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SNP Report

Note

Please be aware that this report was prepared in 2008 and practically everything we now know about SNPs has been discovered since then. Please do not rely on this information or order the tests recommended in this report as there are now much better alternatives. We are looking for someone who understands or is willing to do the work to learn about SNP testing to take over this aspect of the project and we hope that this page will eventually be updated. Meanwhile, interested persons can refer questions to David Neal Ewing. This page reports on actual Single Nucleotide Polymorphism (SNP) testing undertaken by Ewing Project participants several years ago and presupposes a working knowledge of the project and how its groups are organized. Links to our other Results Pages are available in the Results Directory, which can be reached by the link at the top of this page. If you are not acquainted with the organization of the website, please start with the Help link at the top of the page. If you are interested in a considerably more detailed discussion of SNPs and Haplogroups, go to Haplogroups, Haplotypes and Clusters for the Flustered.

Background

In genetic genealogy the most useful markers are Short Tandem Repeats (STR) on the Y-Chromosome. Most of our project participants are tested for 37 STR markers; the results of these tests are posted in the Group Results Tables. Here, we report the results of those project participants who have had SNP testing. SNPs are more useful in genetic anthropology for studying the deep ancestry of the human race long before the period accessible to genealogy. The reason for this is that the mutation rate for SNP markers is very much slower than for STR markers. The mutation rate at SNP markers is so slow that for practical purposes they can be considered unique and permanent; that is, they are not subject to parallel or back mutations. For this reason, they have been used to identify branches in the human family tree, where each branch point is defined by one or more SNPs. These branches are called Haplogroups.

Results

Group-ID		Haplo-group	SNPs
1*	TF	R1b1	P25+, no others tested
1e	JC	M222+	M173+, M207+, M222+, M269+, M343+, P25+, M126-, M153-, M160-, M18-, M37-, M65-, M73-, P66-, SRY2627-
1*	DL	M222+	M173+, M207+, M222+, M269+, M343+, P25+, M126-, M153-, M160-, M18-, M37-, M65-, M73-, P66-,

			SRY2627-
1b	DN	M222+	M173+, M207+, M222+, M269+, M343+, P25+, M126-, M153-, M160-, M18-, M37-, M65-, M73-, P66-, SRY2627-
1a	BE	R1b1	P25+, no others tested
2a	JN	M222+	M173+, M207+, M222+, M269+, M343+, P25+, M126-, M153-, M160-, M18-, M37-, M65-, M73-, P66-, SRY2627-
2a	WR	R1b1	P25+, no others tested
4a	JM	R1b1	P25+, no others tested
4a	DS	R1b1	P25+, no others tested
1c	WC	R1b1	P25+, no others tested
4c	PT	R1b1c*	M173+, M207+, M269+, M343+, P25+, M126-, M153-, M160-, M18-, M222-, M37-, M65-, M73-, P66-, SRY2627-
4*	MK	R1b1	P25+, no others tested
5a	HN	I	M170+, no others tested
5a	WM	I	M170+, no others tested
5a	VC	I	M170+, no others tested

Discussion

In April 2008, the Y-Haplogroup tree that we had been using since 2002 was extensively revised and most haplogroups were given new names, based on the same set of rules that had resulted in the names on the old tree, but taking into account hundreds of new SNPs that had been discovered in the intervening years. What had been called R1b1c7 became R1b1b2e. Since then the name of this haplogroup has been revised at least three more times, and the name and naming system have become impossibly unwieldy. Although in some branches of the tree, the changes resulted in significant restructuring with implications for the relationships of various subclades, in the case of that part of the tree of most interest to us, all that really changed was the name. We have resolved to begin referring to this haplogroup by using simply the name of its defining SNP, M222+. Interested individuals can find information about the tree in many places on the web. Perhaps a good place to start would be the scientific paper that described the changes and the rationale for them. This is available at www.genome.org/cgi/content/abstract/gr.7172008v1. The most recent, or at least a relatively recent iteration of the tree can be found on the ISOGG website at www.isogg.org.

