연 구 / [한국 시베리아연구] 2020년 제24권 2호
https://data.doi.or.kr/10.22892/ksc.2020.24.2.05

Origin of Korean People and DNA Tracking

Lee, Hong Kyu*

▌목 차▌

<국문요약>

유전자 추적으로 살펴본 한국인의 기원

2011년 발간된 “한국인의 기원”이라는 책에서 나는 주류 한국인들이 시베리아에서 내려왔으며, 고고학적, 유전학적 및 인류학적 관점에서 원시-알타이어를 사용했다고 주장했다. 이번 논문에서 나는 고대 DNA 분야에서 최근에 이루어진 새로운 연구결과들을 수용하여 위의 책에서 제

---

* Department of Internal Medicine, Eulji Hospital and Eulji University Medical College

www.kci.go.kr

시한 주요 결론들을 수정했다. 가장 중요한 것은 동아시아인 중에서 두 그룹의 새로운 조상(구성 요소)가 발견된 것이다. 일본 원주민인 조몽인이 초기에 남방을 통해 이동해온 그룹을 대표한다는 것이 중요하다. 나는 북동아시아 및 남동아시아인들이 가진 유전적 요소는 모두 아프리카에서 유래한 해부학적으로 현대적인 호모 사피엔스의 후손이며, 지난 빙하기 동안 고대 북유라시안이 바이칼 호수부근으로 이동하여 북동아시아인과 혼혈되며 진화했다는 추측을 유지했다. '몽골인의 한냉한 기후에 적응된 특징'을 설명하기 위해 이 해석이 필요하다. 마지막 빙하기를 거쳐 살아남은 이 사람들은 빙하기가 끝난 후, 동서남으로 퍼져나가고, 일부는 미대륙으로 이동하여 아메리카 인디안의 선조가 된다. 남으로 내려온 사람들은 중국 북부 황하지역과 만주 요하지역을 포함한 여러 지역에서 현지에 살고 있던 동북아시아인과 혼혈되며 신석기 시대를 열었다. 요하지역에서 발생한 홍산문화는 가장 초기의 문화로서 한국의 문화와 밀접한 관련성을 보인다. 최근에 발견된 유전적 연구는 Y 염색체 일배체 그룹 O2b와 O2b1가 이곳에서 진화하여 한반도와 일본 열도를 침범했음을 보여주고 있다. 나는 데이비드 라이크가 중국 인구 역사를 설명하기 위해 만든 용어 - 양자강 고스트 인구집단 및 황하 고스트 인구집단에 대비하여 홍산문화를 발전시킨 요하 고스트 인구집단이 있다고 제안합니다. 요하 고스트 인구집단의 유전적 특징이 결정되면 한국어와 중국어의 차이와 같은 유전적, 문화적 현상에 대한 많은 미해결 질문이 해결될 수 있다. 이러한 질문에 답을 주기 위하여 많은 다학제적 연구가 필요하며, 특히 홍산문화 유적지에서 발굴된 고인골 DNA의 분석이 꼭 필요하다.

<주제어>

한국인의 기원, 고인골 DNA, 홍산문화, 요하 고스트 인구집단, 황하 고스트 인구집단, 중국인

www.kci.go.kr

I. Introduction

1. “Out of Africa”

In a book, The Origins of Koreans, published by Woori History Research Foundation, in 2011[1], I reconstructed the history of Koreans mainly from genetic anthropological perspectives. This reconstruction was supported by other scientific advances made in nearby scientific fields, particularly historical linguistics and archaeology. I started from the “Out of Africa” theory of Alan Wilson[2], which posits that every people living in outside of Africa are descendants of a small group of people, anatomically modern human (AMH), living in an area in east Africa about 160,000 years ago. In 1986 Wilson and his colleagues at University of Berkley analysed mitochondrial DNA (mtDNA) of 199 people, who represented people living throught the world, and found variations of mtDNA are rather small among humans (in particular compared to chimpanzees) and their types could be interpreted as offsprings of a mtDNA, a small group of women living in Africa, 160,000 years ago, had. This woman is known as Mitochondrial Eve; mother of all humans. This study suggested a small group AMH migrated out of Africans about 60,000 years ago, thus ‘Out of Africa’ hypothesis. Archaeologic anthropologists call these people anatomically modern, as they are very similar to extant people and different from extinct Homos, such as Neanderthals.

