Journal: www.jogg.info
Originally Published: Volume 1, Issue 2 (Fall 2005)
Reference Number: 12.006

RESOLVING THE PLACEMENT OF HAPLOGROUP I-M223 IN THE Y-CHROMOSOME PHYLOGENETIC TREE

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Journal of Genetic Genealogy 1:54-55, 2005

Resolving the Placement of Haplogroup I-M223 in the Y-Chromosome Phylogenetic Tree

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Evidence from four case studies is presented demonstrating that Y Haplogroup I-M223 should properly be considered a subgroup of Haplogroup I1, rather than as a separate Haplogroup I2 as recently proposed. Haplogroup I-M223 had not been discovered when the 2002 Y phylogenetic tree was published by the Y-Chromosome Consortium (YCC). The first study of Haplogroup I-M223 designated it as a subgroup of I1 and named it I1c. A later proposed revision of the Y phylogenetic tree, however, showed I1a and I1b as P38+ and I1c with P38-, which required Haplogroup I-M223 to be renamed as Haplogroup I2. The present study presents four cases in which M223+ men were shown to be also P38+, which reunites Haplogroup I-M223 with I1a and I1b within Haplogroup I1.

Introduction

Y-chromosome Haplogroup I includes about a quarter of all northwest European men. Its largest subgroup, I1a, is common in Scandinavia and Germany and occurs in Britain at a frequency of about 15%. Haplogroup I1b is common in the Balkans and parts of Eastern Europe. The third major subgroup of Haplogroup I is defined by the single nucleotide polymorphism (SNP) M223, as reported by Rootsi et al (2004). Rootsi placed I-M223 as a subgroup of I1, parallel to I1a and I1b, and named it Haplogroup I1c (see **Figure 1**).

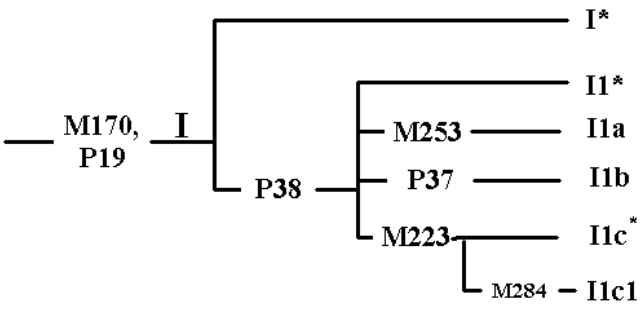

Figure 1 Phylogenetic Chart for Haplogroup I according to Rootsi et al. (2004) (simplified for clarity).

Received: November 7, 2005, accepted November 29, 2005

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The SNP M223 had not been discovered in 2002 when the Y-Chromosome Consortium (YCC) issued the latest official version of the Y phylogenetic tree. Family Tree DNA (FTDNA) in late 2004 published a revision of the YCC-2002 structure. In this new tree, Haplogroup I-M223 was apparently assumed to be P38- and, as such, it was renamed as Haplogroup I2, as shown in **Figure 2**.

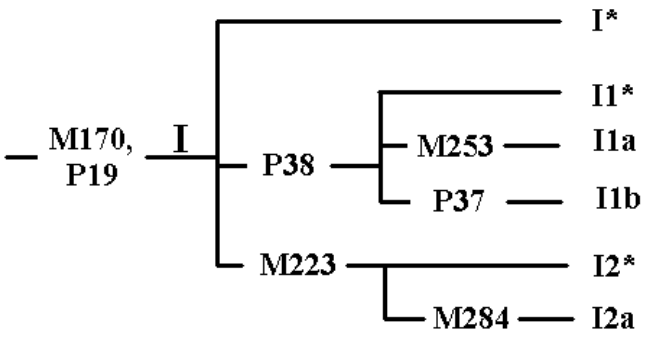

Figure 2 Haplogroup I According to FTDNA's Proposed Revision of the Y Phylogenetic Tree (simplified for clarity).

