



:: Haplogroup Result ::

The testing of your sample shows that you were positive on the above highlighted markers. Additionally, you were negative on the other markers within the panels tested. Knowing this we can determine your haplogroup. An asterisk after the haplogroup would designate that branch-defining markers below the haplogroup have been tested but proved negative - e.g. I* or R1b3*

We determine you to be in Y-chromosome haplogroup **I1a**

Y-chromosomal haplogroup I is defined by markers M170 and P19 according to nomenclature of Y Chromosome Consortium (YCC) (2002; 2003). In earlier literature this haplogroup was referred as Eu 7 plus 8 (Semino et al. 2000) or haplogroup 2 (according to nomenclature of Jobling et al. 1997; Rosser et al. 2000; Helgason et al. 2000). Although the latter 'superhaplogroup' overlaps only partly with haplogroup I, in European populations the coverage is more relevant, as majority of European haplogroup 2 individuals belong to haplogroup I according to more recent studies. Phylogeny of defining markers for haplogroup I in wider context of haplogroup F (lineages defined by M89) according to nomenclature of YCC (2002) was presented in study of Rootsi et al. (2004). An asterisk after the I would designate that markers below M170 have been tested but proved negative - hence I*

The polymorphism M170 represents a putative Palaeolithic mutation which age has been estimated to be about 22 000 years (Semino et al. 2000), similar value of about $23,000 \pm 7,700$ years was given by Rootsi et al. (2004). It has been proposed by Semino et al. (2000) that M170 originated in Europe in descendants of men who arrived from the Near East about 25 000 years ago. The initial spread of the haplogroup I carriers in Europe is usually linked to the diffusion of the largely pan-European Gravettian technology.

Gravettian is the second subdivision of the Upper Paleolithic technological phase in Western Europe (from 27,000 to 21,000 years ago). Gravettian culture earlier phase (c.28,000-23,000 ya) of the European Upper Paleolithic is characterized by a stone-tool industry with small pointed blades used for big-game hunting (bison, horse, reindeer and mammoth). It is divided into two regional groups: the western Gravettian, mostly known from cave sites in France, and the eastern Gravettian, with open sites of specialized mammoth hunters on the plains of central Europe and Russia. The most characteristic artworks made by Gravettian artists were the famous Venus figurines.

Haplogroup I-M170 is a component of the present European Y-chromosome gene pool accounting on average for 18% of the total paternal lineages and is the only major clade of the Y phylogeny, autochthonically arisen, widespread over Europe but virtually absent elsewhere, including Near East (Semino et al. 2000; Barac et al. 2003; Rootsi et al. 2004).

In earlier studies it has been shown that high frequency of hg I (about 40-50%) is characteristic for two distant and distinct regions of Europe - Southern Europe, around the Dinaric Alps (Barac et al. 2003; Semino et al. 2000) and in Nordic populations of Scandinavia (Tambets et al. 2004; Semino et al. 2000; Passarino et al. 2002). In other regions of Europe intermediate or low frequency is characteristic. Relatively high frequencies are characteristic also to some French regions like Low Normandy (24%) and Southern France (16%). Mostly the frequency cline is fluent, but interestingly, a lower frequency of haplogroup I distinguished the Baltic speaking Latvians from their northern neighbors, the Finnic-speaking Estonians. Similar cases of even more significant frequency change over a short geographic distance occur between the Southern Slavic speaking populations and their adjacent neighbors: namely Slovenians versus North Italians, and Macedonians versus Greeks.

This kind of 'peculiar' two-peak distribution pattern in Europe is uncharacteristic to other distinguished Y-chromosomal haplogroups and such a pattern is hard to explain by any possible migratory or expansion event.

The explanation to such irregular distribution-pattern came with further studies, where due to new informative markers the improved resolution of phylogeny of hg I enabled to reveal distinct, clearly defined phylogeographical patterns of haplogroup I sub-clades.

Haplogroup I consists of several distinct sub-clades: I1a (defined by marker M253), I1b (defined by marker P37) and I1c (defined by marker M223), which jointly cover about 95% of hg I individuals. The rest 5% of individuals remain in I* clade (individuals who lack any known downstream markers inside haplogroup) and are distributed with extremely low frequency all over the spread area of the haplogroup. They may represent the real ancestral I* individuals and/or individuals, who belong to sub-clades, so far uncovered (Rootsi et al. 2004). Phylogenetic network of hg I STR haplotypes (Rootsi et al. 2004) points to characteristic haplotype patterns in different sub-clades that allow identifying possible founder haplotypes for the sub-clades. Sometimes (especially in case of typical haplotypes) certain haplotype motifs that are characteristic to specific sub-clade enable even to guess the possible affiliation to certain sub-clade (in case of lack of SNP typing).