Mitochondrion has its own DNA (about 16.5 kilo bases long) from its symbiotic origin; all eukaryotes (all animals belong to this class) were formed by merge of a precursor life form of mitochondrion with an another life form, which had its own DNA (nuclear DNA). Mitochondrion

---

1) Lee HK. *The Origin of Koreans*. Woori History Research Foundation, Seoul, Korea. 2011.

2) Cann RL, Stoneking M, Wilson AC. “Mitochondrial DNA and human evolution.” *Nature*. 325:31-6, 1987.

www.kci.go.kr

became vital servant, which generates energy to its master cell. In animals, which reproduce with sexual system, mitochondria are passed only from mother to daughters, while nuclear DNA is passed to all offsprings. We can track our maternal genetic heritage by analyzing mtDNA. Difference between mtDNAs of chimpanzees and modern human is approximately 1,462 base pairs, while difference is less than 150 base pairs between the humans, a fact that supports extant people are very homogenous[3]. 'Out of Africa' hypothesis is well established now.

Genes in nuclear DNA or somatic genes determine phenotypes; they are blue print of a person. Among the nuclear genome, presence or absence of Y chromosome determine gender of that person. As Y chromosome passes only father to son, we can trace the paternal line by tracking a genetic marker on the Y-chromosome, especially the short tandem repeats (Y-STRs). Y-STRs are quite variable between men. Results from the Y chromosome variations among the modern humans were consistent with the Out of Africa hypothesis[4]. These genetic markers are frequently employed in forensics, paternity testing and genealogical DNA analysis, as well as anthropologic research.

2. How do you know? – Human Genome study

In 2001 Human Genome Project, or sequencing of whole genome a person was completed. Then Human Genome Diversity Project (HGDP) followed, and then sequencing 1,000 Human Genome Project (http://www.internationalgenome.org/).

---

3) Røyrvik EC, Burgstaller JP, Johnston IG. “mtDNA diversity in human populations highlights the merit of haplotype matching in gene therapies.” *Mol Hum Reprod*. 22:809-817, 2016.

4) Kibisild T. “The study of human Y chromosome variation through ancient DNA.” *Hum Genet*. 136:529–546, 2017.

www.kci.go.kr

Advances in gene sequencing technologies and understanding the meaning of sequence variations (bioinformatics), both of which were closely associated with the advances made in the computer sciences. Results from these studies revealed tantalizing details of human genome and some startling evolutionary history of humans. Scientists had developed algorithms, which could pinpoint where a person came from. Now you can find commercial companies providing this kind of service in USA.

For the anthropologic research an ethnic group, for example the Korean, not a person, is target. So we have to analyze DNAs from large numbers of people (population based sequencing) and compare their sequence variations of a population against other ethnic groups.

1. How do you know? – Ancient DNA study

In the Origins of Koreans, I had acknowledged that anatomically modern human (AMH) interbred with Neanderthals soon after they migrated out of Africa and also pointed out that there is another now extinct cousin of human, Denisovan, and they interbred with human somewhere in Asia as well (as shown in Figure 2). I did not know, however, the technologies employed in these ancient DNA studies would revolutionize our understanding of past history. “Ancient DNA revolution” force me to revise many part of my book (not done as yet), but it did not contradict with the main conclusion of my book.

Before going further I will explain several terms used in the archaeological anthropology, summarized in Table 1. Both archaeology and anthropology study the history of human from the origin to the present. Archaeological anthropology specifically studies past humans and cultures through material remains by excavation and collecting artifacts, and then analyses and interprets data along with soils (geologically), where the artifacts were found. It reconstructs the cultural processes of human species

www.kci.go.kr

in large. Genetic anthropologists work mostly with genetic data (obtained by sequencing genes), but work with archaeologists. This kind of collaboration is famously shown in a book[5] written by Svante Pääbo, at Max Planck Institute for Evolutionary Anthropology, Germany. He used bone specimens excavated by an archaeologist, Anatoly Derevianko of Russian Academy of Sciences, Russia, and found Denisovan for the first time. Pääbo and his collaborators developed many new techniques and made a kind of Human Genome Project for ancient human remains (mostly bones). David Reich, a collaborator of Svante Pääbo, summarizes new sciences made by "ancient DNA revolution" and reconstructed human past in a book, "Who We Are and How We Got Here"[6]. This paper is heavily influenced by these books.

