# Genetic History and Identity: The Case of Turkey

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With the rise of genetic studies of human history over the last two decades, criticism has arisen among the disciplines traditionally concerned with this subject, i.e. anthropology, archaeology and history. This criticism is concerned with the biological construct of historical communities – whether they be called tribes, (ethnic) groups, migrants or populations – that neglects decades of scholarly discourse on these matters by transferring these entities into primarily biological spheres. This essay is based on a systematic analysis of scientific articles reporting genetic research in the context of the history of what is now Turkey. Analysing the structure, methods and contents of this genetic research from the perspective of an archaeologist and historian, I discuss the issue of past and present identity according to both sampling criteria and the major research question, i.e. the Central Asian origin of the Turks.

Keywords: genetics; history; identity; sampling; Anatolia; Central Asian origin.

#### Introduction

Genetic history, the topic of this thematic volume and the preceding conference in Berlin in October 2015, is but one of several terms – such as Anthropological Genetics<sup>1</sup>, Archaeogenetics<sup>2</sup>, Historical Genetics<sup>3</sup>, Molecular Anthropology<sup>4</sup> or Population Genetics – that attempt to reconstruct not only human evolution but also the history of human populations and peopling processes through genetic information.<sup>5</sup>

Encountering genetic history for the first time several years ago, I did so in an exclusively non-European context. What intrigued me most – to give but one example – was the socio-political dimension of projects that attempted to correlate geographical, linguistic and genetic borders in troubled regions such as the Caucasus,<sup>6</sup> and the societal and political consequences the results might bear. This led me to consider the origin of such research questions, the composition of research teams that pursued them, the institutions that funded them, and the dissemination and use of their results. Additionally, genetics, as part of biotechnologies, plays a slightly different role in advanced developing countries, as Yulia

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<sup>1</sup> See e.g. Sommer, DNA and Cultures of Remembrance.

<sup>2</sup> Renfrew and Boyle, Archaeogenetics.

<sup>3</sup> Renfrew, Archaeology, Genetics and Linguistic Diversity, 446.

<sup>4</sup> See e.g. Destro-Bisol et al., Molecular Anthropology in the Genomic Era.

<sup>5</sup> The coinage of 'genetic history' may be ascribed to a division of the relevant disciplines in German-speaking countries, whereas in English-speaking countries anthropology essentially comprises all the disciplines that are involved in human history, physically, socially and culturally.

<sup>6</sup> Nasidze and Stoneking, Mitochondrial DNA Variation.

Egorova shows with the example of India.<sup>7</sup> The development of science and technology serves not only to become a power to reckon with in the international arena, it is also often allied to nationalist efforts to overcome past humiliations and install a national identity and political ambition.<sup>8</sup> I would like to add here that not only the state but also the general public in advanced developing countries shows a greater openness to new technologies and their applications as a whole as they are regarded part of the modernisation process. It seemed reasonable then to address these questions in relation to the framework of a current nation state and advanced developing country – Turkey.<sup>9</sup>

### Genetic studies in Turkey

For the study at hand, I analysed 24 genetic research articles from high impact journals, published from 1996 to 2016 (Fig. 1), according to two selection criteria: the studies should have (1) exhibited a historical research question, and (2) involved genetic data from Turkey, including studies that relied on genetic data collected by earlier studies or databases of any kind whatsoever. The studies were analysed according to their structure (composition of research teams, funding, place of publication), their methods (sampling, analysis methods, references to historical literature) and their content (research questions, results/interpretations).

- 7 Egorova, The Substance that Empowers. For the analysis of genetic studies on other non-European countries and regions see, for example, MacEachern, Genes, Tribes and African History; Sanchez-Mazas *et al.*, Genetic Focus on the Peopling History of East Asia.
- 8 Ong, Introduction.
- 9 Additionally, in Africa and the Middle East, Turkey has the biggest market for gene expression services that is continually increasing (Parker, 2009-2014 World Outlook, 11, 61. See also TÜBITAK, Special Focus).
- Alkan et al., Whole Genome Sequencing of Turkish Genomes; Arnaiz-Villena et al., HLA Alleles and Haplotypes in the Turkish Population; Berkman et al., Alu Insertion Polymorphisms; Berkman et al., Asian Contribution to the Turkish Population; Calafell et al., From Asia to Europe; Cinnioğlu et al., Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas et al., Geographic Variation in Human Mitochondrial DNA; Di Benedetto et al., DNA Diversity and Population Admixture; Gokcumen et al., Biological Ancestries, Kinship Connections, and Projected Identities; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Machulla et al., Genetic Affinities among Mongol Ethnic Groups; Mergen et al., Mitochondrial DNA Sequence Variation; Omrak et al., Genomic Evidence; Ottoni et al., Mitochondrial Analysis of a Byzantine Population; Ottoni et al., Comparing Maternal Genetic Variation; Quintana-Murci et al., Where West Meets East; Rasterio and Chikhi, Female and Male Perspectives; Richards et al., Tracing European Founder Lineages; Rosser et al., Y-Chromosomal Diversity; Schönberg et al., High-Throughput Sequencing; Torroni et al., A Signal from Human mtDNA; Underhill et al., Phylogenetic and Geographic Structure; Wells et al., Eurasian Heartland; Yunusbayev et al., Genetic Legacy.
- Four studies were not included to this analysis: In one study, aDNA was extracted from 100 bones from a site in Borkuçu, Muğla, dating to various periods of Classical Antiquity; however, the study addressed only technological aspects of the extraction procedure (Vural and Tırpan, Comparison and Development). Similarly, Arslan *et al.* focused on texting sex determination with morphological and genetic methods, extracting sDNA of Bronze Age individuals from Oylum Höyük (Arslan *et al.*, Optimization of aDNA Extraction Protocol). Furthermore, I did not consider another study that lacked a historical research question (Çakır *et al.*, Y-STR Haplotypes in Central Anatolia). This study was only concerned with the method of aDNA extraction. Another study was improved and reprinted in 2009 (Berkman and Togan, Asian Contribution to the Turkish Population), therefore I considered only the latter.

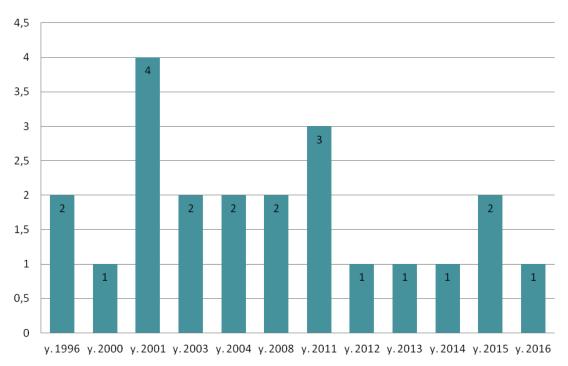


Fig. 1: Dates of publication

## Structure: composition of research teams, funding, place of publication

The composition of research teams was determined according to the authors' institutional affiliation at the time of the study. Eleven studies (46%) had no Turkish participation;<sup>12</sup> nine studies (37%) were conducted by an international project with Turkish participation;<sup>13</sup> and four studies (17%) were solely Turkish projects (*Fig. 2*).<sup>14</sup> Interestingly, no international study has been conducted under Turkish project leadership. This composition of research teams did not change throughout the period investigated.

- 12 Ottoni *et al.*, Mitochondrial Analysis of a Byzantine Population; Ottoni *et al.*, Comparing Maternal Genetic Variation; Quintana-Murci *et al.*, Where West Meets East; Rasterio and Chikhi, Female and Male Perspectives; Richards *et al.*, Tracing European Founder Lineages; Schönberg *et al.*, High-Throughput Sequencing; Torroni *et al.*, A Signal from Human mtDNA; Underhill *et al.*, Phylogenetic and Geographic Structure; Wells *et al.*, Eurasian Heartland; Yunusbayev *et al.*, Genetic Legacy.
- 13 Calafell *et al.*, From Asia to Europe; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups.
- 14 Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Berkman *et al.*, Alu Insertion Polymorphisms; Berkman *et al.*, Asian Contribution to the Turkish Population; Mergen *et al.*, Mitochondrial DNA Sequence Variation.

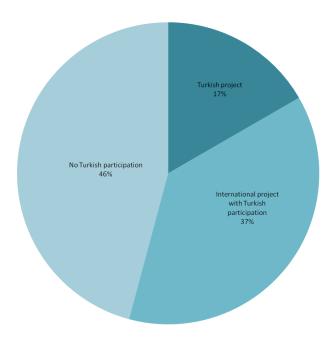


Fig. 2: Composition of research teams

Gathering the data on funding, it became evident that the result appears to be considerably different when including large international projects with many different (and mostly single) national collaborators. Such research projects exhibit research questions that cover at least a continent. As soon as these studies are added to the analysis, a good amount of smaller national or university funding comes in, that supported in nearly all the cases only one of the co-authors and only once. Therefore, I excluded the three studies of this kind from the analysis on funding.<sup>15</sup>

The remaining 21 genetic studies on Turkey were funded by 49 research institutions from thirteen different countries and the European Union (*Table 1, Fig. 3*). Most of the genetic research on Turkey has been funded by Italian institutions: ten funding institutions sponsored five studies. Four of these funding institutions were state-owned (Beni Culturali, Fondo per gli Investimenti della Ricerca di Base, Italian Ministry of Universities, National Research Council of Italy), five were university funds (universities of Ferrara, Pavia, Rome, Urbino and the Italian Consortium of Universities, all of which are public universities) and one was private (*Fondazione Telethon*). Whereas the private institution and the university funds all sponsored only one research project each, the state-owned institution (National Research Council of Italy, *Progetti Ricerca Interesse Nazionale*) funded three or four projects (*Beni Culturali*, Italian Ministry of Universities) respectively.

<sup>15</sup> Rosser *et al.*, Y-Chromosomal Diversity; Underhill *et al.*, Phylogenetic and Geographic Stucture; Yunusbayev *et al.*, Genetic Legacy.

<sup>16</sup> Quintana-Murci et al., Where West Meets East; Rasterio and Chikhi, Female and Male Perspectives.

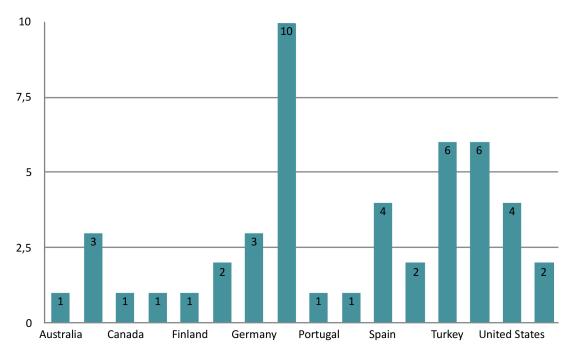


Fig. 3: Funding according to countries (including the European Union)

Six different institutions from Turkey have funded seven different genetic studies of which four were solely Turkish projects<sup>17</sup> and three were international projects with Turkish participation<sup>18</sup>. Four of the Turkish funding institutions were university funds (Ankara University, Boğaziçi University, Hacettepe University and Middle Eastern Technical University), three projects were funded by the national research agency, The Scientific and Technological Research Council of Turkey (*Türkiye Bilimsel ve Teknolojik Araştırma Kurumu, TÜBITAK*) and one by the Turkish State Planning Administration (*Devlet Planlama Teşkilatı, DPT*), one of the most important governmental organizations in Turkey that provides advice to the government on economic, social and cultural state goals. Thus, funding from Turkey derived entirely from state resources.

Six different genetic studies received funding from four different institutions in the United Kingdom,<sup>19</sup> of which the Wellcome Trust sponsored three studies, the Royal Society and the Leverhume Trust two apiece, and the Imperial Cancer Research Fund and the Medical Research Council one each. With the exception of the *Royal Society*, the funding institutions from the United Kingdom are private.

<sup>17</sup> Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Berkman *et al.*, Alu Insertion Polymorphisms; Berkman *et al.*, Asian Contribution to the Turkish Population; Mergen *et al.*, Mitochondrial DNA Sequence Variation.

<sup>18</sup> Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities.

