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Questions and Answers about DNA

Which company and product should I choose for a DNA test?

That depends on what you want to know and how much you are prepared to pay. Firms come and go, and their product ranges change. Two commercial firms that have specialised in genealogy research and individual historical origins are Family Tree DNA and 23andMe. National Geographic sells DNA tests focusing on individual historical origins.

There are many reliable websites that provide up-to-date information for people interested in buying their first test. One of them is run by SSGG, a non-commercial network of Swedish genealogy researchers working with DNA, of which I am a member. Its international counterpart is the International Society of Genetic Genealogy, ISOGG.

How does a test for a private individual work?

Usually, you'll order a test kit online from one of the firms mentioned above. Follow the accompanying written instructions to collect a small DNA sample; this may involve taking a swab from the inside of your cheek using a plastic spatula, or spitting into a test tube. Then send your sample off to the test firm. A few weeks later, you will receive a response online, which will contain some basic information. It may often be a good idea to approach experts who can provide further details, such as project administrators linked with the firm but working on a non-profit basis, or – if the test is particularly sophisticated – other firms that are specialised in interpreting DNA data. One such is YFull, based in Russia.

Who should I have tested first?

That depends on what you want to know. But one practical principle when using DNA as a tool for researching your family

history is to have the oldest individual in a particular line tested before it's too late.

What is DNA?

The acronym stands for deoxyribonucleic acid, a chemical present in nearly all our cells that passes on our biological traits. The DNA molecule comprises four units known as nucleobases. These are called adenine, guanine, cytosine and thymine, and are normally shortened to A, G, C and T. One individual's genetic material comprises over three billion As, Gs, Cs and Ts. Just a few nucleobases in every thousand differ between one randomly selected individual and another, the rest being identical. Although the variations are so small, they can provide a great deal of information about an individual's origins and genealogical relationships.

What are genes?

Genes are specific components of our DNA that code for hereditary characteristics. They account for no more than a tiny percentage of the whole DNA molecule. The rest of our DNA serves a regulatory purpose, has an unknown function, or is simply 'in-between DNA'. It is more important to keep sequences from within the genes confidential, as they can reveal information about the risk of particular medical conditions and other sensitive matters. It is advisable not to let out such information in public. However, it may be a good idea to let project administrators and other experts you may choose to consult know the whole sequence, including the parts that code for information. This may enable you to obtain better information about genealogical relationships.

What are mitochondria?

Mitochondria are tiny structures within cells but outside the cell nucleus that contain small amounts of their own DNA. We inherit our mitochondria solely from our mothers. It is simpler and cheaper to test mitochondrial DNA than other DNA, as we have large numbers of mitochondria in every cell. The downside, however, is that they contain a very small proportion of our total DNA – only

about 16,000 nucleobases, compared with the three billion nucleobases in the cell nucleus. One of their advantages is that mitochondria are passed on unchanged from mother to child for many generations. This means they can be used to trace a person's maternal lineage a long way back in time.

What is nuclear DNA?

Nuclear DNA, which accounts for a large proportion of all DNA, is found in the cell nucleus. Analysing nuclear DNA is generally more difficult and costly than analysing only mitochondrial DNA. The more detailed the analyses you order, the more expensive they tend to be.

What are Y chromosomes?

Y chromosomes are a part of nuclear DNA passed on only by men to their sons. They are sex chromosomes that normally give a man his sexual traits. By comparing mutations in Y chromosomes, a man's paternal lineage can be traced a long way back in time. The most detailed Y-chromosome tests provide far more information than the most detailed tests on mitochondrial DNA, so they will improve your chances of being able to make links with historical sources and the present day.

What are X chromosomes?

X chromosomes are also part of our nuclear DNA. Both men and women inherit an X chromosome from their mother. Women also inherit an X chromosome from their father. This is why women normally have two X chromosomes, while men have one X chromosome and one Y chromosome. The particular way in which X chromosomes are passed on mean they can be used to establish genealogical relationships by means of autosomal tests (see below).

What is autosomal DNA?

Autosomal DNA is DNA from the cell nucleus that an individual inherits from both their father and their mother, and which is randomly mixed each generation. The most common and inexpensive commercial tests for people looking into their family

history are based on autosomal DNA. They can provide information about siblings, parents, cousins, second cousins and other relatives at up to seven degrees of kinship. However, this naturally works only if the other people concerned have also been tested.