2. "We Got Here" through northern and southern routes

To explain genetic component of Korean, which belongs to northern Asian cluster[7][8], I envisaged the majority of Korean came to Korean peninsula through northern route, as displayed at American Museum of Human History, New York, USA (https://www.youtube.com/watch?v=PUwmA3Q0_OE). A recent study on the present-day Han Chinese, Japanese and Korean people confirmed that Koreans are very similar genetically to northern Chinese and Japanese[9]. So the question is 'how they become different?' My answer was

---

[5] Pääbo S. *Neanderthal Man: In Search of Lost Genomes*. Basic Books, New York, USA, 2014.

[6] Reich D. *Who We Are and How We Got Here*. Pantheon Books, New York, USA, 2018.

[7] Rasmussen M, Guo X, Wang Y, et al. "An Aboriginal Australian genome reveals separate human dispersals into Asia." *Science*. 334:94-8, 2011.

[8] Magalhães TR, Casey JP, Conroy J, et al. "HGDP and HapMap analysis by Ancestry Mapper reveals local and global population relationships." *PLoS One.* 7:e49438. doi: 10.1371/journal.pone.0049438. 2012.

[9] Wang Y, Dongsheng L, Chung Y, Xu S. "Genetic structure, divergence and admixture of Han Chinese, Japanese and Korean populations." *Hereditas* 155:19. DOI: 10.1186/s41065-018-0057-5, 2018.

www.kci.go.kr

that they were shaped by 1) initial population divergence (north and south), 2) geographical isolation (glacier and cold weather in Siberia), 3) sequence of gene flows and 4) possibly regional natural selection.

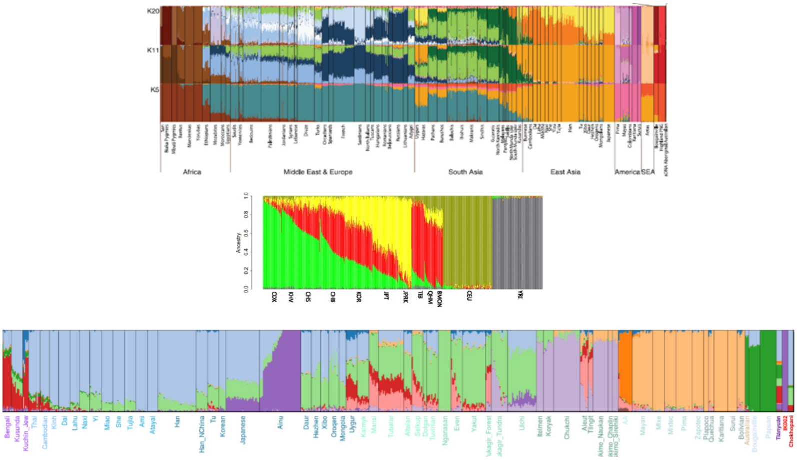

Figure 1. ***Upper panel***. Ancestry proportions of the studied 1,220 individuals from 79 populations and the ancient Aboriginal Australian with the ADMIXTURE program by Rasmussen et al.7), shown in K = 5, K = 11, and K = 20. A stacked column represents each individual, with fractions indicated on the y axis for the choice of K. Asians (shown in yellow color) appear homogenous in K11, but show two subgroups in K20, northern Asians (represented by Oroqens) and southern Asians (represented by Dai and Lahu). Japanese are closer to Oroqens, Han Chinese are closer to Dai. About half of Uygurs and Hazaras alleles are of northern Asians. ***Middle panel.*** Further analysis by Wang et al.9) revealed three dominant ancestral components in East Asian populations; Northern East Asian component (in red), Southern East Asian component (in green) and Ryukyuan component (in yellow). BMON; Buryat Mongolian, CDX; Chinese Dai in Xishuangbanna, CEU; Northern Europeans from Utah, USA, CHB; Chinese Han Beijing, CHS; Chinese Han South, JPRK; Japanese in Ryukyu, JPT; Japanese in Tokyo, KHV; Kinh in Ho Chi Minh City, Vietnam, KOR; Korean, QHM; Mongolians in Qinghai-Tibetan Plateau, TIB; Tibetan, YRI; Yoruba in Ibadan, Nigeria. Lower panel. Ancestry proportion of various Asian populations(K=10) reported by Gakuhari et al.[10], which shows also two ancient DNAs, a Jomon (IK002) and Tiányuán genome (far right). Jomon is an early migrant moved into Japanese archipelago, then isolated. Ainu (and Ryukyuan component in middle panel) and Japanese are direct descendent. Gakuhari et al. note genetic components of Jomon is found highly in Ami (Taiwan), Ulchi and people living coastal area. See also Figure 6. Tiányuán genome contains Papuan (green), Bengal (red), Jomon (purple) and South Asian (blue) genetic components.