The present distribution of different sub-clades of haplogroup I is considered to be connected to post-LGM re-colonization of Europe. After the end of the Last Glacial Maximum (LGM) climatic improvement, repopulation of Europe started, most likely as expansions from isolated population nuclei from different refugial zones in Europe - Iberia (Franco-Cantabrian refugium), the present Ukraine (Periglacial) and from the northern Balkans (Dolukhanov 2000). The post-LGM expansion of haplogroup I sub-clades is prevalingly connected with expansions from Franco-Cantabrian and Balkan refugial areas.

Sub-clade I1a is defined by marker M253 (Rootsi et al. 2004) and P30 according to Y Chromosome Consortium (YCC 2002; 2003). This clade is the most frequent and characterized by widest distribution area among European-specific haplogroup I sub-clades covering practically whole distribution region of this haplogroup.

I1a is widely spread in northern Europe with its highest frequencies in Scandinavia: in Norwegians (39%), Swedes (39% in Southern and 26% in Northern Swedes) and Saami (29%), accounting for 88-100% of hg I individuals in these populations and showing rapidly decreasing frequency towards both the East European Plain and the northwestern coastal areas of Europe (Rootsi et al. 2004). Since the Scandinavian Peninsula was completely depopulated during the Last Glacial Maximum (LGM), two main European refugia, the Iberian Peninsula/Southern France and the Ukraine/Central Russian Plain (Dolukhanov, 2000) could be considered as possible source regions of Scandinavian I1a chromosomes.

Although Hg I occurs in the Ukraine at higher incidence (22%) than in western Europe (11% in France), the virtual absence in Scandinavia of the most represented East European I1b* sub-clade together with the higher I1a microsatellite (STR) diversity background points to western Europe as a source of the Scandinavian I1a chromosomes. The STR diversity of I1a decreases from Western to Eastern Europe, showing a significant negative correlation with longitude. In France, I1a is the leading sub-clade, representing 45% of the Hg I lineages, while in Ukrainians I1a covers only 5% of haplogroup I. Therefore, combined analysis of STR diversity and relative portion of I1a sub-clade among all I lineages suggests that France or possibly more precisely - the Franco-Cantabrian refugial area - could have been the source region of the spread of I1a during the post-LGM re-colonization of Europe. The same may apply to the spread of the less common sub-clade I1c. This scenario is also supported by high positive correlation between the geographic distributions of I1a and I1c.

A temporal interpretation of the phylogeographical spread based on the results of the STR length variation (Zhitovskiy et al. 2004) in the individual sub-haplogroups of I was reported in Rootsi et al. (2004) and the results support the view that I1a, I1b and I1c, all have diverged from I* in the Late Upper Palaeolithic/Mesolithic period, possibly during the re-colonization of Europe after the LGM. Yet the expansion phase of I1a and I1b seems to have occurred later, around the early Holocene. The possible migration to Northern Europe was following the coastal route through the north-western Europe reaching up in Scandinavia. As this region was unoccupied after the LGM, the present high frequency of I1a was most likely influenced by founder effect, as the carriers of I1a were almost certainly among the first settlers, who moved across the already melted area of coastal region of Scandinavian Peninsula.

The most frequent haplotype of I1a sub-clade defined by 6 STRs (DYS19, 388, 390, 391, 392 and 393) with characteristic haplotype 14-14-23-10-11-13 corresponds to the earlier named Nordic Haplotype (Barac et al. 2003). This haplotype is most frequent among Scandinavian populations, while in Western European, North-Western mainland European populations and also populations of British Isles possess a one step derivative from the Nordic haplotype as the most frequent one (differs in DYS 390 allele 22 as most frequent instead of 23). This may reveal that the initial modal haplotype for this clade might be 14-14-22-10-11-13 haplotype and Nordic haplotype, although being most frequent in Scandinavian population, is a derivative from the ancestral type, probably having arisen during the northward migration of I1a carriers after the LGM.

