore, maternal relatives will have the same mtDNA profile. In male or female missing person cases, mtDNA is used to match UHR against any maternal relative such as a mother, grandmother, sister or brother (or children if the missing person is female). Additionally, mtDNA is the most sensitive DNA marker, and may provide DNA results for old and degraded samples when nDNA profiling is unsuccessful.

DNA Identification:

UHR are examined to collect an optimal bone or tooth sample for DNA testing. In many cases, laboratory techniques can then be used to extract DNA from the sample and analyse specific DNA markers to develop nDNA and/or mtDNA profiles for comparison or upload on to state, national and international DNA databases. To identify the UHR, the DNA profile from the sample must be matched to a DNA profile from the missing person (direct reference sample) or their relative/s (family reference sample). Alternatively, a secure database of DNA profiles from missing persons, missing persons relatives and other relevant categories can be used for direct and/or kinship searching to find DNA matches.

Direct Reference Sample:

A direct reference sample is a DNA sample collected from a missing person's medical specimen (e.g. newborn screening card, clinical biopsy sample) or personal item (e.g. toothbrush, razor). Medical specimens are preferred if available as documentation can be used to verify the sample origin, with newborn screening cards being considered the gold standard. Additionally, for historical missing persons cases personal items are unlikely to be available. If personal items are the only direct reference sample available, the DNA profile produced can be compared to family reference samples if available to confirm the sample is attributable to the missing person.

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Family Reference Sample:

A family reference sample is a volunteer DNA sample collected from a suitable biological relative of the missing person. Samples from at least two biological relatives should be collected for DNA testing if available. The most informative samples are from close biological relatives such as parents, children or siblings. Ideally, one sample should be a maternal relative (e.g. mother) to facilitate mtDNA testing and the other should be a paternal relative (e.g. father) to facilitate Y-DNA testing. For historical missing persons cases, samples from distant maternal or paternal relatives may only be available and in these scenarios mtDNA or Y-DNA testing will be useful. In missing parent cases, it is valuable to collect a reference sample from the missing person's spouse (or parent-in-common) if the missing person's biological child/ren have provided family reference sample/s.

Direct Matching:

DNA profiles from UHR can be directly compared in a pairwise search to DNA profiles from direct reference samples. If a direct match is obtained, it provides probabilistic support for inferring that the UHR (i.e. bone sample) and the donor of the direct reference sample (i.e. saliva sample) are the same person. This is possible because an individual's DNA is the same in every cell of the body.

Kinship Matching:

When a DNA database search does not result in any direct matches, DNA profiles from UHR can be indirectly compared in either a pairwise or pedigree search to DNA profiles from one or more family reference samples, respectively. If a kinship match is obtained, it provides probabilistic support for inferring that the UHR and the donor of the family reference sample are genetically related. This is possible because relatives share similar and predictable patterns in their DNA profiles. For example, a child's DNA profile is a combination of atDNA markers inherited equally from the mother and father. Additionally, Y-DNA and mtDNA markers can be useful to support a biological relationship between UHR and putative relative/s.

Further Information

For more information on the Program please go to our dedicated webpage: www.missingpersons.gov.au/national-dna-program-unidentified-and-missing-persons or contact the NMPCC: missing@afp.gov.au.



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