---

10) Gakuhari T, Nakagome S, Rasmussen S et al. "Jomon genome sheds light on East Asian population history." https://www.biorxiv.org/content/biorxiv/early/2019/03/15/579177.full.pdf

www.kci.go.kr

3. Who We Are?

We know Koreans are similar to and different from other ethnic groups or people of other countries, particularly Chinese, Mongolians and Japanese. What we are interested in? This question is shown in Figure 2. I will use genetic anthropological approach to get answers, but then we have to agree first what “Korean” means. Before going into further, we have to appreciate what anthropologist Jonathan M. Marks stated:

“As any anthropologist knows, ethnic groups are categories of human invention, not given by nature”11). I also agree with statements made by the American Society of Human Genetics (ASHG)12) in that humans cannot be divided into biologically distinct subcategories, although there are clear observable correlations between variation in the human genome and individuals are identified by race. The study of human genetics challenges the traditional concept of different races of humans as biologically separate and distinct; most human genetic variation is distributed as a gradient, so distinct boundaries between population groups cannot be accurately assigned. There is considerable genetic overlap among members of different populations. Such patterns of genome variation are explained by patterns of migration and mixing of different populations throughout human history.

Koreans usually think they are all the descendants of Dangun (檀君), a son of Hwanwoong (桓雄), who is a son of Hwanin (桓因), a deity. Koreans think they have been living in Korean peninsula and Manchuria more than 5,000 years, when Gojoseon was established by Dangun. Japanese believe they are descendants of a deity, Amaterasu (天照), and Chinese believe similarly, for example, Yeowa (女媧), a deity. These myths, quasi-multiregional theories of the origins of individual ethnic groups, are

---

11) Marks J. *What it means to be 98% chimpanzee* Berkeley: University of California Press. pp. 202-7. USA. 2002.

12) ‘ASHG Denounces attempts to link genetics and racial supremacy.’ *Am J Hum Genet* 103:636, 2018.

www.kci.go.kr

simply wrong. But Koreans are apparently different from Chinese and Japanese; they differ in their cultures, particularly their languages. I will not discuss the details of cultural differences, but still will use "Korean" mainly in cultural terms, whatever that might mean.

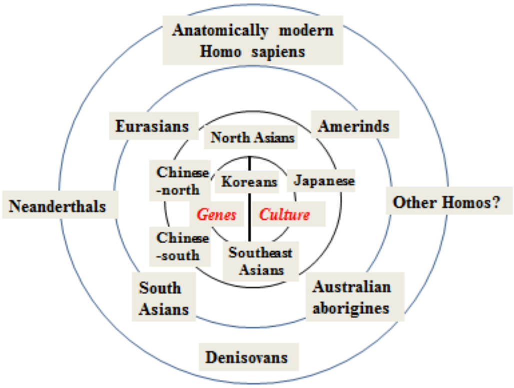

Figure 2. Koreans are relative of Japanese, Chinese and other Asians, who belong to the larger groups of people living in the world. Genetically all the extant people are descendants of anatomically modern human, with small genetic components derived from Neanderthals, Denisovans and other subspecies of Homo. We humans identify other people by classifying and naming them, mostly based on the physical appearance and culture.

Ⅱ. Evolution of humans-genetico-and paleo-anthropology

1. Anatomically modern humans (AMH) and terms used in paleoanthropology

he term AMH or anatomically modern Homo sapiens (AMHS) refers in paleoanthropology to individual members of the species Homo sapiens with an appearance consistent with the range of phenotypes in modern humans. When the "Out of Africa" hypothesis was proposed, anthropologists knew there were other kind of archaic human living in Europe in 200,000 years ago; Neanderthals. We now know that Neanderthals and Denisovans had

www.kci.go.kr

evolved from Homo heidelbergensis inside of Africa about 600,000 ~ 744,000 years ago and diverged each other about 200,000 years later[13]. “Anatomically modern” emphasize the fact that they did not show evidences for “behaviorally modern” characteristics as yet.

