

WHAT MITOCHONDRIAL DNA CAN AND CAN'T DO IN PARENTAGE TESTING

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Mitochondrial DNA has unique features that make it an excellent alternative to nuclear DNA testing in certain circumstances. The high copy number of mitochondrial DNA molecules in each cell allow typing where sample is limited or degraded. Particularly in missing persons cases or “no-body” homicides, where skeletal remains, telogen hairs, or even blood samples are available that might have been contributed by the missing individual, mitochondrial DNA typing provides a direct link to any single maternally related family member of that individual. For this reason, mitochondrial DNA testing in these cases effectively becomes “parentage” or “kinship” testing. The biological tissues of individuals many generations removed from each other, as long as they are connected by a female lineage, would be expected to have the same type of mitochondrial DNA. In deficiency cases where parents are not available, there would be no need to type multiple family members as would be done in nuclear DNA testing to generate confidence in the genotype match. In some situations, it is possible to avoid exhumations or otherwise difficult typings by seeking out alternative living, though more distantly related, maternal relatives when one wishes to confirm maternal relatedness.

While mitochondrial DNA is useful for identifications and to evaluate maternal relatedness, it cannot play any role in establishing paternity. However, when paternity can otherwise be confirmed by standard methods, typing the mitochondrial DNA of the father of an individual may, in a rare case, be helpful in evaluating ethnicity, for example, Native American heritage.

We will review the inheritance pattern of mitochondrial DNA, and briefly discuss some scenarios where it can be used to evaluate kinship or maternal relatedness. The match statistics observed in mitochondrial testing will be described.