

ANCIENT MITOCHONDRIAL DNA FROM STONE AGE LITHUANIA AND THE POSSIBLE ORIGINS OF THE FIRST INHABITANTS

RIMANTAS JANKAUSKAS

Abstract

This paper discusses recently published data on mitochondrial DNA (mtDNA) extracted from Stone Age burials in Lithuania in a broader European context, and data from modern Lithuanians on the basis of recent literature. Several major processes (initial Palaeolithic colonisation, recolonisation after the LGM and Younger Dryas cold relapse, the spread of the Neolithic, and possible small-scale migrations in the Eneolithic age) could have left traces on the modern gene pool. From four Lithuanian samples where data on mtDNA were available, one (Spiginas 4) belonged to haplogroup U4, and three (Donkalis 1, and Kretuonas 1 and 3) to U5b2. In total, out of 17 individuals from Central and East European non-farming cultures (Mesolithic and Neolithic Ceramic, spanning a period from circa 7800 BC to 2300 BC), a majority of them had mtDNA type 'U'. An exceptionally high incidence of U5-types (more than 45%) occurs among the modern Saami (Lapps) of northern Scandinavia, perhaps the closest modern European equivalent of Mesolithic hunter-gatherers. Genetic time estimates based on modern mtDNA have suggested that the U5-type arose by mutation about 50,000 to 40,000 years BP. This age implies that around the glacial maximum 20,000 years BP, U5 types were already present and could have repopulated Central and northern Europe as soon as northern areas were deglaciated. Both western (Franco-Cantabrian) and eastern (Pontic) refugia could be sources of this repopulation. In the recent Lithuanian population, U5 and U4 haplogroups are infrequent. The mtDNA homogeneity observed across modern Europe is a more recent phenomenon, less than 7,000 years old, according to these ancient mtDNA results. We can refer to the third millennium BC, internal European migrations from the Eneolithic that significantly modified the genetic landscape, as a time window little explored by archaeogeneticists. The imprecise chronology of mtDNA mutations should in the first instance be based on audited archaeological sources.

Key words: mtDNA, U5 haplogroup, Stone Age, Lithuania.

Introduction

Europe as a subcontinent has witnessed several dramatic changes in archaeological cultures and population since since anatomically modern humans displaced the Neanderthal population around 30,000 to 40,000 years ago. Palaeolithic hunter-gatherers survived the Last Glacial Maximum (LGM) about 25,000 years ago in southern and eastern refugia (Richards *et al.* 1996; Torroni *et al.* 2001), and resettled Central and Eastern Europe following the retreat of the ice sheets. With the end of the Ice Age ~9600 BC, their Late Palaeolithic and Mesolithic descendants or successors recolonised large parts of the deglaciated northern latitudes. Starting in 7000 BC, the Mesolithic way of life of hunting, gathering and fishing was rapidly or gradually replaced by the Stone Age farming cultures. Several major processes (initial Palaeolithic colonisation, recolonisation after the LGM and Younger Dryas cold relapse, the spread of the Neolithic, and possible small-scale migrations in the Eneolithic age) could have left traces on the modern European gene pool (Barbujani, Bertorelle 2001; Soares *et al.* 2010). The potential impact of such transitions on the genetic ancestry of modern Europeans has been extensively debated (Cavalli-Sforza *et al.*

1994; Richards *et al.* 1996; Sampietro *et al.* 2007). The application of ancient DNA studies added new stimulus to these discussions, as archaeogenetic research can help in reconstructing details of demographic history (Renfrew 2010), and mitochondrial DNA (mtDNA) at present is considered to be the most informative genetic marker system for studying European prehistory (Soares *et al.* 2010). Some investigations have shown that early Central European farmers had a very limited genetic impact on the present European mtDNA pool (Haak *et al.* 2005; Burger *et al.* 2006). However, it appears that there cannot be a universal 'pan-European' model of cultural and population-biologic processes, as each climato-geographic region could have its own specificity (Zvelebil 1998), and populations are likely to differ in their history and genetic composition. During the last few years, advances in ancient DNA studies have provided a rapidly growing amount of data that serves as a basis for the verification or creation of new hypotheses, and also on a regional level, in our case, the eastern Baltic area (Bramanti *et al.* 2009). In this paper, we will attempt to summarise available relevant mtDNA data, and to discuss their impact on possible trends for future research.