When FTDNA does our STR testing, they automatically “predict” a haplogroup for each participant. Even though haplogroups are defined on the basis of SNPs, this usually can be done with a high degree of confidence without testing any SNPs because certain patterns of STR results are very characteristic of specific haplogroups. Indeed, Whit Athey has used similar logic to construct a “haplogroup calculator” that will not only predict the haplogroup for any specific haplotype but will give the probability that the prediction is correct. When FTDNA feels that they cannot confidently predict the haplogroup for a haplotype, they automatically do a “Backbone SNP test” at no extra charge. As near as I can tell, this involves testing just one or a few SNPs, and they report only the SNP that confirms membership in one of the haplogroups. So far, the only haplogroups represented in the Ewing project are R1b1 and I; P25+ is enough to confirm membership in haplogroup R1b1, and M170+ is enough to confirm membership in haplogroup I. In the results table shown on the Ewing FTDNA site, predicted haplogroups are shown in red and haplogroups that have been confirmed by SNP testing are shown in green. We have not bothered with this in the Results Tables on this website, but all of those whose haplogroups appear in green on the FTDNA site are shown in the table above. Please notice that in the table above TF, BE, WR and WC all appear as R1b1 rather than as M222+ because they have been tested only for P25 and not for M222. Because their haplotypes (and in the case of BE, his conventional genealogy) show a close relationship with men who have been tested M222+, we can be certain enough that they are also M222+ that they should not waste their money getting the additional test. On the other hand, JM, DS and MK also have only been tested P25+ and so are also shown as R1b1, but their haplotypes are not like anyone who has been tested and we do not know whether or which subclade of R1b1 they might fall into if they were to have additional SNP testing.

FTDNA also offers for sale “deep clade testing” for members of haplogroups I and R1b1 for an additional charge of $79, which involves testing another fourteen or fifteen SNPs in order to determine what sub-branches participants may belong in. So far, we have had only four men order this testing. Comparing the “old” R1b1 phylogram with the “new” R1b1 phylogram will give you an idea of what is happening to our nomenclature system and understanding of branch structure as new SNPs are discovered.

In the first diagram, work backward from R1b1c7 and you can see that men in this branch will have M222+, M269+, P25+, M343+, M173+ and M207+. Notice that JC, DN, JN and PT all have M207+, M173+, M343+, P25+ and M269+. In order, these markers put them in R, R1, R1b, R1b1 and R1b1c. Now notice that PT is negative for all ten of the remaining markers. In this diagram, that leaves him in R1b1c, but properly speaking, this should be shown as R1b1c*, where the asterisk signifies that he has been tested for all the SNPs marking sub-branches of R1b1c and did not have any of them. JC, DN and JN are M222+, but negative for all nine of the remaining markers. This puts them in the R1b1c7 sub-branch. JC, DN and JN are in Ewing Groups 1e, 1b and 2a respectively, all in the large

closely related group of Ewings, which has a rather tight cluster of haplotypes. Because of this, we believe that all of the men in the large closely related group of Ewings (which includes all the men in Ewing Groups 1 and 2) are certain to have the same set of SNPs, so we think that ordering more deep clade SNP testing would be a waste of money for them. What is more, the men in Group 3 are even closer to the M222+ STR modal, so they are also sure to be M222+, and we do not recommend deep clade testing for them, for the same reason. PT is in Ewing Group 4c, and though his cousin RL2 is certain to have the same SNPs as he does, this is not necessarily true of the other men in Group 4, and their deep clade testing might be interesting. Since the men in Group 4a are another tight cluster, only one of them would have to get this testing for all of them to know their results. Perhaps I should say that we can be reasonably confident that all of our R1b participants are actually in R1b1c, and though we cannot predict which of them might have one of the downstream SNPs that would put them into one of the sub-branches of R1b1c, most of them will turn out to be R1b1c*. Though none of them will get additional information of genealogical applicability from additional SNP testing, they might learn something interesting about deep origins of their line.[1]

The second diagram does not show quite as much of the tree—it starts at the top left with M343, already down to the R1b level on the first diagram. On the second diagram, the SNPs in green and red are the ones that appear in the first diagram. Folks in the M222+ subclade (old R1b1c7, new R1b1b2a1b5) are positive for the green ones and negative for the red ones, just as they have been right along. The folks in our project who have had deep clade testing have actually been tested for all of the green and red SNPs, and all of them are M222+ except for PT in Group 4c. He was found to have M269+, but none of the SNPs downstream for that, and therefore was found to be in old group R1b1c (as FTDNA called it; this is more properly R1b1c*). In the new diagram, he would be in Rb1b1 because he is M73-. Neither PT nor the rest of us have moved anywhere; it is just that a bunch of new folks have moved into the neighborhood and they changed our address. The short story is that we know what the result would be for the men in our Groups 1, 2 and 3 on all of the new SNPs because the men in Groups 1, 2 and 3 are all M222+, and all M222+ individuals have the same set of upstream SNPs, whether these have been discovered or not. Undoubtedly, someday some SNPs downstream from M222 will be discovered, and that will result in the subdivision of this group. One of the “Holy Grails” of genetic genealogy is to discover a downstream SNP that will distinguish Scottish M222+ from Irish M222+. Don’t hold your breath.