<sup>19</sup> Ottoni *et al.*, Comparing Maternal Genetic Variation; Quintana-Murci *et al.*, Where West Meets East; Richards *et al.*, Tracing European Founder Lineages; Torroni *et al.*, A Signal from Human mtDNA; Wells *et al.*, Eurasian Heartland.

Funding institution	Projects
Academy of Finland (FI)	1
Alfred P. Sloan Foundation (US)	1
Belgian Programme on Interuniversity Poles of Attractions (BE)	2
Beni Culturali (IT)	4
Boğaziçi University Research Funds (TR)	1
CNRS (FR)	1
CNRS/NATO Cooperation (FR)	1
Complutense University (ES)	1
Comunidad de Madrid (ES)	1
Department of Forensic Sciences at Ankara University Medical School (TR)	1
Deutsche Forschungsgesellschaft (DE)	1
Deutscher Akademischer Austauschdienst (DE)	1
Direccion General de Investigacion Científica y Tecnica (ES)	2
ERC Advances Grant (EU)	1
EU Science and Technology Cooperation (EU)	1
Fondo d'Ateneo, University of Pavia (IT)	2
Fondo per gli Investimenti della Ricerca di Base (IT)	1
Fundação para a Ciência e Tecnologia (PT)	2
Grandi Progetti Ateneo Università di Roma "La Sapienzia (IT)"	1
Hacettepe University, Department of Physical Anthropology (TR)	1
Imperial Cancer Research Fund (UK)	1
Italian Ministry of Universities (IT)	4
La Trobe University (AU)	1
Leverhulme Trust (UK)	2
	1
Max Planck Society (DE)  Medical Research Council (UK)	1
Middle Eastern Technical University Research Fund (TR)	2
Ministerio de Educación y Ciencia (ES)	1
Ministry of Science and Technology of the Repulic of Croatia (HR)  National Institutes of Health (US)	6
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National Research Council of Italy (IT)	3
Progretti Ricerca Interesse Nazionale (IT)	3
Research Foundation Flanders (BE)	2
Research Fund of the K.U. Leuven (BE)	2
Romanian Ministry of Research and Technology (RO)	1
Royal Society Visiting Fellowship (UK)	1
Royal Society/NATO Postdoctoral Fellowship (UK)	1
Social Sciences and Humanities Research Council (CA)	1
Spanish Ministry of Educacion (ES)	1
Swedish Foundation for Humanities and Social Sciences (SE)	1
Swedish Research Council (SE)	1
Telethon-Italy (IT)	1
TÜBITAK (TR)	3
Turkish State Planning Administraiton (TR)	1
University of Ferrara (IT)	1
University of Pennsylvania (US)	2
University of Urbino (IT)	1
Wellcome Trust (UK)	4
Wenner Gren Foundation (US)	1

Table 1: Number of projects according to funding institutions

Three Belgian institutions funded two genetic studies<sup>20</sup>, three German institutions sponsored three projects<sup>21</sup>, two French institutions funded two projects<sup>22</sup>, four Spanish institutions supported five studies<sup>23</sup>, two Swedish institutions sponsored one project<sup>24</sup> and four US institutions funded four projects<sup>25</sup>. Funding from Australia, Canada, Croatia, Finland, Portugal and Romania supported in each case only one single project each. Besides solely Turkish projects that were supported only by Turkish institutions, as described above, three more projects received such a national funding, each from a German<sup>26</sup>, a Spanish<sup>27</sup> and a US institution<sup>28</sup>.

All of the 24 studies were published in high impact journals (*Table 2*)<sup>29</sup>. Only one of these journals covers arts and humanities and social sciences (*American Anthropologist*), one journal addresses sciences and social sciences (*American Journal of Physical Anthropology*), while all of the other journals are dedicated solely to the sciences.

Journal	no. of studies
American Anthropologist	1
American Journal of Human Genetics	4
American Journal of Physical Anthropology	2
Annals of Human Genetics	2
BMC Genomics	1
Current Biology	1
Discrete Applied Mathematics	1
European Journal of Human Genetics	3
Human Genetics	1
Journal of Genetics (Indian Academy of Sciences)	1
Molecular Biology and Evolution	1
PLOS Genetics	1
PLOS ONE	1
PNAS (Proceedings of the National Academy of Sciences)	1
Royal Society Open Access	1
Tissue Antigens	2

Table 2: Places of publications and numbers of studies

<sup>20</sup> Ottoni et al., Mitochondrial Analysis of a Byzantine Population; Ottoni et al., Comparing Maternal Genetic Variation.

<sup>21</sup> Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Schönberg *et al.*, High-Throughput Sequencing; Richards *et al.*, Tracing European Founder Lineages.

<sup>22</sup> Quintana-Murci et al., Where West Meets East; Rasterio and Chikhi, Female and Male Perspectives.

<sup>23</sup> Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population; Calafell *et al.*, From Asia to Europe; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Rosser *et al.*, Y-Chromosomal Diversity; Torroni *et al.*, A Signal from Human mtDNA.

<sup>24</sup> Omrak et al., Genomic Evidence.

<sup>25</sup> Calafell et al., From Asia to Europe; Cinnioğlu et al., Excavating Y-Chromosome Haplotype Strata in Anatolia; Gokcumen et al., Biological Ancestries, Kinship Connections, and Projected Identities; Omrak et al., Genomic Evidence.

<sup>26</sup> Schönberg et al., High-Throughput Sequencing, from the Max Planck Society.

<sup>27</sup> Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population, from the Spanish Ministry of Education and the Comunidad de Madrid.

<sup>28</sup> Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, from the National Health Institute.

<sup>29</sup> In accordance with the San Francisco Declaration on Research Assessment (see www.ascb.org/dora/; retrieved 27 August 2016), I refrained from listing the journals' impact factor.

### Methods: sampling, analysis methods, references to historical literature

Sampling was defined not only as the geographical region from which the samples were obtained, but also where and how the researchers acquired the tissue samples or genetic data – whether they were obtained from a tissue or databank, collected at a hospital or were already in the researcher's possession.

Three studies analysed ancient DNA (aDNA),<sup>30</sup> one study combined modern and aDNA by using one of the largest aDNA datasets available.<sup>31</sup> The remaining 20 studies drew their conclusions from modern DNA (*Fig. 4*). Eight of the studies that analysed modern DNA generated it from blood samples only (33%),<sup>32</sup> one study amplified DNA from hair roots (4%),<sup>33</sup> one from a combination of blood and buccal samples (4%)<sup>34</sup> and another one from a combination of blood samples, saliva samples and buccal swab (4%)<sup>35</sup>. Of these 20 studies, one obtained DNA from the Turkish Heart Study,<sup>36</sup> one from the International Society of Genealogy database<sup>37</sup> and another one from two laboratories in Istanbul where the samples were already typed.<sup>38</sup> Five studies did not specify the nature of their samples and combined their data with data from literature,<sup>40</sup> and two studies relied entirely on samples from literature<sup>41</sup>; these nine studies were summarised in one category (42 %).<sup>42</sup> The three studies that analysed aDNA extracted it from excavated bones or bones and teeth respectively (9 %)<sup>43</sup> (*Fig. 5*).

- 30 Omrak *et al.*, Genomic Evidence; Ottoni *et al.*, Mitochondrial Analysis of a Byzantine Population; Ottoni *et al.*, Comparing Maternal Genetic Variation.
- 31 Rasterio and Chikhi, Female and Male Perspectives.
- 32 Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population; Calafell *et al.*, From Asia to Europe; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups; Mergen *et al.*, Mitochondrial DNA Sequence Variation.
- 33 Comas et al., Geographic Variation in Human Mitochondrial DNA.
- 34 Berkman et al., Alu Insertion Polymorphisms.
- 35 Schönberg et al., High-Throughput Sequencing.
- 36 Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 129.
- 37 Underhill et al., Phylogenetic and Geographic Structure, 126.
- 38 Machulla et al., Genetic Affinities among Mongol Ethnic Groups, 293.
- 39 Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Quintana-Murci *et al.*, Where West Meets East; Rosser *et al.*, Y-Chromosomal Diversity (»collections from the authors«); Torroni *et al.*, A Signal from Human mtDNA; Wells *et al.*, Eurasian Heartland.
- 40 Richards *et al.*, Tracing European Founder Lineages; Underhill *et al.*, Phylogenetic and Geographic Structure; Yunusbayev *et al.*, Genetic Legacy.
- 41 Berkman and Togan, Asian Contribution to the Turkish Population; Rasterio and Chikhi, Female and Male Perspective.
- 42 Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Wells *et al.*, Eurasian Heartland.
- Omrak *et al.*, Genomic Evidence; Ottoni *et al.*, Mitochondrial Analysis of a Byzantine Population. Ottoni *et al.*, Comparing Maternal Genetic Variation.

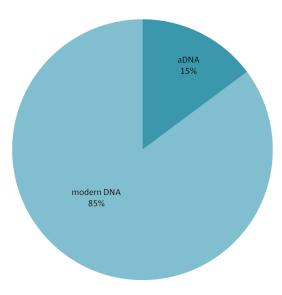


Fig. 4: Percentage of studies with aDNA and modern DNA

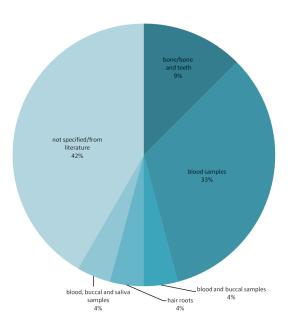


Fig. 5: Provenance of samples

The 'Turkish' feature of DNA samples is generally inferred from the geographic origin of the samples. Information on the geographic origin of samples varies from simply "Turkey« Turkey» or "a large territory of Turkey excluding main cities and coastal areas» or "Anatolia» or specified cities in Anatolia, unspecified villages in Anatolia or specified cities or unspecified cities in Turkey. In several studies, regional aspects formed an additional criterion in the sampling. Three research teams tried to avoid urban areas due to "a higher probability to be recent (im) migration» or to "ensure the autochthony of the sample» when sampling in villages and rural areas. Four studies, on the other hand, collected their samples in urban areas. Three studies added language to geographical features as an expression of Turkishness. Three studies affiliated the Turkishness of their samples primarily or entirely with language, either by excluding non-Turkish speakers or by establishing sampling criteria according to language families and languages. One study assessed the Turkish feature in sampling via self-assessed ancestry of the donors.