The current study presents the results of testing of SNPs in four men who were known or suspected to be members of I-M223. In all cases, they were found to be both M223+ and P38+, demonstrating that the phylogenetic structure of Haplogroup I as presented in Rootsi et al (2004) (see **Figure 1**) is correct.

Methods

Four subjects were chosen from four different surname projects where the Y-STR values suggested strongly that they were members of I-M223. Three subjects had

Table 1 – Y-STR Values* for All Subjects

ID	393	390	19	391	385a	385b	426	388	439	389i1	392	389i2	458	459a	459b	455	454	447	437	448	449	464a	464b	464c	464d	464e	464f	460	GATAH4	YCAIIa	YCAIIb	456	607	576	570	CDYa	CDYb	442	438
101	15	24	15	10	15	15	11	13	13	14	12	31	15	8	10	11	11	25	14	21	27	11	14	14	15															
102	14	22	15	10	15	15	11	13	11	13	12	29	15	8	10	11	11	25	15	20	28	11	14	15	15			12	10	19	19	17	16	16	20	34	36	12	11
103	14	23	15	10	14	15	11	13	11	14	12	32	14	8	10	11	11	25	14	19	27	11	14	15	15															
104	14	24	15	10	15	17	11	13	11	13	12	29	16	8	9	11	11	26	15	23	27	11	11	11	11	14	15	11	10	19	21	15						12	10

* Results are presented in the same order as in an FTDNA report.

results from testing either 25 or 37 Y-STR values carried out at Family Tree DNA (FTDNA, Houston, TX), and one from testing 45 markers at Relative Genetics (Salt Lake City, UT). The Y-STR values, shown in **Table 1**, were used with the permission of the participants, and were known prior to the present study.

Participants in the present study were designated as Subjects 101-104. The subjects displayed a diverse set of Y-STR haplotypes and (where known) geographic origin. Subject 101 has an uncertain geographic origin, but his Y-STR values are similar to I-M223 haplotypes from northwest continental Europe as described by Nordtvedt (2005). Subject 102's lineage is from Spain and Subject 103's lineage is from Germany. Subject 104 has Y-STR values similar to those found in Britain for this haplogroup (Nordtvedt 2005). Thus, each subject in the study represents one of the major European populations in which the haplogroup is found.

For the present study, SNP testing was carried out on all subjects by Ethnoancestry (Cyprus, CA) for SNPs M223 and P38. At Ethnoancestry, Y chromosome SNPs were amplified by PCR with standard primers giving products from 200 to 500 bp in length. PCR products were then sequenced using dye terminator chemistry with electrophoresis on a capillary ABI sequencer. Alleles were called in the software package, Sequencher, by alignment with chromosomes of known allelic state (positive and negative controls).

Table 2 SNP Results for All Subjects, This Study*

ID	SNP Status (+ or -)					
101	M170+	P38+	M223+	M253-	P37-	
102	P19+*	P38+	M223+*			M284-
103	M170+	P38+	M223+	M253-	P37-	
104	M170+	P38+	M223+	M253-	P37-	M284+

* All results are from Ethnoancestry except those marked with an asterisk. The P19 test was carried out at FTDNA, and one M223 test was carried out at Trace Genetics.

Results

The results for all of the SNP tests from the present study are shown in **Table 2**. All subjects were found to be derived (positive) for both P38 and M223. Where additional SNP testing was done, these results are also included in Table 2 for completeness.

Conclusion

Since all subjects were P38+ and M223+, the correct phylogenetic structure of Haplogroup I is shown in the chart published by Rootsi (see **Figure 1**). Thus, the alternate structure shown in **Figure 2** is incorrect. Since Haplogroups I-M253, I-P37, and I-M223 are parallel sub-clades of Haplogroup I1-P38, they are properly named I1a, I1b, and I1c.

Electronic-Database Information

http://www.northwestanalysis.net/
Haplogroup I Information

References

Nordtvedt K (2005) Population Varieties within Y-Haplogroup I and their Extended Modal Haplotypes. Web site, URL = http://www.northwestanalysis.net/.

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