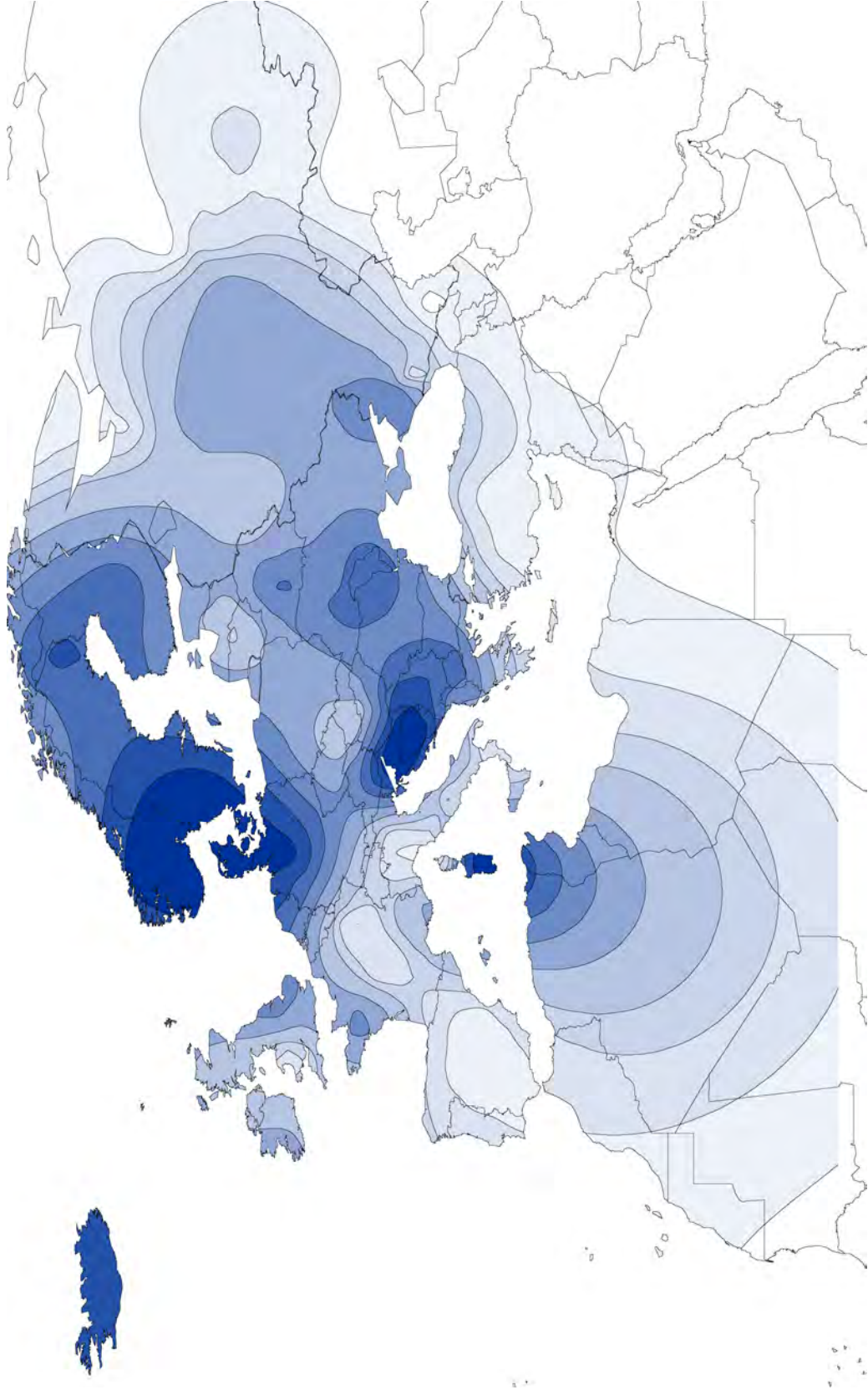
There also exists a specific STR marker, namely DYS 455, which allele 8 distinguishes I1a clade from all other I sub-clades that all are characterized by allele 11. This difference is sometimes used to define the clade (used as a 'surrogate SNP') by people interested in their genealogy.

At present phylogenetic resolution level there has been defined a minor sub-clade inside I1a, called I1a4 and characterized by marker M227. This sub-clade has been found mainly as single observations scattered in East and Southeast Europe.