Among the 15 studies in which the origin and nature of samples were given seven emphasised the fact that the donors were »not related individuals«<sup>59</sup> or »not related and healthy«<sup>60</sup> respectively. Two studies were careful to generate DNA from healthy individuals.<sup>61</sup> In one

- 44 Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Berkman *et al.*, Alu Insertion Polymorphisms; Calafell *et al.*, From Asia to Europe; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups; Mergen *et al.*, Mitochondrial DNA Sequence Variation; Richards *et al.*, Tracing European Founder Lineages; Schönberg *et al.*, High-Throughput Sequencing; Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Torroni *et al.*, A Signal from Human mtDNA.
- 45 Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Richards *et al.*, Tracing European Founder Lineages; Torroni *et al.*, A Signal from Human mtDNA.
- 46 Calafell et al., From Asia to Europe.
- 47 Machulla et al., Genetic Affinities among Mongol Ethnic Groups.
- 48 Berkman *et al.*, Alu Insertion Polymorphisms; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Mergen *et al.*, Mitochondrial DNA Sequence Variation.
- 49 Comas et al., Geographic Variation in Human Mitochondrial DNA.
- 50 Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Schönberg *et al.*, High-Throughput Sequencing.
- 51 Cinnioğlu et al., Excavating Y-Chromosome Haplotype Strata in Anatolia.
- 52 Calafell et al., From Asia to Europe, 37; Di Benedetto et al., DNA Diversity and Population Admixture, 145.
- 53 Comas et al., Geographic Variation in Human Mitochondrial DNA, 1068.
- 54 Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia, 128; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 129; Schönberg *et al.*, High-Throughput Sequencing, 989; Quintana-Murci *et al.*, Where West Meets East, 828.
- 55 Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture.
- 56 Arnaiz-Villena et al., HLA Alleles and Haplotypes in the Turkish Population, 309.
- 57 Wells  $\it et al.$ , Eurasian Heartland, 10244; Yunusbayev  $\it et al.$ , Genetic Legacy, 5.
- 58 Gokcumen et al., Biological Ancestries, Kinship Connections, and Projected Identities.
- 59 Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population, 309; Di Benedetto *et al.*, DNA Diversity and Population Admixture, 145; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA, 1068; Berkman *et al.*, Alu Insertion Polymorphisms, 12.
- 60 Mergen et al., Mitochondrial DNA Sequence Variation, 40; Quintana-Murci et al., Where West Meets East, 828.
- 61 Calafell *et al.*, From Asia to Europe, 36 ("unaffected families with common Mendelian disorders"); Wells *et al.*, Eurasian Heartland, 10244.

study, the mother's birthplace was additionally recorded,<sup>62</sup> one study documented the birth place of the paternal grandfather,<sup>63</sup> one study the donor's birthplace,<sup>64</sup> one the paternal residency<sup>65</sup> and one study the birthplace of the donors, their parents and their grandparents.<sup>66</sup> Finally, one study tried to avoid donors from ethnic minorities.<sup>67</sup>

In only 15 of 24 studies (68%) were the absolute numbers of samples given.<sup>68</sup> The ratio of Turkish samples to the total samples of these 15 studies is shown in Figure 6, with the exception of Torroni *et al.*'s study, as its total sample number is so large that it would have undermined all the other studies on this illustration.<sup>69</sup>

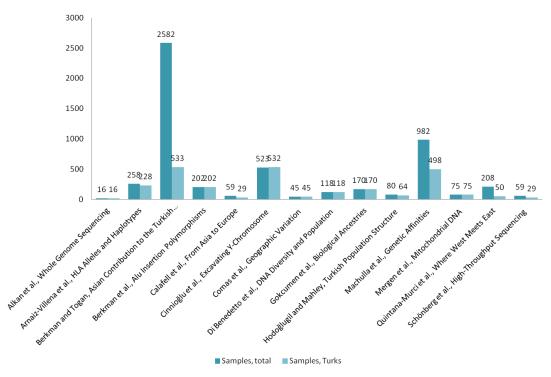


Fig. 6: Ratio of Turkish samples to the total amount of samples

- 62 Comas et al., Geographic Variation in Human Mitochondrial DNA, 1068.
- 63 Rosser et al., Y-Chromosomal Diversity, 1528.
- 64 Calafell et al., From Asia to Europe, 37.
- 65 Cinnioğlu et al., Excavating Y-Chromosome Haplotype Strata in Anatolia, 128.
- 66 Schönberg et al., High-Throughput Sequencing.
- 67 Calafell et al., From Asia to Europe, 36.
- 68 Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population; Berkman and Togan, Asian Contribution to the Turkish Population; Berkman *et al.*, *Alu* Insertion Polymorphisms; Calafell *et al.*, From Asia to Europe; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups; Quintana-Murci *et al.*, Where West Meets East; Schönberg *et al.*, High-Throughput Sequencing; Torroni *et al.*, A Signal from Human mtDNA.
- 69 Torroni *et al.*, A Signal from Human mtDNA, had a panel of 10,365 individuals from 56 populations of western Eurasia and northern Africa of which 606 were Turkish samples; this is about four times more total samples than the 2583 samples of Berkman and Togan, Asian Contribution to the Turkish Population, and about 647 times more samples than the study of Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes (see Fig. 6).

Studies of genetic variation in the Turkish population examined mtDNA sequence variation, polymorphic markers on the Y-chromosome, polymorphic loci on autosomal chromosomes, *Alu* insertions (YAP), HLA (Human leukocyte antigen) allele frequencies and whole genome sequences (*Fig. 7*).

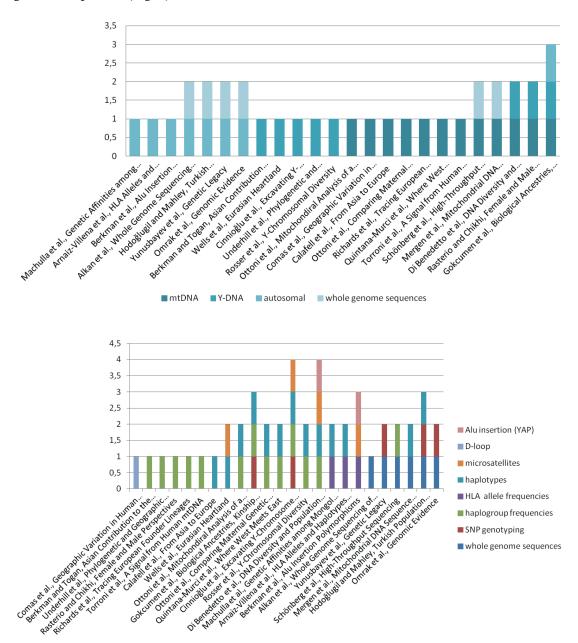


Fig. 7: Elements of DNA analysed

<sup>70</sup> Admixture analysis: 2000-2016; phylogenetic trees: 1996-2014; principal component analysis: 2000-2014.

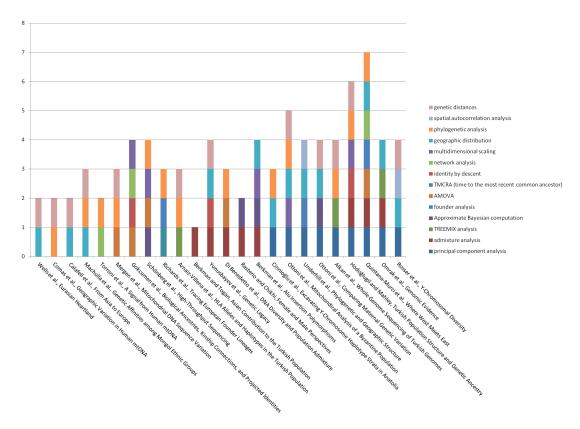


Fig. 8: Biostatistical methods applied

Among biostatistical methods for interpreting the data, three methods were favoured: phylogenetic trees (13 studies), principal component analysis (9 studies) and admixture analysis (8 studies); all of these methods were used throughout the 20 year period investigated (*Fig. 8*).<sup>70</sup>

All of the genetic studies included in this analysis had an underlying historical research question, and accordingly all of them used historical literature, including archaeological and anthropological scholarship. However, there were differences in the quantity and quality of the historical literature. Compared to all of the references in the bibliographies of each study, the percentage of historical references amounts to between 3% and 59%, with an average of 16% (*Fig. 9*). The largest share of historical literature was used in the genetic studies that featured an interdisciplinary research setting; one of them displayed a social-anthropological context,<sup>71</sup> two were embedded in an interdisciplinary project in the context of archaeological excavations and additionally analysed aDNA.<sup>72</sup> One study, which referred to a comparatively great amount of historical literature (27%), consulted somewhat outdated literature as well

<sup>71</sup> Gokcumen et al., Biological Ancestries, Kinship Connections, and Projected Identities: 50% of all references.

<sup>72</sup> Ottoni *et al.*, Comparing Maternal Genetic Variation: 42%; Ottoni *et al.*, Mitochondrial Analysis of a Byzantine Population: 26 %.

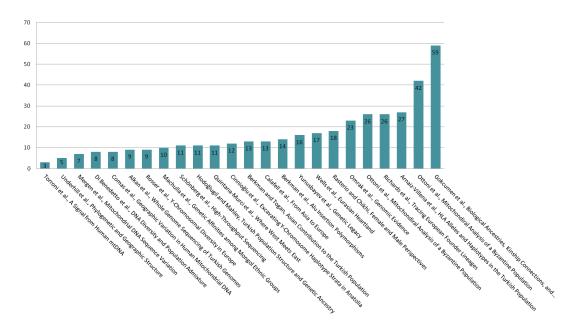


Fig. 9: Quantity of historical references in the studies analysed

as popular scientific literature and exhibited a focus on linguistics.<sup>73</sup> Four of the five studies that used the least amount of historical literature favoured historical-linguistic literature as well.<sup>74</sup> A lower quantity of historical literature, though, does not imply lesser quality: Schönberg *et al.* quoted few references that were all up to date;<sup>75</sup> Alkan *et al.*, made their point using a lot of literature on population exchanges;<sup>76</sup> the Rasteiro and Chikhi and Richards *et al.* studies were in the mid-range in regards the total amount of referred literature, but also exhibited an excellent choice of historical references in terms of up-to-dateness and diversity.<sup>77</sup> However, the way the authors made use of historical references in their interpretation of the genetic data differed considerably. Whereas Rasteiro and Chikhi used genetic data to shed new light on to two traditional historical models of the Neolithic transition, i.e. the demic and the cultural model, Richards *et al.* referred to historical literature mainly in their introduction but hardly included it in their discussion. The same holds true for Hodoğlugil

<sup>73</sup> Arnaiz-Villena et al., HLA Alleles and Haplotypes in the Turkish Population, 316-317.

<sup>74</sup> Torroni *et al.*, A Signal from Human mtDNA; Underhill *et al.*, Phylogenetic and Geographic Structure; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA – in this order according to the amount of historical literature they used.

<sup>75</sup> Schönberg et al., High-Throughput Sequencing.

<sup>76</sup> Alkan et al., Whole Genome Sequencing of Turkish Genomes.

<sup>77</sup> Rasterio and Chikhi, Female and Male Perspectives; Richards et al., Tracing European Founder Lineages.

and Mahley who made only marginal use of their well-chosen literature on the early Middle Ages.<sup>78</sup> Mergen *et al.* seems even more intriguing as the selected historical literature focuses on Turkish identity in the past and in the present.<sup>79</sup> The most popular author referred to was by far Colin Renfrew, being quoted in 12 (of 24) studies with 8 different publications.<sup>80</sup>

#### Content: research questions, results and interpretation

Two major research questions emerged in the analysed genetic studies: the Central Asian origin (10 studies/25%)<sup>81</sup> and the Anatolian impact on the Neolithic transition (6 studies/41,7%)<sup>82</sup> – one study aimed to analyse both topics<sup>83</sup>. The remaining studies exhibited individual research questions (*Table 3*).

Results and interpretations of these two main topics, however, differ considerably, especially on the question of the Central Asian impact on the Turkish gene pool (*Table 4*).

<sup>78</sup> Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry.

<sup>79</sup> Mergen et al., Mitochondrial DNA Sequence Variation, quoted Güvenç, Türk Kimliği, and Roux, Histoire des Turcs.

<sup>80</sup> Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population; Berkman *et al.*, *Alu* Insertion Polymorphisms; Calafell *et al.*, From Asia to Europe; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Omrak *et al.*, Genomic Evidence; Quintana-Murci *et al.*, Where West Meets East; Richards *et al.*, Tracing European Founder Lineages; Rosser *et al.*, Y-Chromosomal Diversity, Schönberg *et al.*, High-Throughput Sequencing; Wells *et al.*, Eurasian Heartland.

<sup>81</sup> Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Berkman and Togan, Asian Contribution to the Turkish Population; Berkman *et al.*, *Alu* Insertion Polymorphisms; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Quintana-Murci *et al.*, Where West Meets East; Wells *et al.*, Eurasian Heartland; Yunusbayev *et al.*, Genetic Legacy.

<sup>82</sup> Omrak *et al.*, Genomic Evidence; Mergen *et al.*, Mitochondrial DNA Sequence Variation; Rasterio and Chikhi, Female and Male Perspectives; Richards *et al.*, Tracing European Founder Lineages; Rosser *et al.*, Y-Chromosomal Diversity.

<sup>83</sup> Cinnioğlu et al., Excavating Y-Chromosome Haplotype Strata in Anatolia.