Data from ancient Lithuanian DNA: analysis and discussion

A recent study (Bramanti *et al.* 2009) provides the following clades of mitochondrial DNA of Lithuanian Mesolithic and Neolithic samples:

Spiginas 4 (GIN-5571: 7470 ± 60 BP) **U4**
 Donkainis 1 (Cultural context - Mesolithic) **U5b2**
 Kretuonas 3 (OxA-5926: 5580 ± 65 BP) **U5b2**
 Kretuonas 1 (OxA-5935: 5350 ± 130 BP) **U5b2**

In total, out of 17 individuals from Central and Eastern European non-farming cultures (Mesolithic and Neolithic Ceramic, spanning a period from circa 7800 BC to 2300 BC), a majority of them had mtDNA type 'U': 11 belonged to the mitochondrial lineage U5, and two further individuals were U4-types. The presence of U-types, and in particular U5-types, in Stone Age Europeans has been predicted by some authors (Richards *et al.* 2000), based on the relative high frequency (about 10%) in the modern European population and on a founder analysis. It should be noted that an exceptionally high incidence of U5-types (more than 45%) occurs among the Saami (Lapps) of northern Scandinavia (Tambets *et al.* 2004), traditional reindeer herders, and therefore perhaps the closest modern European equivalent of Mesolithic hunter-gatherers. Interestingly, U5b2 subclade was not noted among contemporary Lithuanians (Kasperavičiūtė *et al.* 2004).

Genetic time estimates based on modern mtDNA have suggested that the U5-type arose by mutation about 50,000 to 40,000 years BP (Richards *et al.* 1996) or 55,000 to 30,000 years ago (Soares *et al.* 2010). This age implies that around the glacial maximum 20,000 years BP, U5 types were already present and could have repopulated Central and northern Europe as soon as northern areas were deglaciated. A number of authors have suggested that repopulation started out from the Franco-Cantabrian (Torroni *et al.* 2001; Achilli *et al.* 2004; Tambets *et al.* 2004; Zvelebil and Pettit 2006) and Pontic (the western Caucasus and southern European peninsulas) (Malyarchuk *et al.* 2008) glacial refuges around 15,000 years BP. On the basis of these data, it can be confirmed that the repopulation of northern and eastern Europe with U5 types had indeed occurred by the Late Palaeolithic-Mesolithic period, in agreement with the extrapolation from modern genetic data. Furthermore, the high variability of U5 types observed in the sample (eight different U5 types out of 11 individuals carrying U5) confirms that the mitochondrial U5 lineage had a long time to develop prior to the Mesolithic. The high frequency and diversity of U5 in this sample is mitochondrial evidence for an ancient, shared Mesolithic population stratum that underlies modern Europe and adjacent areas.

The interaction between ancient farmers and hunter-gatherers is also of interest. A qualitative comparison between the northeast European ancient mtDNA and the roughly contemporaneous Central European ancient mtDNA presented in another study (Haak *et al.* 2005) shows that, while the mtDNA in Central Europe consisted of up to 25% N1a-types and 0% U5 or U4-types, the north and east European sample examined contains no N1a-types and is dominated by U5-types. East European ancient DNA thus demonstrates that the early farmers and the hunter-gatherers exchanged female genetic lineages between the two areas only to a very limited extent, if at all.

Indeed, in the recent Lithuanian population (Kasperavičiūtė *et al.* 2004), U5 and U4 haplogroups are infrequent. The most frequent from a total sample of 180 individuals representing all the regions of Lithuania are listed below.

Haplogroup H – 33.3%. Its distribution reflects a second intra-European expansion from the Franco-Cantabrian region circa 13,000 years BP (Achilli *et al.* 2004; Pereira *et al.* 2005).

Haplogroup T – 7.2%. It is generally considered as one of the main genetic signatures of the Neolithic expansions (time of origin circa 10,000 to 12,000 years BP in Fertile crescent) (Genographic project, 2010).

Haplogroup V – 5%. It originated around the western Mediterranean region, circa 13,600 years BP, possibly in Iberia (Soares *et al.* 2009).

Haplogroup U frequency is only 2.8%.

Subclade U4 frequency among modern Lithuanians is 5% (time of origin circa 20,000 years BP), U5 is 0.6%, U5a is 2.8% (time of origin circa 20,000 years BP) (Soares *et al.* 2010), U5a1 is 1.1% (time of origin circa 17,000 years BP) (Soares *et al.* 2010); U5b is 3.3% (time of origin circa 30,000 years BP) (Soares *et al.* 2010), and U5b1 is 1.1% (time of origin circa 25,000 years BP) (Soares *et al.* 2010). The time of origin of U5b2, not found among contemporary Lithuanians, is estimated at circa 23,000 years BP.

In general, the founder analysis of haplogroups H (and its subgroups H1, H3, H5), V and U5b1 (pooled frequency among contemporary Lithuanians 46.6%) indicates that they originated in southwest Europe and expanded after the Ice Age and at the end of the Younger Dryas cold event with several dispersal routes (Soares *et al.* 2010). However, the western (Franco-Cantabrian) Ice Age and Younger Dryas refuge, which is more analysed from a population demographic and genetic point of view (Gamble *et al.* 2005), seems not to be the single source of subsequent repopulation: the

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presence of U4 may be the result of expansions from the eastern (Pontic) refuge (Malyarchuk *et al.* 2008). Concerning the area of the eastern Baltic, both ancient and modern mtDNA data could be indicative of post-LGM repopulation from both refugia.