Archaeologists classify Old Stone Age of human of Africa into 3 stages; Early Stone (Lower Paleolithic) Age, Middle Stone (Middle Paleolithic or Mesolithic) Age and the Later Stone (Upper Paleolithic to Neolithic) Age. Then human culture progresses to bronze age and iron age. The Middle Stone Age (MSA) is a term applied to African prehistory in particular by archaeologists, which is generally considered to have begun around 280,000 years ago and ended around 50–25,000 years ago. Some researchers consider The beginnings of particular MSA stone tools have their origins as far back as 550–500,000 years ago. The MSA in Africa is different from the Middle Paleolithic of Europe, frequently misunderstood due to their roughly contemporaneous time span. However, an entirely different hominin population, Homo neanderthalensis, was the maker of the Middle Paleolithic of Europe. The Middle Stone Age in Africa is associated with both anatomically modern humans (Homo sapiens), as well as archaic Homo sapiens, sometimes referred to as Homo helmei.

“Behaviorally modern” is central characteristics of Homo sapiens. British museum at London made an Ice Age art exhibit under the title of “Arrival of Modern Mind” in 2013. Curators explained exhibits are the “masterpieces created by artists with modern minds like our own”. Emergence of “behaviorally modern” Homo sapiens (BMH) is linked to the transition from Middle to Upper Paleolithic Age. This subject is a hot topic in anthropology[14]. Hun-Jong Lee at Mokpo University, Mokpo, who is an

---

[13] Rogers AR, Bohlender RJ, Huff CD. “Early history of Neanderthals and Denisovans.” *Proc Natl Acad Sci U S A.* 114:9859-9863. doi: 10.1073/pnas.1706426114, 2017.

[14] Roebroeks W. “Time for the Middle to Upper Paleolthic transition in Europe.” *J Hum Evol.* 55:918-26, 2008.

www.kci.go.kr

expert in this subject, told me Russian archaeologists believe Siberia is the second birthplace of Homo sapiens. Professor Lee pointed out one of his colleague, Ted Goebel at University of Arizona, USA, showed the Upper Paleolithic cultures spread from the east Europe to west, citing archaeological observations made in Russia, particularly Kostenki 14 site (42,000 ~ 30,000 years old), near the Don river[15)][16)].

Archaeological studies to find the evidences which support the fact that behaviorally modern human (BMH) initiated the Initial Upper Palaeolithic Age are ongoing at the Denisova cave, where the bones belonging to Neanderthals and Denisovans were previously discovered[17)]. This site was repeatedly excavated since 1940 and many ancient artefacts typical of early H. sapiens were also found. However it was not clear when the modern human lived there. Recently Douka and her colleagues reported[18)] that two bone artefacts and assemblages excavated at the cave were of at the onset of the Initial Upper Paleolithic period, 42,660~48,100 and 41,590~45,700 years old, respectively, using a newly developed radiocarbon dating method. These results show Upper Paleolithic Age in Denisova cave predated Kostenki-14.

Dennell suggested that ornamental artifacts discovered with two bones, such as pendants made from bones and other decorative items made with mammoth ivory are made by modern humans. Because bones did not yield sufficient DNA for genetic analysis, authors appreciated the positive identification is yet to be done. In an accompanying news article, Dennel raised a possibility is that the Initial Upper Paleolithic at Denisova cave

---

15) Goebel T. “Anthropology; The missing years for modern humans.” *Science*. 315:194-196, 2007.

16) Anikovich MV, Sinitsyn AA, Hoffecker JF et al. “Early Upper Paleolithic in Eastern Europe and implications for the dispersal of modern humans.” *Science*. 315:223-6, 2007.

17) Dennell R. “Dating of hominin discoveries at Denisova.” *Nature*. 565:571-572, 2019.

18) Douka K, Slon V, Jacobs Z. “Age estimates for hominin fossils and the onset of the Upper Palaeolithic at Denisova Cave.” *Nature*., 565:640-644, 2019.

www.kci.go.kr

was made by hybrids of Denisovans and H. sapiens, provided the undeniable evidence that interbreeding between Neanderthals, Denisovans and H. sapiens had happened here. Homo sapiens had already reached north Siberia at least 46,880 to 43,200 years ago, as evidenced by the ancient DNA study on Ust-Ishim man.