Research question	Reference	
Investigate the complex population history of Turkey via clustering genetic variation with South European and ancestral East Asian populations	Alkan $\it et al.$ , Whole Genome Sequencing of Turkish Genomes, 1.	
Obtain for the first time the Turkish HLA gene profile and characteristic haplotypes, studying the relative contribution of ancient Mediterraneans to the genetic pool of present day Turkish people. A Kurd group is also analyzed and both ethnic groups will be compared with the HLA genetic structure of neighbouring Armenians and other Mediterraneans	Arnaiz-Villena <i>et al.</i> , HLA Alleles and Haplotypes in the Turkish Population, 309.	
Male genetic contribution from Central Asia to the Turkish population	Berkman and Togan, Asian Contribution to the Turkish Population, 2341.	
Genetic contributions of Central Asia to Anatolia with respect to the Balkans	Berkman et al., Alu Insertion Polymorphisms, 11.	
Gain insight in Turkish and Bulgarian population and knowledge of mtDNA variation in Europe and West Asia	Calafell <i>et al.</i> , From Asia to Europe, 35.	
Understand how the succession of events spanning millennia have contributed to the current genetic composition of Turkey to illuminate the Holocene expansions, the contributions of agriculturalists to the European gene pool and the genetic assessment of Caucasian and Central Asian gene flows	Cinnioğlu <i>et al.</i> , Excavating Y-Chromosome Haplotype Strata in Anatolia, 128.	
Understand the role of Turkey in the history and making of European populations	Comas <i>et al.</i> , Geographic Variation in Human Mitochondrial DNA, 1068.	
Contribution of Central Asian genes to the current Anatolian gene pool	Di Benedetto <i>et al.</i> , DNA Diversity and Population Admixture, 145.	
Investigate the influence of recent historical and social dynamics on local population structure and explore their influence on overall Anatolian genetic diversity	Gokcumen <i>et al.</i> , Biological Ancestries, Kinship Connections, and Projected Identities, 117.	
Investigate Turkish population structure in relation to Central Asia and Europe	Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 128.	
Analyse the relationship between three Mongolian populations and their relationship to Turkish and German samples	Machulla <i>et al.</i> , Genetic Affinities among Mongol Ethnic Groups, 293.	
Obtain information about the distribution of the existing mitochondrial D-loop sequence variations in the Turkish population of Anatolia which is a junction connecting the Middle East, Central Asia and Europe	Mergen et al., Mitochondrial DNA Sequence Variation, 39.	
Anatolian contribution to the European Neolithic gene pool	Omrak <i>et al.</i> , Genomic Evidence 270.	
Investigate whether the historical events of the Plague of Justinian in the 6th century CE, an earthquake in the seventh century CE and the abandonment of Sagalassos around 1200 CE resulted in demographic changes across time	Ottoni et al., Comparing Maternal Genetic Variation, 1.	
Reconstruct the genetic signature potentially left in this region of Anatolia by the many civilizations, which succeeded one another over the centuries until the mid-Byzantine period (13th c. AD)	Ottoni <i>et al.</i> , Mitochondiral Analysis of a Byzantine Population, 571-572.	
Evaluate the effect of Southwest and Central Asian migrations on the current landscape of the Iranian plateau, the Indus Valley and Central Asia	Quintana-Murci <i>et al.</i> , Where West Meets East, 827.	
Test the demic diffusion model and the cultural diffusion model for the Neolithic transition $$	Rasterio and Chikhi, Female and Male Perspectives, 1.	
Investigate the colonization of Europe and estimate the proportion of modern lineages whose ancestors arrived during each major phase of settlement	Richards <i>et al.</i> , Tracing European Founder Lineages, 1232.	
Investigate the demographic history of Europe, discuss demic versus cultural diffusion $% \left( 1\right) =\left( 1\right) \left( 1\right) \left$	Rosser <i>et al.</i> , Y-Chromosomal Diversity, 1526.	
Investigate the genetic structure of three Caucasian (Armenian, Azeri and Georgian) groups and one group from Iran and Turkey each to infer their demographic history	Schönberg <i>et al.</i> , High-Throughput Sequencing, 988.	
Geographic distribution of haplogroup V mtDNA to precise geographical extent and timing of the western recolonization	Torroni et al., A Signal from Human mtDNA, 835.	
Phylogenetic and geographic structure of Y-chromosome haplogroup R1a across Eurasia	Underhill <i>et al.</i> , Phylogenetic and Geographic Structure, 124.	
Reveal traces of historical migrations with a particular focus on Central Asia	Wells <i>et al.</i> , Eurasian Heartland, 10244.	
Prehistoric origin of nomadic Turkish speakers; identify explicit genetic signals shared by all Turkish peoples that have likely descended from putative prehistoric nomadic Turks; trace back the hypothesized "Inner Asian Homeland"	Yunusbayev et al., Genetic Legacy, 3,	

Table 3: Research questions

Result/interpretation	Reference
Genetic variation of the contemporary Turkish population is best described within the context of the Southern European/Mediterranean gene pool, but also shows signatures of relatively recent contribution from ancestral East Asian populations.	Alkan <i>et al.</i> , Whole Genome Sequencing of Turkish Genomes, 1, 8.
The present-day Turkish HLA profiles reflect an old Mediterranean substratum, not very different from the Jewish or Lebanese. The input from the Altai mountains was relatively low. The "out of Anatolia" origin for the Hittites and related people who are considered by some on a linguistic basis as Indo-Europeans has to be doubted according to the results of the study. Turks, Kurds and Armenians are very close genetically and all of them seem to have been living in the area for many millennia, because typical Asian HLA genes are not found. Probably, small different "elite" invaders imposed different languages on these three different groups who originally spoke a similar pre-Indo-European language.	Arnaiz-Villena <i>et al.</i> , HLA Alleles and Haplotypes in the Turkish Population, 314-315.
Anatolia is genetically more closely related with the Balkan populations than to the Central Asian gene pool. Central Asian contribution is only 13%.	Berkman <i>et al.</i> , <i>Alu</i> Insertion Polymorphisms, 11.
Male contribution from Central Asia to the Turkish population with reference to the Balkans is 13% . The genetic similarity between Anatolia and the Balkans seems to be high within males. Comparison of the admixture estimate for Turkey with those of the neighbouring populations point out that the Central Asian contribution was lowest in Turkey.	Berkman <i>et al.</i> , <i>Alu</i> Insertion Polymorphisms, 2341, 2347.
The Turkish sample showed a higher and more diverse polymorphism than the Bulgarians. Bulgarians are close to Western Europeans in terms of levels of internal differentiation and genetic distances, whereas Turks present a close affinity to Middle Eastern populations in terms of higher internal differentiation and shorter genetic distances.	Calafell <i>et al.</i> , From Asia to Europe, 39, 46.
The major components (haplogroups) are shared with European and neighbouring Near Eastern populations (94%) and contrast with only a minor share of haplogroups related to Central Asian (3,4%), Indian (1,5%) and African (1%) affinity. The variety of Turkish haplotypes is witness to Turkey being both an important source and recipient of gene flow.	Cinnioğlu <i>et al.</i> , Excavating Y-Chromosome Haplotype Strata in Anatolia, 127.
The Turkish population presents the shortest genetic distance with the British, but at the same time Turkey is the population with the shortest genetic distance to the Middle East. Once again, Turkey's intermediate genetic position between the Middle East and Europe is shown.	Comas <i>et al.</i> , Geographic Variation in Human Mitochondrial DNA, 1075.
With one exception, the estimated m values converge in suggesting a Central Asian contribution to the current Turkish gene pool of around 30%. An instantaneous input of Asian alleles, accounting for 30% of the current gene pool, means that the 11th century invasion entailed a massive movement of people, females as well as males.	Di Benedetto <i>et al.</i> , DNA Diversity and Population Admixture, 152-153.
Strong structuring of paternal lineages among villages, but not for autosomal and maternal lineages. Based on ethnographic observations, this paternal genetic structuring is delineated from the cultural isolation between the villages, which are rationalized mostly on differences in ancestry and religion.	Gokcumen <i>et al.</i> , Biological Ancestries, Kinship Connections, and Projected Identities, 116.
Turkish population has a close genetic similarity to the Middle Eastern and European populations and some degree of similarity to South Asian and Central Asian populations. Results from the samples collected in Turkey overlapped without a clear sub-population structure, suggesting a rather homogenous and distinct genetic ancestry.	Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 137.
Turks and Germans are equally distant to all three Mongolian populations: lack of a strong genetic relationship between Mongols and Turks despite close relationship of languages and geographic neighbourhood. Despite a shared central Asian history and common linguistic features, Turks and Mongolians are not genetically related.	Machulla <i>et al.</i> , Genetic Affinities among Mongol Ethnic Groups, 292.
Turkic Central Asian, Turkish, British and Finnish populations are placed on one side, German, French, Bulgarian and Greek on the other side of the phylogenetic tree. Turkish samples are at a lower distance from Turkic Central Asian populations and at a somewhat higher distance from European populations. The study provides further support for the intermediate location of Anatolia between Europe and Asia.	Mergen <i>et al.</i> , Mitochondrial DNA Sequence Variation, 45-46.
Genetic similarities to the early European Neolithic gene pool and modern-day Sardinians, as well as a genetic affinity to modern-day populations from the Near East and the Caucasus. Anatoli's central geographic locations appears to have served as a connecting point with other areas of the Near East and Europe, throughout and after the Neolithic.	Omrak et al., Genomic Evidence 270.

Table 4: Results and interpretations

Comparing mtDNA variation in three period groups (Roman, Middle Byzan-Ottoni et al., Comparing Maternal Genetic Varitine and modern), and by simulating possible scenarios inferred by historical ation, 8. and archaeological evidence, the data suggests that concurrently witht the abandonment of the city of Sagalassos in the early thirteenth century CE, the population of the region may have been drastically reduced by almost 90%, mostlikely due to migrations towards farther regions in southwest Anatolia. Furthermore, an earlier but milder contraction in population size may have taken place in the sixth-seventh centuries CE, either owing to the Plague of Justinian and/or an earthquake in the region. High haplotype diversity, all characteristic of West Eurasians. No East/South Ottoni et al., Mitochondiral Analysis of a Byzan-Asian haplogroup M and Sub-Saharan haplogroups; significant maternal getine Population, 573. netic signature of Balkan/Greek populations, as well as ancient Persians and populations from the Italian peninsula. Some contribution from the Levant has also been detected, whereas no contribution from Central Asian population could be ascertained. The Central Asian Uzbeks, Turkmen, and Shugnan tend to be closer to popula-Quintana-Murci et al., Where West Meets East tions from the Anatolian/Caucasus/Iranian regions, rather than to Indus Val-835, 838, ley populations. The eastern Eurasian contribution to the west is negligible. Males and females underwent the same admixture history and both support Rasterio and Chikhi, Female and Male Perspecthe demic diffusion mode. The patterns of genetic diversity found in extant and ancient populations demonstrate that both modern and aDNA support the demic diffusion model. Some differences between male and female markers suggest that the effective female population size was larger than that of the males, probably due to different demographic histories that might be connected to various shifts in cultural practices and lifestyles that followed the Neolithic transition, such as sedentarism, she shift from polygyny to monogamy or the increase of patrilocality. There has been substantial back-migration into the Near East; the majority Richards et al., Tracing European Founder of extant mtDNA lineages entered Europe in several waves during the Upper Lineages, 1251. Palaeolithic: there was a founder effect or bottleneck associated with the Last Glacial Maximum 20,000 years ago, from which derives the largest fraction of surviving lineages; the immigrant Neolithic component is likely to comprise less than one quarter of the mtDNA pool of modern Europeans. Geographic proximity may be a better predictor of Y-chromosomal genetic af-Rosser et al., Y-Chromosomal Diversity, 1537 finity than is language. In the range of this genetic affinity, the Turks lie between the geographically neighbouring but linguistically distant Armenians and Greeks. The randomly sampled complete mtDNA genome sequences indicated ext-Schönberg et al., High-Throughput Sequencing, raordinarily high genetic diversity in the groups from the South Caucasus, 991-993 Iran and Turkey. Central/East Asian groups were found only in a few individuals from the Azeri and Turkish groups, suggesting some Central Asian influence especially on these groups; the low frequency of these mtDNA lineages is in good agreement with previous estimates of low levels of gene flow from Asia into Anatolia. The complete mtDNA genome sequences do reveal some additional genetic similarity between the two Turkish-speaking groups (Azeri and Turks) that was not evident in previous studies. The BSP for the mtDNA sequences from Turkey suggests that the ancestors of the group from Turkey have a different history than the ancestors of the Caucasian and Iranian group in this study. Specifically, these results suggest that the ancestors of the group from Turkey did not expand after the LGM. Haplogroup V is virtually absent in the southern Balkans, Turkey, the Cauca-Torroni et al., A Signal from Human mtDNA, sus and the Near East. The initial episodes of haplogroup R1a diversification likely occurred in the Underhill et al., Phylogenetic and Geographic vicinity of present-day Iran and eastern Turkey. Possibly the R1a lineages ac-Structure, 130. companied demic expansions initiated during the Copper, Bronze and Iron Ages, partially replacing previous Y-chromosome strata. The Turkish and Azeri populations are atypical among Altaic speakers in ha-Wells et al., Eurasian Heartland, 10248. ving low frequencies of M130, M48, M45, and M17 haplotypes. Rather, these two Turkic-speaking groups seem to be closer to populations from the Middle East and Caucasus, characterized by high frequencies of M96- andyorM89-related haplotypes. This finding is consistent with a model in which the Turkic languages, originating in the Altai-Sayanregion of Central Asia and northwestern Mongolia, were imposed on the Caucasian and Anatolian peoples with relatively little genetic admixture-another possible example of elite dominance-driven linguistic replacement.