The strict biological definition of autosomal DNA is DNA that is neither mitochondrial nor from sex chromosomes. However, genealogy companies also include X chromosomes in autosomal DNA tests, making it easier to trace your relatives.

What is a haplogroup?

A haplogroup is a group of individuals with the same set of mutations, who thus share a common female or male ancestor some considerable way back in history. Another way of expressing the concept is that all the individuals in a haplogroup belong to the same branch of a genealogical tree.

What does STR mean?

STR stands for 'short tandem repeats'. A short tandem repeat is the location where the DNA molecule gets stuck, as it were, and a short sequence is repeated a number of times. The number of repetitions at each location is hereditary and differs between individuals. This makes STR a practical tool for ascertaining whether a family relationship exists. It has been used in forensics since the 1980s and is now also a popular tool in genealogical research, especially for comparing Y chromosomes. There is no known link between STR and hereditary traits, which makes its use less controversial. The commercial Y-chromosome tests known as Y-DNA37, Y-DNA67 and Y-DNA111 calculate the number of repeats at 37, 67 and 111 locations on the Y chromosome respectively. The higher the number, the greater the precision and the closer the genealogy researcher can get to the present day. A compromise that often provides good value for money is to test a moderate number of STRs and combine the results with tests on single nucleotide polymorphisms, SNPs.

What does SNP mean?

SNP stands for 'single nucleotide polymorphism'. It is a single location on the DNA molecule, the nucleobase, where people can

have different variants. SNPs are often called ‘snips’. Geneticists often test a selection of snips instead of all of a person’s hereditary material, as this is generally simpler and less costly. Snips have long been used in medical research, but they are now also beginning to be used in DNA-based genealogical research. There are thousands of different kinds of SNP tests, so you should discuss your needs with a specialist before selecting one, to be sure of which test will provide the most relevant information in your particular case.

What are HVR1 and HVR2?

These abbreviations stand for ‘hypervariable region’ 1 and 2. They are two small parts of our mitochondria where mutations occur more frequently than in the rest of our DNA. Analysing them shows which mitochondrial haplogroup a person belongs to. But this is a rough division which can only provide information about a person’s origin in the maternal line several thousand years ago. Looking into closer genealogical and family relationships and getting closer to the present day requires more complete DNA analyses of whole mitochondria.

What are CRS and rCRS?

CRS stands for Cambridge Reference Sequence, which is used for the purpose of comparing mitochondrial DNA. This involves identifying the mutations that differentiate a mitochondrion from the first human mitochondrion ever sequenced; this was done in Cambridge in the 1970s. The improved version is known as rCRS, the revised Cambridge Reference Sequence.

What is RSRS?

RSRS stands for Reconstructed Sapiens Reference Sequence. It is a more recent method for comparing mitochondrial DNA that has been in use since 2012. This method is based on the sequence that, according to scientists’ calculations, was borne by the female ancestor we all share, ‘mitochondrial Eve’.

What are mutations?

Mutations are random changes in the DNA molecule whereby certain units – nucleobases – are replaced, added or disappear.

There are roughly 30 new mutations per human generation. The older a father is, the more new mutations his children will have. Mitochondria undergo one mutation every 2,000 years on average within the same line of descent.

What do all the letters and figures in my mitochondrial DNA result mean?

They are terms for different types of mutations. To give an example, C40624T means that the nucleobase called cytosine, C, has been replaced by thymine, T, at location 40624. If the result begins with A, T or G instead, and ends with a different letter, this means that another nucleobase – adenine, thymine or guanine – in the reference sequence used (see rCRS and RSRS above) has been replaced.

Sometimes the result is followed by an exclamation mark; for example, C40624T!. The exclamation mark stands for a reverse mutation. This means that the original version – in mitochondrial Eve or the Cambridge Reference Sequence – had a T at exactly the same location. Within the haplogroup as a whole – the larger branch to which the sample belongs – that T has been replaced by another nucleobase. But the side branch on the tree to which the tested sample belongs has since mutated back to the original T.

Sometimes you can see 315,1 or 315+C. That means that an extra nucleobase, a C in this instance, has been added after position 315. The opposite of that would be 315D. D stands for deletion, meaning that the nucleobase at the location indicated has been deleted.

There are a number of other abbreviations for specific types of mutations.