The quest for knowledge of ones relatives and ancestors now takes an exciting leap forward with the inclusion of DNA testing. Family Tree DNA has already made Y chromosome testing available to those individuals interested in genealogical research through male family lines. Mitochondria DNA testing is similarly used to follow female ancestral lines and may be used to search for unknown relatives through testing and data basing the results.

Human mitochondria DNA (mtDNA) have a number of distinct properties that make it an invaluable tool for genealogical and anthropological study. mtDNA is maternally inherited and therefore the study of mtDNA is the study of female genetic lines within human populations. Additionally, there is little reshuffling or recombination in mtDNA. Mutations, when they occur, accumulate in a linear or chronological manner allowing scientists to study these changes. Using proper documentation and historical data, these mutations can also be correlated to the geographic area in which they first occurred. Ancient migration patterns can then be constructed based on the presence of these mutations in human populations.

First a simple review of cellular biology may be helpful. Mitochondria are organelles present in all human cells. They are present in the cytoplasm of each cell and contain their own DNA. In the process of human reproduction mtDNA is passed in the egg cytoplasm only. Males do not contribute mtDNA to the offspring but carry mtDNA contributed from their mother. A female receives mtDNA from her mother who received mtDNA from her mother along the female line. Therefore, mtDNA is handed from mother to daughter along the entire female line and to the male offspring, but the male does not pass this on to his next generation.

Specifically, mtDNA analysis is performed looking for both similarities and differences among individuals. Over 16,500 base pairs of genetic (mtDNA) material have been identified in humans and 400 base pairs encompassing the entire HVS-1 area, (hyper variable control region 1), are used for genealogical and genetic analysis. A base pair is a specific component of the DNA and is made of either purine bases Adenine (A), and Guanine (G), or pyrimidine bases Cytosine(C) and Thymine (T). Therefore, our report will express your results as a series of letters, representing the purine and pyrimidine bases specifically found in your HVS-1 control region. The HVS-1 area of the control region (CR) is used because mutations occur more frequently in this region and therefore changes can be followed to delineate specific human lineages more easily.

The physical process of analyzing your mtDNA begins with the simple scraping of your inner cheek using the brush provided in your sampling kit. The cells and the DNA they contain are preserved in the solution in the sample tube. When the lab

receives the test tubes, the DNA is extracted using a solution of chemicals and enzymes. This DNA must then be amplified or copied in order to have enough for the tests. This technique called the Polymerase Chain Reaction, was invented by Kary Mullis, a California scientist who later won the Nobel Prize for this invention. Each of the bases, (A) Adenine, (G) Guanine, (T) Thymine, and (C) Cytosine, are tagged with a fluorescent dye and run through an electric field which separates them into a series of colored bands. A laser detector then identifies the color and plots the read-out in a series of peaks and troughs. This is then computer analyzed to give the final sequence. This basic technique was developed twenty years ago by British scientist, Fred Sanger, and resulted in his second Nobel Prize.

Using this information, an analysis of the 400 base pairs surrounding and including the entire HVS-1 has been performed and a standard 400 base pairing referred to as the Cambridge Reference Sequence has been determined. Commonly mtDNA pairings are compared to this standard and differences are shown.

Your analysis highlights these differences and may be compared to other individual's pairs. Our database will be helpful in finding other individuals with exactly the same mtDNA. This exact duplication of the mtDNA means two individuals shared a common female ancestor. Research over the last decade has suggested several maternal lines ultimately all originating from the first woman "Eve" approximately 140,000 years ago in Africa. Further details may be reviewed in the journal *Nature Genetics*, November 2000 or in *Science*, November 2000.

How To Read Your mtDNA Certificate:

Your sequence is displayed on your certificate. Below your individual sequence you will notice four rows with 10 columns each, This is the Cambridge reference Sequence, with which all samples are compared. Begin reading at the upper left and continue to the end at the lower right.

Where you have RED numbers under your name this indicates that you have a mutation from the Cambridge Reference Sequence at that particular point. The red letters are the mutations to which you have moved. A red-letter C in position 16154, for example, means that at the 154th of the four hundred base pairs that are commonly studied, you have a C in place of the T listed for that position in the Cambridge sequence. A position like this that shows variation is called a polymorphism.

You may compare your polymorphism to other people in the database to determine whether you share a common maternal ancestor. We will soon have a database that you will be able to search against of the Mitochrondia of others who have tested with us to see if you have a common ancestor, or descend from the same branch of the tree.

The mitochondria area we examine begins at position 16,001 and extends to position 16,400.

Please allow yourself time to contemplate both the wonders of science that allow the evaluation of your mtDNA and the magnitude of human development over the past 140,000 years. Family Tree DNA is proud to be part of the process of helping you learn more about your family ancestral lineage.

In a few days we will begin to compare your specific sequence to other people in our database of mtDNA results. If we find someone who has the exact changes within their sequence as you do we will inform both of you by email, provided, of course you have signed the Family Tree DNA Release Form. In this way we will be able to link you up with your genetic cousins and you may then employ conventional genealogical techniques to see if you can find where that person (s) might actually fit on your family tree!

Supplemental data can be found from the site below where you can investigate your specific haplogroup and its relative frequency among the sample taken by a team at Oxford. It might take a few minutes of searching but the site below will allow you to find the Haplogroup that you descend from on your maternal side.

http://www.stats.ox.ac.uk/~macaulay/founder2000/index.html

If you want to read an in depth article on mtDNA and the settling of Europe, go here:

http://www.stats.ox.ac.uk/~macaulay/papers/richards_2000.pdf

Another detailed article worth reading is located at:

http://www.stats.ox.ac.uk/~macaulay/papers/980656.web.pdf

If you have questions about this report, or questions in general, please fill free to email us at info@familytreedna.com. Of course we encourage you to visit our web site http://www.familytreedna.com from time to time to see what new genealogical opportunities we have developed for you.

It has been a pleasure to serve you. If you have questions please visit our web site at: **FamilytreeDNA.com** and read our ever-expanding FAQ or inquire via email to **info@familytreedna.com**.

Your Privacy Code: _____

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