Table 4: Results and interpretations

Turkic-speaking peoples sampled across the Middle East, the Caucasus, Eastern Europe and Central Asia share varying proportions of Asian ancestry that originate in a single area, southern Siberia and Mongolia. The findings reveal genetic traces of recent large-scale nomadic migrations and map their source to a previously hypothesized area of Mongolia and southern Siberia. Although we report a single admixture date for each population, we note that it is likely that the contemporary Turkic peoples were established through several migration waves. Indeed, Turkic peoples closer to the SSM area (those from the Volga-Ural region and Central Asia) showed younger dates compared to more distant populations like Anatolian Turks, Iranian Azeris, and the North Caucasus Balkars.

Yunusbayev et al., Genetic Legacy, 2, 12.

Table 4: Results and interpretations

#### Discussion

The majority of the studies examined based their research on recent DNA samples (85%),<sup>84</sup> only four studies relied on aDNA,<sup>85</sup> of which one compared modern DNA with aDNA.<sup>86</sup> Two aDNA studies, however, were conducted under the same research project,<sup>87</sup> the excavations of Sagalassos, which are well known for the interdisciplinary evaluation of their results.<sup>88</sup> The fact that most of the studies drew their conclusions about historical migrations from modern genetic data pre-eminently represents the state of technical possibilities. As these are constantly improving, many more genetic studies of aDNA, not only from present-day Turkey, are to be expected in the near future.<sup>89</sup>

Before discussing the contents and results of the analysed genetic studies, I would like to make some methodological remarks on sampling criteria and statistical methods. As the *Race, Ethnicity, and Genetics Working Group* has stated, sampling design can have a critical influence on the result of genetic studies.<sup>90</sup> In many cases of plant and animal population genetics, definition of exact boundaries and particular demes<sup>91</sup> and the assignment of individuals of the species under study to one or another deme is not crucial, and patterns of genetic variability can be detected by a fairly straightforward random sampling procedure in

<sup>84</sup> Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population; Berkman *et al.*, *Alu* Insertion Polymorphisms; Berkman *et al.*, Asian Contribution to the Turkish Population; Calafell *et al.*, From Asia to Europe; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups; Mergen *et al.*, Mitochondrial DNA Sequence Variation; Quintana-Murci *et al.*, Where West Meets East; Richards *et al.*, Tracing European Founder Lineages; Rosser *et al.*, Y-Chromosomal Diversity; Schönberg *et al.*, High-Throughput Sequencing; Torroni *et al.*, A Signal from Human mtDNA; Underhill *et al.*, Phylogenetic and Geographic Structure; Wells *et al.*, Eurasian Heartland; Yunusbayev *et al.*, Genetic Legacy.

<sup>85</sup> Rasterio and Chikhi, Female and Male Perspectives; Omrak *et al.*, Genomic Evidence; Ottoni *et al.*, Comparing Maternal Genetic Variation; Ottoni *et al.*, Mitochondrial Analysis of a Byzantine Population.

<sup>86</sup> Rasterio and Chikhi, Female and Male Perspectives.

<sup>87</sup> Ottoni et al., Comparing Maternal Genetic Variation; Ottoni et al., Mitochondrial Analysis of a Byzantine Population

<sup>88</sup> Cf. the website of the Sagalassos project: www.sagalassos.be (retrieved 30 October 2016).

<sup>89</sup> Destro-Bisol et al., Molecular Anthropology in the Genomic Era, 106-107; Der Sarkissian et al., Ancient Genomics.

<sup>90</sup> Race, Ethnicity, and Genetics Working Group, Use of Racial, Ethnic, and Ancestral Categories, 521.

<sup>91</sup> Originally, a deme describes any specified assemblage of taxonomically closely related individuals. With time, however, the use of the term 'deme' by biologists took on the added implications of a local inbreeding population, a feature not essential to the original proposal of the term (Summers, Demes).

the territory under study. Population geneticists cannot apply these procedures when studying human genetic variation. Researchers interested in the historical analysis of human genetic variability are faced with the problem of associating genetic variability with identifiable modern populations in some explicit way.<sup>92</sup>

Several sampling criteria have been applied in the analysed genetic studies; however, the main criterion was geographical. Some research teams were explicitly cautious to avoid urban or coastal areas for sampling in order to bypass the genetic impact of »recent migrations«.93 The timespan of >recent< has never been indicated in any of the surveyed studies - and I cannot decide what recent means from the geneticist's perspective. Anatolia's prehistoric demography is subject to much discussion among specialists, 94 not least due to missing data such as cemeteries in certain periods or uncertainty upon household size, often due to partial excavation of settlements.95 Even with the availability of written sources, the evidence of population data (censuses, tax registers, land registers) is not as comprehensive and clear as might be desired by historians. Nevertheless, it is a fact that the Ottoman Empire witnessed intensive state-induced population movements in the form of internal migrations and immigration from beyond its borders, from the sixteenth century until its dissolution in 1922. During its expansion from the sixteenth to the eighteenth century, the Ottoman Empire pursued a deportation and resettlement policy based on military, administrative, economic and political considerations, i.e. to fill empty land, to facilitate the dispatching of troops and to supply provisions as well as to disperse heterodox population groups whom the state perceived as religious and political troublemakers.96 From the second half of the nineteenth century to 1913, the Ottoman lands became a shelter for Muslim refugees coming from the Caucasus and the Balkans, i.e. the Crimean Tatars who fled the annexation of their homelands by the Russian Empire in the last quarter of the eighteenth century; the Circassians who were subjected to the Russian policy of expulsion in the 1860s or the Muslim Turks who fled the Balkans in the aftermath of the Russo-Turkish war (1877-1878). 97 A massive deportation of Greeks from Asia Minor started with the Balkan wars (1912-1913), and especially after these wars in 1914.98 From 1906 onwards99, the Committee of Union and Progress (İttihat ve Terakki Cemiyeti) directed internal migration, following a policy of ending existing demographically homogenous regions by mixing the Turkish Muslim population with the non-Turkish Muslim population. It is estimated that nearly one million Balkan refugees, approximately 2 million Kurdish and Turcoman nomads, 5,000 Arab families from

<sup>92</sup> MacEachern, Genes, Tribes, and African History, 361. For a detailed discussion of sampling by reference to the Human Genome Diversity Project see Reardon, *Race to Finish*, chapter 4.

<sup>93</sup> Calafell *et al.*, From Asia to Europe, 37; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA, 1068; Di Benedetto *et al.*, DNA Diversity and Population Admixture, 145.

<sup>94</sup> Düring, Breaking the Bond; see there also references for details. See also Cohen, Implications for the NDT (as well as other chapters in the same volume).

<sup>95</sup> Düring, Early Holocene Occupation.

<sup>96</sup> Seker, Forced Population Movements, 3-4; for examples see ibid., 4.

<sup>97</sup> Şeker, Forced Population Movements, 5.

<sup>98</sup> Dundar, Settlement Policy, 35.

<sup>99</sup> The Committee of Union and Progress was already active between 1895 and 1897, being concerned with the Armenian question, see Dundar, Settlement Policy, 34.

Syria, nearly 1,5 million refugees from eastern Anatolia, some Arab refugees from Tripoli and Benghazi whose number is unknown, nearly 400,000 new Balkan refugees and the Circassians who escaped from Syria, were displaced. To summarise, this means that more than one third of the Muslim population of Asia Minor was transferred from their original habitat to places far away. If the massive reduction of the non-Muslim population, i.e. nearly 1,2 million Greeks and more than 1,5 million Armenians, is added to this number, it is evident that only one half of Asia Minor's population was displaced by the Committee's policy. 100

Additionally, individual migration in the Ottoman Empire has to be considered: peasants, for instance, migrated for survival, for under the conditions of the fifteenth-and sixteenth-centuries, mountainous regions and islands typically did not produce enough to feed their populations. Besides seasonal migrations, some migrants who had left their villages for longer, ultimately found their way back to their home villages, while others stayed away for good, transferring their families to the localities where they had found a source of livelihood. Some permanent migrants married local women and rapidly became part of the society into which they had entered.<sup>101</sup> Finally, from the Neolithic period and well into the twentieth century, a good part of the population in present-day Turkey consisted of nomads, semi-nomadic pastoralists and peripatetic nomads.

Lastly, it should be mentioned here that Turkic tribes already settled throughout the thirteenth and fourteenth centuries in Byzantine lands, adopted Christianity and Greek language, married local Greeks or Slavs and thus entered Byzantine society. Rustam Shukurov describes this Turkic minority as the »Byzantine Turks«, a term that was originally coined by the Ottomans in the first half of the fifteenth century. De do not know the total extent of this Turkic settlement and all the regions of the Byzantine Empire it affected. However, the fact that this minority is well documented in Byzantine sources shows that ethnocultural realities were already more complex in the thirteenth and fourteenth centuries than they are presented in the binary model that many scholars use to describe relations between Greeks and Turks. Thus, genetic admixture took place in a more continuous and less sudden way than the conception of medieval nomadic invasions would suggest. Taking into account only the known migrations of the last five centuries, any suggested autochthony of samples taken in rural areas of present-day Turkey seems illusory.

Of all the analysed genetic studies, it was only Gokcumen *et al.* who considered the more recent Turkish migration history in greater detail and contrasted it with the self-assigned identity of their donors. Thus, they could show that, at the village level, paternal genetic diversity is structured among settlements, whereas maternal genetic diversity is distributed more homogenously, reflecting strong patrilineal cultural traditions that transcend larger ethnic and religious structures. Local ancestries and origin myths, rather than ethnic or religious affiliations, delineate the social boundaries and projected identities among the villages.<sup>104</sup>

<sup>100</sup> Dundar, Settlement Policy, 39. For the population exchange with Bulgaria in 1913, see also İçduygu *et al.*, Politics of Population, 364. For the population exchange between Greece and Turkey in 1923, see Hirschon, *Crossing the Aegean*.

<sup>101</sup> Faroqhi, Ottoman Population, 394.

<sup>102</sup> Shukurov, Byzantine Turks, 9.

<sup>103</sup> Shukurov, Byzantine Turks, 9.

<sup>104</sup> Gokcumen et al., Biological Ancestries, Kinship Connections, and Projected Identities, 119-121.

Other research teams accomplished their sampling in urban areas<sup>105</sup> which have been and still are subject to massive internal and external migrations since at least the formation of the Turkish Republic.<sup>106</sup> In all cases this choice seems to have been a pragmatic one as the investigators drew on blood samples from other studies,<sup>107</sup> simply asked the staff and students of the medical department on site to donate samples,<sup>108</sup> or both.<sup>109</sup> Given the rural migration in the last centuries, the decision to collect available blood samples in urban areas amounts to the same thing.