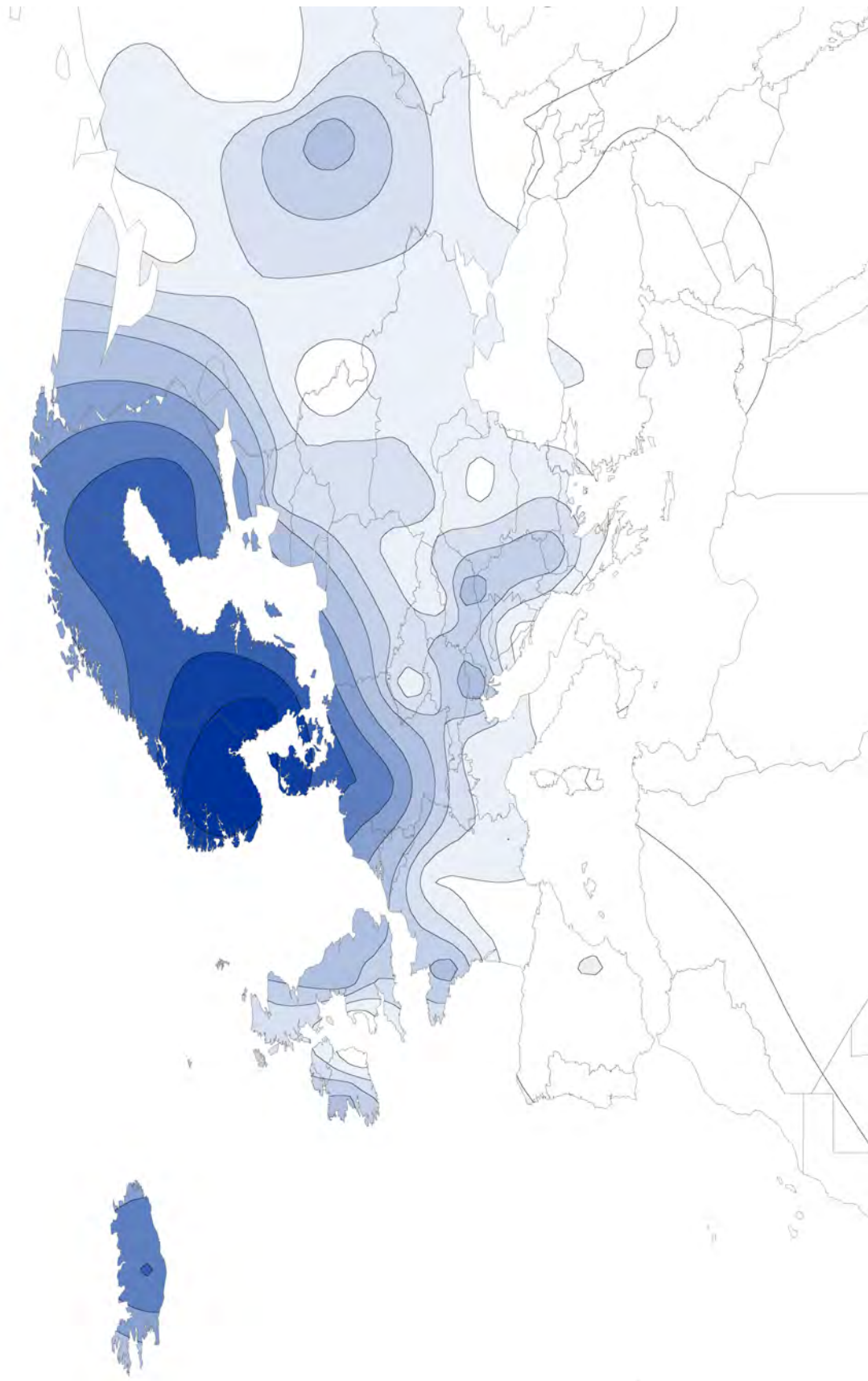
Interesting fact to note is that sub-clade I1a shows a distribution similar to the second PC of the synthetic maps

based on classical genetic loci (Cavalli-Sforza et al. 1994) and reveals a significantly positive correlation with mtDNA haplogroups V and U5b that have been suggested to be the markers of a postglacial population expansion from Iberia (Torroni et al. 1998; 2001; Tambets et al. 2004).

Distribution map for Y-chromosome
haplogroup I
(note: map is a composite of sub-haplogroups of I)



Distribution map for Y-chromosome
haplogroup I1a



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