So far, none of our project participants in haplogroup I have had deep clade testing, though all three of the Group 5a men have been tested for M170 because FTDNA was having trouble predicting their haplogroup, and all three

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[1] Vincent Vizachero has prepared some maps of the distribution of R1b1c6, 7, 9 and 10.

tested M170+. I was rather keen on at least one of the Group 5a men getting Haplogroup I deep clade testing, because arguments have been made that some of the sub-branches are more characteristic of Vikings and others more characteristic of Eastern Europeans, etc. Ken Nordtvedt has done a fair amount of work on the branch structure of haplogroup I.[2] His STR cluster analysis has our Ewing Group 5a men falling into what he presently terms the "I1b2a1-Isles/English" cluster, which any sensible person would assume to be a sub-branch of what appears as I1b2a on the diagram above. Be careful of the terminology, though; it is constantly changing and, in fact, Nordtvedt is using a variation on an updated nomenclature that has re-labeled the braches on the tree above, and our Group 5a is a sub-branch of what appears there as I1c. To say this same thing in a way that is not so subject to terminological vagaries, Nordtvedt would predict on the basis of his STR cluster analysis that if our Ewing Group 5a men were tested for these SNPs, they would test P38+, M253-, M307-, P30-, P37.2-, and M223+. He would also predict that they would test M284+ (which is not presently included in FTDNA's version of deep clade testing, and does not appear on the diagram above). M284 is a downstream SNP that characterizes a branch that includes two STR clusters, our guys' I1b2a1-Isles/English cluster and a related cluster, I1b2a1-Isles/Scot, which is more commonly found in Scotland. To say this in English, our Group 5a men appear to belong to a subclade of haplogroup I that is found mostly in England and not to speak of on the continent, which suggests that he may be descended from haplogroup I indigenous British that were in England long before any Anglo-Saxon or Scandinavian invasions. Now, it looks like we do not need to get the deep clade testing for these guys, because two of them have recently had the 67-marker upgrade and this essentially confirmed that they are M284+ without actually testing the SNP. How can this be? Well, it turns out that both these men had a so-called null result at DYS 425. This means that the assay found no repeats. The reason for this is that there has been a mutation in the primer region of that marker and the PCR primer could not bind to it, which prevents counting the repeats. By coincidence, this mutation is almost certainly the M284+ mutation. What a coincidence. M284+ is a SNP (a change at a single nucleotide) that just happens to be in the primer region for one of the STR markers. So a null value at that marker practically confirms that M284+ is present. I say "practically" because it is also possible that in this kindred there was some other sort of damage in the primer region, but the fact of a null DYS 425 marker combined with Nordtvedt's cluster analysis is all but proof certain.

On the other hand, the only Ewing man (JD) with 37-marker results in Group 2, who is also in haplogroup I, falls into a different Nordtvedt cluster, I1b*, which I think fits in the diagram above at I* or I1*, I cannot tell which because Nordtvedt seems not to have included P38 in his analysis. It is very difficult to speak with much confidence about the timing of most recent common ancestors, but although JD (in 5b) and the men in Group 5a have a common ancestor, he must have lived several hundred generations before their branches diverged, probably

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[2] You can see some of his work at http://knordtvedt.home.bresnan.net/

more than 25,000 years ago. I know nothing about the distribution of the haplotype cluster that JD belongs to.

To Order Deep Clade Testing

Remember, if you are in Ewing Group 1, 2, 3 or 5a you don't need this testing. But I would like to see at most one man from each cluster in Group 4 get the test. I would be happy to discuss with anyone who may be interested whether this testing might be useful for him. If you would like to add this testing, please go to your FtDNA personal page, click on ***Haplogroup*** in the menu on the left-hand side of the page, then under Haplogroup Test click on ***Continue for more Information***, and then click on ***Continue to order*** and fill out the billing information. It will not be necessary to submit another specimen, because FtDNA already has the one you submitted previously in storage. If you have trouble ordering the test or are not sure whether it will be an informative test for you, please send an email to david_ewing@clanewing.org and he will help you.