Five studies categorised their samples as Turkish according to language. Linguistic models and archaeological models that often originated from them influenced genetic history from its very beginnings; and indeed, four of the five studies date to the early period of genetic research on human history. Already Rosser *et al.* have shown in the same early period of genetic research that geographic proximity – at least in regards to the Y-chromosome – may be a better predictor for genetic affinity. In the same study, Rosser *et al.* made another important point: the Altaic language of the Turks was acquired as a result of Turkic invasions from the eleventh to the fifteenth century, and if this language is believed to be acquired by elite dominance, the genes of populations like the Turks are unlikely to be separated from surrounding populations by genetic barriers.

Gokcumen *et al.* have already stated that Anatolia was a multilingual region and that it is not only likely but is also historically documented that a considerable number of communities changed their language over the past centuries. Language concerns in regard to homogenisation of language and proper Turkish started already with the *Tanzimat* period, the process of Westernisation that began in 1839. The foundation of the Republic of Turkey that necessitated the process of forming a national consciousness, and language was used as a significant instrument to create Turkishness as a collective identity. A remarkable example

<sup>105</sup> Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia, 128; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 129; Schönberg *et al.*, High-Throughput Sequencing, 989; Quintana-Murci *et al.*, Where West Meets East, 828.

<sup>106</sup> İçduygu et al., Politics of Population in a Nation-Building Process

<sup>107</sup> Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 129, from the Turkish Heart Study; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups, 293 used samples »collected by two other studies, available from two laboratories in Istanbul where the samples were already typed.«

<sup>108</sup> Arnaiz-Villena et al., HLA Alleles and Haplotypes in the Turkish Population.

<sup>109</sup> Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia, used 359 blood samples from blood banks, 61 from paternity clinics, 103 from staff and students enrolled at Istanbul University.

<sup>110</sup> Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population, 309; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Wells *et al.*, Eurasian Heartland, 10244; Yunusbayev *et al.*, Genetic Legacy, 5.

<sup>111</sup> Cavalli-Sforza, Genes, Peoples, and Languages; Renfrew, Archaeology, Genetics and Linguistic Diversity.

<sup>112 1996:</sup> Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; 2001: Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Wells *et al.*, Eurasian Heartland. Yunusbayev *et al.*, Genetic Legacy, is an exception as the study was published in 2015.

<sup>113</sup> Rosser et al., Y-Chromosomal Diversity, 1537.

<sup>114</sup> Rosser et al., Y-Chromosomal Diversity, 1528.

<sup>115</sup> Gokcumen et al., Biological Ancestries, Kinship Connections, and Projected Identities, 127.

<sup>116</sup> Aydıngün and Aydıngün, Role of Language, 416; see there also for details on language policy before the formation of the Turkish Republic.

is the »Citizen, Speak Turkish!« (»Vatandaş, Türkçe konuş!«) campaign during the first two decades of the Turkish Republic that aimed at putting pressure on non-Turkish speakers to speak Turkish in public.117 Starting with an initiative of students of the Istanbul University to which the Ministry of Interior granted permission, the Commission for the Protection and Expansion of Turkish Language was formed. This commission arranged newspaper announcements, the instalment of posters and above all public meetings that frequently culminated in outbursts of violence. This campaign was a minor event in the history of the Turkish Republic, but the fact that it was initiated by the state and accomplished by the people it could mobilise, shows that language homogenisation was also a concern of the general public. 118 The »Citizen, Speak Turkish!« campaign was but one of the many state-induced means to spread Turkish language throughout Anatolia (and Thrace, too, of course)119 in the course of the »Turkification« (Türklestirmek) policy. During the 1920s and especially the 1930s, many municipalities imposed fines on those who did not speak Turkish.<sup>120</sup> In 1926, the parliament passed a law that made use of the Turkish language compulsory in all correspondence among corporations; in 1931, the state required all Turkish children to have their primary education in Turkish; in 1934, the Law of Surnames required all citizens to take Turkish surnames; names of cities, towns and villages were subsequently changed into Turkish.<sup>121</sup>

Therefore, it might be more valuable to document the languages the grandparents and great-grandparents of the donors spoke (and not their birthplaces) when sampling current Turkish population according to the language spoken.

The criteria of geography and language may suggest that the assumption of ethnicity mattered in some of the studies. Hodoğlugil and Mahley queried their donors about their ethnicity and included only participants who indicated Turkish or Kyrgyz ethnicity in their study.<sup>122</sup> Ethnic groups in Mongolia were the point of departure for Machulla *et al.*'s study; similarly this was the case in Arnaiz-Villena *et al.*'s study that invested the genetic relatedness of Turks, Kurds and Armenians.<sup>124</sup> Torroni *et al.* equated geographic and ethnic origin.<sup>125</sup> Calafell *et al.* applied ethnic criteria when excluding sindividuals from ethnic minorities« from their study.<sup>126</sup> In contrast, some studies explicitly rejected any ethnic assignment of their donors. Alkan *et al.* included their donors sirrespective of their mother-tongue/ethnicity« by referring to all of their donors »collectively as Turkish«.<sup>127</sup> Di Benedetto *et al.* mentioned in their sampling description that they did not record any self-assigned ethnic affiliations« of their donors.<sup>128</sup>

<sup>117</sup> Aslan, »Citizen, Speak Turkish!«.

<sup>118</sup> Aydıngün and Aydıngün, Role of Language, 267.

<sup>119</sup> N.N., Les Israélites de Turquie ; Bali, 1934 Trakya Olayları, 12-13, 170.

<sup>120</sup> Başak, Citizenship and Identity in Turkey, 61.

<sup>121</sup> Aydıngün and Aydıngün, Role of Language, 252, 265.

<sup>122</sup> Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 129.

<sup>123</sup> Machulla et al., Genetic Affinities among Mongol Ethnic Groups, 292.

<sup>124</sup> Arnaiz-Villena et al., HLA Alleles and Haplotypes in the Turkish Population, 309.

<sup>125</sup> Torroni et al., A Signal from Human mtDNA, 845.

<sup>126</sup> Calafell et al., From Asia to Europe, 36.

<sup>127</sup> Alkan et al., Whole Genome Sequencing of Turkish Genomes, 3.

<sup>128</sup> Di Benedetto et al., DNA Diversity and Population Admixture, 146.

The bulk of the analysed genetic research was done on the genetic affiliations of modern populations with the investigation of prehistoric and historic relationships among modern human groups in mind. Genetic studies of Turkey cover a time span of nearly two decades. Methodologies have continually developed throughout this period, so that different methods were applied. As an archaeologist and historian not trained in genetics, I cannot judge to what extent the results of classical methods (from blood protein polymorphisms and gene frequencies) and molecular methods (DNA sequencing, genomics) can be compared with one another.<sup>129</sup>

In order to interpret modern genetic data for historical and prehistoric periods, they are extrapolated with the help of statistical methods. These statistical methods have already been subject to criticism. One issue concerns the sample size and therefore the studies' relevance and significance.<sup>130</sup> In all of the genetic studies, the proportion of Turkish samples when compared with the Turkish population in total<sup>131</sup> is at best a per mille value with at least two zeros after the decimal place (Table 5). Robert V. Krejce and Daryle W. Morgan provided, besides the necessary formula, a table for determining sample size from a given population, according to which the sample size for a population size of 1,000,000 has to amount to 384 or more. However, they noted that as the population increases the sample size increases at a diminishing rate and remains relatively constant at slightly more than 380 cases. 132 Based on the total population size of 74,525,696, 133 with a margin of error 134 of 5% and a confidence level of 95%, the recommended sample size for the current Turkish population amounts to 385. 135 In terms of percentage, 385 samples constitute 0,00052% or 0,0052‰ of the total Turkish population respectively. Of the 15 studies which stated the absolute number of their samples, four studies exhibited this recommended sample size,136 whereas eleven studies did not. 137 In the cases with the smallest sample sizes, this lead to margins of error of 24,5% 138, 18, 2%139 and 14, 61%140. This margin of error, however, refers to the current population and I frankly assume that it must increase retrospectively to time when the data is retrojected. 141

<sup>129</sup> For an example of the incomparability of methods that causes a problem when comparing results, see Rasterio and Chikhi, Female and Male Perspectives, 5-6, who explained this in detail.

<sup>130</sup> For early remarks on this topic see Pohl, Identität und Widerspruch, 26.

<sup>131</sup> Turkish Statistical Institute, Population and Housing Census: total 74,525,696.

<sup>132</sup> Krejcie and Morgan, Determining Sample size for Research Activities, 607, table 1.

<sup>133</sup> Turkish Statistical Institute, Population and Housing Census: total 74,525,696.

<sup>134</sup> The margin of error expresses the amount of random sampling error in a survey's result.

<sup>135</sup> Cf. The sample size calculator: www.raosoft.com/samplesize.html (retrieved on 30 October 2016).

<sup>136</sup> Berkman and Togan, Asian Contribution to the Turkish Population; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups; Torroni *et al.*, A Signal from Human mtDNA.

<sup>137</sup> Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Arnaiz-Villena *et al.*, HLA Alleles and Haplotypes in the Turkish Population; Berkman *et al.*, Alu Insertion Polymorphisms; Calafell *et al.*, From Asia to Europe; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry; Mergen *et al.*, Mitochondrial DNA Sequence Variation; Quintana-Murci *et al.*, Where West Meets East; Schönberg *et al.*, High-Throughput Sequencing.

<sup>138</sup> Alkan et al., Whole Genome Sequencing of Turkish Genomes.

<sup>139</sup> Calafell et al., From Asia to Europe.

<sup>140</sup> Comas et al., Geographic Variation in Human Mitochondrial DNA.

<sup>141</sup> Pohl, Identität und Widerspruch, 27, made the same assumption. See also Geary and Veeramah in this volume.

Reference	Samples, total	Samples, Turks	% Turkish population
Alkan et al., Whole Genome Sequencing	16	16	0,00021
Arnaiz-Villena et al., HLA Alleles and Haplotypes	258	228	0,00359
Berkman and Togan, Asian Contribution to the Turkish Population	2582	533	0,00715
Berkman et al., Alu Insertion Polymorphisms	59	29	0,00039
Calafell et al., From Asia to Europe	59	29	0,00039
Cinnioğlu et al., Excavating Y-Chromosome	523	532	0,00714
Comas et al., Geographic Variation	45	45	0,00064
Di Benedetto et al., DNA Diversity and Population	118	118	0,00158
Gokcumen et al., Biological Ancestries	170	170	0,00228
Hodoğlugil and Mahley, Turkish Population Structure	80	64	0,00086
Machulla et al., Genetic Affinities	982	498	0,00888
Mergen et al., Mitochondrial DNA	75	75	0,00010
Quintana-Murci et al., Where West Meets East	208	50	0,00067
Schönberg et al., High-Throughput Sequencing	59	29	0,00039
Torroni et al., A Signal from Human mtDNA	10365	606	0,00813

Table 5: Ratio of Turkish samples to the total Turkish population

This assumption raises the issue of time depth in data simulation. The timing of biological processes that brought genetic variation into being is entirely unknown<sup>142</sup> and is instead estimated with the help of computer simulation or bioinformatics respectively. Sometimes it seems the cart is put before the horse when genetic data is used to date demographic events instead of being interrogated as to whether it can supply proof of that kind. Richards *et al.*, e.g., used the age of mtDNA mutations and haplogroups to date major demographic events.<sup>143</sup> This kind of approach has been criticised as it can lead to misinterpretation of the data.<sup>144</sup>

In the studies analysed, three methods of data simulation were favoured: phylogenetic trees (13 studies), principal component analysis (9 studies) and admixture analysis (8 studies); each of these methods was used throughout the 20 year period investigated. Marianne Sommer has argued extensively the problematic nature of phylogenetic trees and their mappings which freeze the hierarchical kinship systems that are meant to represent a state before great historical population movements. Previously, Colin Renfrew called attention to the problem of correlating gene frequency maps to long-term population history as it is not known how stable spatial structures in gene frequencies are through time, even when populations are relatively isolated. On the other hand, some of these genes and their phenotypes may not be adaptively neutral, so that some of the variation is to be associated with environmental variables. Furthermore, the spatial aspect of mating patterns may vary at different periods of history and thus influence the gene flow. Finally, a gene frequency map presents a contemporary map. Although it is agreed that early demographic processes may have had a determining impact on genetic distributions, it is unknown so far which gene frequencies are pertinent. 146

<sup>142</sup> MacEacher, Genes, Tribes, and African History, 360.