Conclusions and trends for future research

It may be concluded that the genetic continuity in Europe exemplified by the ancient and European-specific U5 type is recognisable in ancient mtDNA samples spanning over 50,000 years. Both 'western' and 'eastern' refugia remain as plausible candidates of postglacial repopulation of the area. Although exchanges of goods are archaeologically evident during the Stone Age, there were no detectable genetic contacts between the early farmers of Central Europe (*Linienbandkeramik*) and the residual hunter-gatherer groups inhabiting areas to the north and east, at least concerning the female lines. Europeans today seem to have inherited Mesolithic lineages (U4 and U5-types) coming from a common European Mesolithic substrate, whereas the female lines from the early Central European Neolithic farmers (N1a types) have not left a significant impact today. Overall, a pattern is emerging where the genetic lineages of modern Europeans have evidently been resident in Europe since the last Ice Age, and for a time at least, in genetically distinct regional pockets. One possible explanation is that many Neolithic pioneers were replaced by autochthonous groups who successfully adopted Neolithic technologies. The mtDNA homogeneity observed across Europe is a more recent phenomenon, less than 7,000 years old, according to these ancient mtDNA results. We can refer to the third millennium BC, internal European migrations occurring in the Eneolithic that significantly modified the genetic landscape, as a time window little explored by archaeogeneticists (Renfrew 2010). This requires further examination, as it has a bearing on a general issue of archaeogenetics. Moreover, as Gamble *et al.* (2005) pointed out, the simple mapping of genetic lineages on to traditional archaeological cultures often yields unsatisfactory results, as some archaeological cultures might not have genetic signatures. In any case, the imprecise chronology of 'genetic clock' should in the first instance be based on audited archaeological sources.

Perhaps the most important general point that can be drawn from this review might be that 'we have not yet learnt how to interpret the data very effectively' (Renfrew 2010).

Written in English by the author

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Rimantas Jankauskas,
Department of Anatomy, Histology and Anthropology,
Faculty of Medicine, Vilnius University,
Čiurlionio street 21, Vilnius LT-03101
Lithuania
Rimantas.jankauskas@mf.vu.lt

MITOCHONDRINĖ DNR AKMENS AMŽIAUS LIETUVOJE IR GALIMA PIRMŪJŲ GYVENTOJŲ KILMĖ

RIMANTAS JANKAUSKAS

Santrauka

Remiantis naujausiomis publikacijomis straipsnyje aptariami neseniai paskelbti mitochondrinės DNR (mtDNR), išskirtos iš akmens amžiaus palaidojimų Lietuvoje, rezultatai platesniame Europos ir dabartinių Lietuvos gyventojų duomenų kontekste. Šiuo metu manoma, kad bent penki ryškesni procesai – pradinė kolonizacija vėlyvajame paleolite, rekolonizacija po paskutinio ledynmečio ir vėlyvojo driasio atšalimo, neolito plėtra, galimos nedidelio masto vario-bronzos amžiaus migracijos – galėjo palikti pėdsakų dabartinių gyventojų genofonde. Iš keturių lietuviškų mėginių, iš kurių neabejotinai pavyko išskirti mtDNR, vienas (Spiginas 4) priklauso U4 haplogrupei, trys (Donkalis 1, Kretuonas 1 ir 3) – U5b2. Apskritai iš visų 17 Vidurio ir Rytų Europos individų, priskiriamų žemdirbyste nesivertusioms kultūroms (mezolitas ir neolitas tik pagal keramikos buvimo kriterijų, apimančių laikotarpį nuo apie 7800 BC iki 2300 BC), dauguma priklausė „U“ tipui. Nepaprastai aukštas U5 tipų dažnumas (daugiau kaip 45 %) dabar randamas tik tarp Šiaurės Skandinavijos samių (lapių) – greičiausiai artimiausio mezolito medžiotojų-rankiotųjų atitiktens šiuolaikinėje Europoje. Šiuolaikinės mtDNR įvairovė paremtas datavimas leidžia manyti, kad U5 haplogrupė dėl mutacijos atsirado maždaug prieš 50 000–40 000 metų. Toks amžius liudija, kad paskutinio ledynmečio maksimu-

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mo metu (prieš 20 000 metų) U5 tipai jau buvo paplitę ir vėl apgyvendino Vidurio ir Rytų Europą ledynui nuslinkus. Šio antrinio apgyvendinimo šaltiniai galėjo būtų tiek vakarinė (Franko-Kantabrijos), tiek ir rytinė (šiaurinės Juodosios jūros pakrantės) slėptuvės. Tarp šiuolaikinių Lietuvos gyventojų U5 ir U4 haplogrupės yra retos. Remiantis šiais senovinės mtDNR duomenimis, dabartinės Europos mtDNR homogeniškumas yra vėlesnis fenomenas, gali būti mažiau kaip 7000 metų senumo. Galima būtų įtarti trečiąjį tūkstantmetį pr. Kr. – vario–bronzos amžiaus migracijas Europos viduje, kurios padarė ženklią įtaką Europos genetinei struktūrai. Deja, netikslus mtDNR mutacijų datavimas neleidžia daryti kategoriškų išvadų, ir hipotezės pirmiausia turi būti patikrintos remiantis patikimais archeologiniais duomenimis.