<sup>143</sup> Richards et al., Tracing European Founder Lineages.

<sup>144</sup> Rasterio and Chikhi, Female and Male Perspectives, 5-6; see there also for further references concerning critique.

<sup>145</sup> Sommer, Population-Genetic Trees. See also Pohl, Identität und Widerspruch, 26, who critically discussed genetic mapping some 15 years earlier.

<sup>146</sup> Renfrew, Archaeology, Genetics and Linguistic Diversity, 463.

Geoffery Anderson Clark has described the way in which principal components analysis is used to establish correlation coefficient matrices for patterns in genetic polymorphisms in order to isolate a number of principal components, expressed geographically, and which are interpreted as time-successive, quasi-historical migration events in the form of a »post-hoc accommodative argument« that develops explanations after an analysis is completed to account for patterns in the data.<sup>147</sup>

Furthermore, differences in patterns of exogamous marriage and post-marital residence can have significant effects on genetic distributions and genetic drift. I estimate that this should be considered in data simulation, especially when extrapolating data to past centuries or millennia. Turkey is a country with a high level of consanguinity. The rate of cousin marriage was found to be at 28,4% in 1969, 149 24,53% in 1983, 150 23,06% in 1987, 151 and 22% in 2003. Regional differences in this rate, however, may vary considerably; the study by Akbayram *et al.* for the Lake Van region, for instance, even showed a percentage of 34,4% cousin marriages of which 75% were first degree cousin marriages. Interestingly, Hodoğlugil and Mahley considered consanguinity in the interpretation of their results when detecting an extended homozygosity in a locus with two identical alleles in Middle Eastern and South Asian populations; however, they excluded Turkey (besides Central Asia, Europe and Northeast Asia) from these considerations as these countries showed less homozygosity in terms of number and size. Mergen *et al.* interpreted the differences they observed in the nucleotide pairwise distributions between Eastern and Western Anatolia as being due to consanguinity and migration events.

Besides consanguinity, polygyny looks back at a rich history on Anatolian grounds: Walter Scheidel described the conditions in the Greco-Roman world as prescriptively universal monogamous marriages that co-existed with (male) resource polygyny, especially for elites; and stated the same for the pre-Islamic and Islamic Middle East. Laura Betzig has argued a close relationship between stratification, despotism and polygyny in early agrarian societies that are relevant for prehistoric periods in Turkey.

With the development of bioinformatics in the last decade, computer simulation programs became increasingly complex, matching modern genetic, historical and ecological data sets, modelling life cycles, mating systems and even phenotypes, even if they still have a wide array of options, capabilities, limitations, input formats and assumptions. <sup>159</sup> For

<sup>147</sup> Clark, Comment on MacEachern, 372.

<sup>148</sup> Renfrew, Archaeology, Genetics and Liguistic Diversity, 471. See, especially, the groundbreaking study of Cavalli-Sforza *et al.*, *Consanguinity, Inbreeding and Genetic Drift in Italy*.

<sup>149</sup> Şayli, Anadolu'nun genetik yapısı, 1.

<sup>150</sup> Başaran, Anadolu'nun genetik yapısı, 5.

<sup>151</sup> Ulusoy and Tunçbilek, Consanguineous marriage in Turkey.

<sup>152</sup> Koc, Prevalences and Sociodemographic Correlates; Ulusoy and Tunçbilek, Consanguinity in Turkey in 1988.

<sup>153</sup> Akbayram et al., Frequency of Consanguineous Marriage, 212.

<sup>154</sup> Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 139.

<sup>155</sup> Mergen et al., Mitochondrial DNA Sequence Variation, 45.

<sup>156</sup> Scheidel, A Peculiar Institution?

<sup>157</sup> Scheidel, Sex and Empire.

<sup>158</sup> Betzig, Sex, Succession, and Stratification.

<sup>159</sup> Hoban et al., Computer Simulations, 110-111.

historical interpretation, computer simulation programs are used for statistical inference and evaluation of statistical genetics methods, and some of these simulators consider polygamy, polyandry and other complex mating behaviours. <sup>160</sup> Of all the analysed studies, only Rita Rasteiro and Lounès Chikhi considered polygyny (and the shift to monogamy) in the interpretation of their results. <sup>161</sup>

Thematically, the genetic studies on populations in Turkey analysed in this study are concerned with two main topics: the Central Asian origin of the Turks (10 studies) and the migrations in the course of the Neolithic transition (6 studies). In accordance with these topics, genetic studies were conducted on two different scales, a macro scale (covering an area of several countries, one continent or more) and a micro scale (covering regions of Turkey or Turkey entirely). In macro-scale studies, Turkish DNA samples constituted only one sample category among many, whereas some micro-scale studies gathered DNA samples solely from Turkey and compared them to published data (see Fig. 6). This observation, though, is restricted to the 15 studies that provided information about the number and origin of their samples.

I would like to discuss here the topic of the Central Asian origin of the Turks that is of greater interest for the readers of this journal. Various estimates exist on the proportion of gene flow associated with the arrival of Central Asian Turkic speaking people to Anatolia. Di Benedetto *et al.* compared mtDNA control region sequences and one binary and six STR Y-chromosome loci analyzed in 118 Anatolian samples with those found in Central Asia and suggested roughly a 30% Central Asian impact on the Anatolian gene pool. According to Di Benedetto *et al.*, these data reflect the occurrence of a single admixture event related to the flow of Central Asian mtDNA into the Anatolian gene pool. <sup>164</sup>

In contrast, according to Berkman *et al.*, who investigated 10 *Alu* insertion polymorphisms, the Central Asian contribution to the Anatolian gene pool amounts only to 13%. <sup>165</sup> Cinnioğlu *et al.* detected in their study of Y-chromosome variation only a minor share of Anatolian haplogroups related to Central Asian that amounts to 3,4%. <sup>166</sup> Hodoğugil and Mahley attested the Turkish population only had »some degree of similarity to South Asian and Central Asian populations:«<sup>167</sup> depending upon the number of chosen components of parental ancestry,

<sup>160</sup> Hoban *et al.*, Computer Simulations, 119. See *ibid.* also for simulation programs on historical events in general, esp. 116, Fig. 3.

<sup>161</sup> Rasterio and Chikhi, Female and Male Perspectives, 6-7.

<sup>162</sup> Machulla *et al.*, Genetic Affinities among Mongol Ethnic Groups; Quintana-Murci *et al.*, Where West Meets East; Rasterio and Chikhi, Female and Male Perspectives; Richards *et al.*, Tracing European Founder Lineages; Rosser *et al.*, Y-Chromosomal Diversity; Schönberg *et al.*, High-Throughput Sequencing; Torroni *et al.*, A Signal from Human mtDNA; Underhill *et al.*, Phylogenetic and Geographic Structure; Wells *et al.*, Eurasian Heartland; Yunusbayev *et al.*, Genetic Legacy.

<sup>163</sup> Alkan *et al.*, Whole Genome Sequencing of Turkish Genomes; Berkman *et al.*, *Alu* Insertion Polymorphisms; Cinnioğlu *et al.*, Excavating Y-Chromosome Haplotype Strata in Anatolia; Comas *et al.*, Geographic Variation in Human Mitochondrial DNA; Di Benedetto *et al.*, DNA Diversity and Population Admixture; Gokcumen *et al.*, Biological Ancestries, Kinship Connections, and Projected Identities; Mergen *et al.*, Mitochondrial DNA Sequence Variation.

<sup>164</sup> Di Benedetto et al., DNA Diversity and Population Admixture, 144.

<sup>165</sup> Berkman et al., Alu Insertion Polymorphisms, 11.

<sup>166</sup> Cinnioğlu et al., Excavating Y-Chromosome Haplotype Strata in Anatolia, 127, 136.

<sup>167</sup> Hodoğlugil and Mahley, Turkish Population Structure and Genetic Ancestry, 137.

the Central Asian contribution amounted between 9% (K = 4) and 15% (K = 3). <sup>168</sup> Several other studies confirm the small Central Asian impact on the Anatolian gene pool, even if they did not express it in numbers. Quintana-Murci *et al.*, analysing mtDNA, found "the eastern Eurasian contribution to the west negligible.«<sup>169</sup> Schönberg *et al.*, who genotyped complete mtDNA sequences, found "Central/East Asian groups only in a few individuals from the Azeri and Turkish groups, suggesting some Central Asian influence especially on these groups« that, nevertheless, confirmed "previous estimates of low levels of gene flow from Asia into Anatolia.«<sup>170</sup> Machulla *et al.*, who found in their analysis of five HLA loci that "Turks and Germans are equally distant to all three Mongolian populations and stated that "despite a shared Central Asian history and common linguistic features, Turks and Mongols are not genetically related.«<sup>171</sup> Mergen *et al.* who analysed mitochondrial D-loop region sequence variations abstained from a judgment of their data when ascertaining that the "Turkish samples are at a lower distance from Turkic Central Asian populations and at a somewhat higher distance from European populations.«<sup>172</sup>

Several studies agree with traditional Turkish historiography concerning the timing of Central Asian genetic contribution. Alkan *et al.* state that the genetic variation of contemporary Turkish population is best described within the context of the Southern European/Mediterranean gene pool; however, it »also shows signatures of relatively recent contribution from ancestral East Asian populations.«<sup>173</sup> Wells *et al.*, who found the »Turkish and Azeri populations atypical among Altaic speakers but closer to populations from the Middle East and the Caucasus«, applied a linguistic model of the Turkic language expansion and concluded for these both groups »a possible example of elite dominance-driven linguistic replacement.«<sup>174</sup>

Of great interest are the aDNA results on this matter. In their analysis on the eleventh to thirteenth century Byzantine population of Sagalassos, Ottoni *et al.* could not ascertain any genetic contribution from Central Asia.<sup>175</sup> However, the result of a single Byzantine site might not be representative for the rest of the Byzantine Empire for which a continuous Turkic immigration since the thirteenth century is attested, as mentioned above.

Aram Yardumian and Theodore G. Schurr have discussed some of the genetic studies mentioned here – as well as the genetic evidence from Central Asia – at length and contrasted it with the linguistic historical and archaeological evidence. Based on this evidence, they concluded that the genetic profile of present-day Anatolians is not the product of mass westward migrations from Central Asia and Siberia nor of small-scale migrations into an emptied sub-continent, but instead one of small-scale, irregular punctuated migrations that engendered large-scale shifts in language and culture among the diverse autochthonous inhabitants.<sup>176</sup>

 $<sup>168 \,\,</sup> Hodoğlugil \,\, and \,\, Mahley, Turkish \,\, Population \,\, Structure \,\, and \,\, Genetic \,\, Ancestry, \, 128.$ 

<sup>169</sup> Quintana-Murci et al., Where West Meets East, 838.

<sup>170</sup> Schönberg et al., High-Throughput Sequencing, 991-993.

<sup>171</sup> Machulla et al., Genetic Affinities among Mongol Ethnic Groups, 292.

<sup>172</sup> Mergen et al., Mitochondrial DNA Sequence Variation, 45-46.

<sup>173</sup> Alkan et al., Whole Genome Sequencing of Turkish Genomes, 1, 8.

<sup>174</sup> Wells et al., Eurasian Heartland, 10248.

<sup>175</sup> Ottoni et al., Mitochondrial Analysis of a Byzantine Population, 571.

<sup>176</sup> Yardumian and Schurr, Who Are the Anatolian Turks?

The Central Asian ancestry of the Turks is anything but a new research topic in the history of the late Ottoman Empire and the Republic of Turkey. Due to the influence of European Turkologists, particularly Léon Cahun and his Introduction to the History of Asia in which he portrayed a just, egalitarian, well-organised society in the Turkish states of Central Asia, and to the influx of intellectuals from the Crimea, Azerbaijan and Central Asia, pre-Ottoman Turks appeared on the scene for the first time in the late nineteenth century. The revolutionary Young Turks (1908-1918) turned from Ottomanism to pan-Turkism and introduced a new period of history writing, promoting Ottoman history on a scientific basis. In the years before the First World War, interest in pre-Ottoman Turks and in early Turkish Central Asian states increased again.<sup>177</sup> Most notably, Rıza Nur emphasized the Central Asian origin of the Turks in his Türk Tarihi (Turkish History). 178 Ever since the foundation of the Republic of Turkey by Mustafa Kemal Atatürk in 1923, history gained particular importance for the newly created nation state. In his »Great Speech« Nutuk, delivered by Atatürk over six consecutive days in front of the Republican Party in Ankara in October 1927, he told the nation's story. In this speech, Atatürk stretched Turkish history from the dawn of civilisation to the present, glorified the pre-Islamic ancestors in Central Asia and offered a heroic image of a new, non-Muslim ancestor. Moreover, in his narrative, Turkish history is a linear succession of various Turkish states, such as the empire of the Huns or the Seljuqs.<sup>179</sup> Accordingly, Atatürk himself promoted a research group of ministers, parliamentarians, professors and teachers to collect all possible sources on the history of the Turks. The result was the *Türk Tarihinin* Ana Hatları (Main Lines of Turkish History) that was devoted almost entirely to ancient and medieval history and gave a major role to the ancestral Turks of Central Asia. 180 This conception finally culminated in the »Turkish History Thesis«, according to which the Turks from Central Asia migrated in several waves and initiated the progress of civilization in the rest of the world.<sup>181</sup> The citizens of the Turkish Republic were portrayed as the direct descendants of ethnic Turks from Central Asia. 182 The Turkish History Thesis could be found in Turkish schoolbooks until the late 1980s; 183 accordingly, one might assume that it is still present in the minds of large parts of the Turkish population. In 1972, the Central Asian origin of the Turks was enhanced by Islam in the framework of the »Turkish-Islamic Synthesis«. 184 This reappraisal of Islam, embedded in a nationalism that goes hand-in-hand with the ruling political parties and various incidents over the last few decades, paved the way to Neo-Ottomanism, the current state ideology. Therefore, it seems nearly impossible not to think about Turkish politics when questioning the Central Asian origin of the Turks.

<sup>177</sup> Foss, Kemal Atatürk, 827.

<sup>178</sup> Nur, Türk Tarihi, 304-305. This 14 volume *History* was reprinted in the Latin alphabet but unchanged otherwise in 1978-1981.

<sup>179</sup> Morin and Lee, Constitutive Discourse of Turkish Nationalism, 492, 498-500.

<sup>180</sup> Türk Tarihi Heyeti, Türk Tarihinin Ana Hatları, chapter 9, 401-546.

<sup>181</sup> Başak, Citizenship and Identity in Turkey, 69.

<sup>182</sup> Morin and Lee, Constitutive Discourse of Turkish Nationalism, 499. For the sake of completeness, though, it has to be mentioned here that there were simultaneous attempts to construct an explicit Anatolian identity by announcing the Sumerian and the Hittites the forefathers of the Turks; see Türk Tarihi Heyeti, *Türk Tarihinin Ana Hatları*, chapter 7; see also, i.e., Erimtan, Hittites, Ottomans and Turks; Foss, Kemal Atatürk; Lewis, *Turkish Language Reform.* 

<sup>183</sup> Kabapınar, Başlangıcından Günümüze Türk Tarih Tezi, 164-177.

<sup>184</sup> Eligür, Mobilization of Political Islam in Turkey, 96.

The proximity of research questions to trends in Turkish historiography and in Turkish politics, therefore, was the reason for the detailed analysis of funding sources and the composition of research teams. Although the contiguity between research questions and Turkish historiography seems striking at the first glance, funding and the composition of research teams do not prove any intended political agenda of the genetic studies at hand. As a matter of fact, though, results are often presented in a rather catchy and subjective manner in public media as well as in websites and forums operated by private individuals and associations.

Concerning the Central Asian origin, I would like to pose the question Rudi Paul Linder asked in 1982: »What was a nomadic tribe?« In early anthropological research, tribes were traditionally construed as a group or community sharing a common territory, speaking a common language or dialect, sharing a culture and religious tradition, united under a single political organization, and having a common economic pursuit. Additionally, the terms "tribe" and "ethnic group" were often used interchangeably. Geoff Emberling discussed several items that do not allow for the determination of a tribe: tribal names (self-assigned or not), language(s), political system(s), physical variation or race. As early as 1969, Fredrik Barth rejected the equation of race, culture and language that were entailed in early perceptions of ethnic groups. Instead he suggested that "ethnic groups are categories of ascription and identification by the actors themselves". This definition implies that ethnic identity was part of a dynamic social process and it introduced the possibility of change in actor's group membership. 187

The same has to be stated for tribes: nomadic tribes were of hybrid and dynamic natures; shared concerns – such as pastures, raids and the will to expand – played a much greater role in medieval tribal formation than kinship did; external pressure –be it from sedentary societies or other nomadic tribes – was the major factor in their formation. Tribal organisation provided a simple and efficient means to channel and escalate conflict by involving additional groups according to an essentially binary logic of inclusion or exclusion. Finally, the common descent of a tribe does not necessarily describe a biological fact, but rather a genealogical and thus cultural construction. These tribal genealogies may serve as an idiom or charter that nomads use to explain their history and politics. Tribal genealogies are rearranged and a particular chief's lineage is discovered to be the senior lineage. The utility of genealogies as political supports derives paradoxically from the inability of most steppe nomads to render a precise and complete account of their descent. Typically, tribal genealogy is quite clear for the fairly immediate antecedents, becomes hazy and often contradictory for a number of more distant generations, but finally emerges precise and unequivocal in describing the apical ancestor of the tribe and his sons.

<sup>185</sup> Emberling, Ethnicity in Complex Societies, 297-298; see there for further literature.

<sup>186</sup> Barth, Introduction, 10. For the context of ethnicity and genetic history see Renfrew, Roots of Ethnicity, 21-29.

<sup>187</sup> Emberling, Ethnicity in Complex Societies, 299.

<sup>188</sup> Linder, What was a Nomadic Tribe?, 698-699.

<sup>189</sup> Emberling, Ethnicity in Complex Societies, 302; Pohl, Archaeology of Identity, 14.

<sup>190</sup> Linder, What was a Nomadic Tribe?, 696-697.

Genealogies, however, provide the notion of a common ancestry. In the past, this common ancestry served predominantly to create kinship relations, in a mythic sense rather than biological, and to support social cohesion of group members. <sup>191</sup> The »forefather-group« – a descent group who shared common patrilineal ancestors up to the seventh generation before their own – is a standard feature in the kinship society model of Turkic »nomads«. The ethnographic evidence for this is rather slight. <sup>192</sup> For the Eurasian steppe in particular, David Sneath observed that the deployment of descent and genealogies serve as technologies of power and forms of government that administer political subjects. Thus descent groups are more likely to reflect contingent historical conditions and forms of government in the wider sense than the kinship structures supposed in earlier anthropological models. <sup>193</sup>

In the present, though, the notion of common ancestry suggests a collective memory of a former unity, of a time when a group was geographically united. Often, in this past, the group was autonomous or held political control. A frequent, but not universal, concomitant of this memory of past togetherness is the hope of a political reunification in the future.<sup>194</sup>

In the end, genetic research also reinforces this conclusion: Chaix *et al.* tested the hypothesis of common ancestry from the geneticists' perspective, examining genetically ethnographically assigned descent groups (lineages, clans and tribes) from Central Asia with the assumption that if patrilineally organised descent groups correspond to a genetic reality, there should be a correlation between Y-chromosome diversity and group affiliation. However, the genetic kinship coefficients of people of the same tribe (but from different clans and lineages) were all slightly negative. Their genetic kinship was not significantly higher than the mean kinship of the whole population, and, in two populations, even significantly lower. Thus, Chaix *et al.* concluded, tribes do not correspond to a real genetic entity; their claimed common ancestry is likely to be socially constructed. However, the genetic stripe is the stripe of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population and the population of the whole population of the whole population and the population of the whole population and the population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population of the whole population

Furthermore, in their comparative analysis of genetic diversity (mtDNA and NRY) in pastoral and farming societies in Central Asia, Chaix *et al.* detected that the »molecular signature of pastoral social organisation disappears over a few centuries only after conversion to an agricultural way of life.«<sup>197</sup> They concluded the discussion of their results with an essential question: »To which extent could social organisation have modified the action of natural selection or the intensity of drift during recent human evolution?«<sup>198</sup>

So, following Lindner – as well as Yardumian and Schurr  $^{199}$  – the question remains: »What was a Turk?«

<sup>191</sup> Weber, Economy and Society, 389. For the Islamic worlds see, i.e., Savant and de Felipe, *Genealogy and Knowledge*. 192 Sneath, *Headless State*, 226, n. 20.

<sup>193</sup> Sneath, *Headless State*, 202-203. For Eurasia see also Gingrich, Medieval Eurasian Communities by Comparison, 485-489. For the early Ottomans, see Lindner, *Nomads and Ottomans in Medieval Anatolia*, 32-36.

<sup>194</sup> Emberling, Ethnicity in Complex Societies, 303.

<sup>195</sup> Chaix et al., Genetic or Mythical Ancestry, 1113.

<sup>196</sup> Chaix et al., Genetic or Mythical Ancestry, 1114-1115.

<sup>197</sup> Chaix et al., From Social to Genetic Structures in Central Asia, 43.

<sup>198</sup> Chaix et al., From Social to Genetic Structures in Central Asia, 47.

<sup>199</sup> Yardumian and Schurr, Who Are the Anatolian Turks?

#### Conclusion

In the introduction of his 1992 lecture on »The Roots of Ethnicity: Archaeology, Genetics and the Origins of Europe« Colin Renfrew emphasized its underlying topic: identity. One level indeed, in this study the issue of identity was encountered on two different levels. One level concerned the sampling criteria, in the case of modern DNA, according to which the identity of donors was assigned as Turkish. In most of the cases, these sampling criteria consisted of geography or language or even both; this implies – intended or not – the assignment of ethnicity. The second level is related to the past identity of when Turks« that comprises of an even a more fuzzy set of social and political components.

Discussing the topic of the Central Asian origin of the Turks, which was a major research objective in the studies at question, I showed the complexity of applied sampling criteria by illustrating settlement and language policies that had a great impact on who is considered to be Turkish today. Moreover, I traced the topic of the Central Asian origin of the Turks which, at the latest with the foundation of the Turkish Republic in 1923, became a state-induced idiom. In the first instance, the Central Asian origin served to create a Turkish identity by establishing a collective memory with the help of a continuous narrative of Turkish states since the fifth century, thus legitimising territorial claims in Anatolia and excluding the non-Muslim population. I ended my discussion with the initial question what was a Turkish tribe« from a social-anthropological and historical view and illustrated the hybrid and dynamic nature of tribal formation that is least based on a common genetic ancestry. Ultimately, this conception has been confirmed by genetic research in Central Asia.

Thus, the past and the present identity of the »object of investigation«, i.e. »the Turks«, remains uncertain.

As I mentioned in my introduction, the results of this analysis are presented from an archaeologist and historian's perspective. History cannot resist being affected by the developments of genetic research as it opens new and important avenues for the study of human history, e.g. migration. Bearing in mind the ongoing and future development of aDNA studies, several of my arguments brought forward here will become obsolete. However, it seems evident to me that multidisciplinary research teams have already yielded with past and contemporary genetic methodology the most convincing and significant results. I do hope that future genetic studies will be arranged in similar ways. Furthermore, I hope that my discussion of the Turkish context has indicated the usefulness of interdisciplinary research in genetics, i.e. the involvement of the traditional disciplines in order to avoid some pitfalls in the initial research setting, to contextualise the genetic data and to gain a meaningful interpretation.

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