Middle to Upper Paleolithic transition in Siberia implies that BMH might have newly emerged somewhere between the Kostenki-14 site and Denisova cave, around 42,000~48,000 years ago. This period belongs to an Ice Age. It is critically important for us to recognize that Upper Paleolithic culture did not appear in China, implying that BMH did not migrate into east Asia and the Neolithic Age start to appear around 15,000~13,000 years ago, coinciding the end of Ice Age. I interpreted this phenomenon suggest that (1) there was a natural barrier (glacier?) between Siberia and northern China- Manchuria region during the Ice Age and (2) after the disappearance of it by the end of Ice Age, BMH moved down to east Asia starting the Neolithic Age.

Table 1. Three age chronology of human genome- culture evolution

Stages	Culture and tools	Regions	Time frame (yrs BP)	Species name
Lower Paleolithic	Acheulian	Africa and other regions	2.5 to 0.2 million	Homo erectus, Homo heidelbergensis
Middle Paleolithic	Prepared-core technique	Africa		Homo helmei, H. sapiens
Middle Paleolithic	Mousterian	Europe	300,000-30,000	H. neanderthalensis
Upper Paleolithic	Aurignacian and other behaviorally modern cultures	Europe	50,000 to 10,000	H. neanderthalensis, H. Denisovan, H sapiens
Epipaleolithic (Mesolithic)	Microlithic tools	Eurasia	10,000 to 6,000	H. sapiens sapiens
Neolithic	Ceramics	North Asia	13,000 to 6,000	H. sapiens sapiens

www.kci.go.kr

2. Neanderthals

Neanderthal (specimen) was first found in the Neander Valley in the Germany in the year 1856. Thereafter numerous fossils as well as stone tool assemblages were found throughout Europe, which belonged to the so-called Mousterian culture. We now know this culture was of Neanderthals living within Europe from 400,000 until 40,000 years ago. Compared to modern humans, Neanderthals were heavy with shorter legs and bigger bodies, suggesting that they were adapted to cold climate. Recent evidences suggest they expanded to middle east and Altai mountain region, but not into Asia. This fact has an important implication in understanding the Korean.

Neanderthal genome revealed many interesting features of humans. Most importantly for us, all the extant people except those living sub-Sahara region carry about 1~2.5% of genes derived from Neanderthals, suggesting that they interbred AMH soon after AMH migrated out of Africa. (Northern Africans carrying Neanderthal genes are people migrated back to Africa). This interbreeding made AMH stronger, may be wiser for them to adapt colder environment, where Neanderthals were living for millennia. Archaeologists say that two Homos did not live well together mostly, resulting in extinction of Neanderthals.

3. Siberian Denisovans and Australo-Denisovans

In March 2010, Svante Pääbo and his colleagues reported that an undated finger bone fragment found in the Denisova cave in the Altai Mountains in Siberia, belonged to a finger bone of juvenile girl of yet unknown hominin[19]. Analysis of mtDNA showed it was quite different

---

[19] Krause J, Fu Q, Good JM, et al. "The complete mitochondrial DNA genome of an unknown hominin from southern Siberia." *Nature*. 464:894-7, 2010.

www.kci.go.kr

from modern humans and Neanderthals; it differed by 385 bases (nucleotides) out of about 16,500 bases of total mtDNA sequence of modern humans, whereas the difference between modern humans and Neanderthals was around 202 bases. The base difference between chimpanzees and modern humans is approximately 1,462 bases. This 385 bases difference suggested a divergence time around one million years ago. The nuclear genome of an another Neanderthal found nearby the Denisova cave had 17% of the Denisovan genome. From these differences they calculated that Denisovans and Neanderthals split from Homo sapiens around 600,000 up to 744,000 years ago and diverged from each other about 200,000 years ago. Recent discovery of a 116,000 years old Denisovan jawbone with two molar teeth at Baishiya Karst cave at Tibet shows that indeed they were living in Asia[20].

4. Extinct Homos, not characterized or ghost – Super-archaic human

Denisovan genome has about 3-6% genetic component which do not belong to Homo sapiens, or sub-Saharan African's genome. These genetic components were not present in Neanderthals, suggesting that they originated from an archaic hominin who interbred with ancestor of Denisovans. David Reich designated this particular population lived in sub-Saharan region, "super-archaic human".

---

[20] Chen F, Welker F, Shen CC, et al. "A late Middle Pleistocene Denisovan mandible from the Tibetan Plateau." *Nature*. doi: 10.1038/s41586-019-1139-x. 2019.

www.kci.go.kr

5. Neanderthal, Siberian- Denisovan and Australo-Denisovan genetic elements among worldwide populations

Genome of sub-Saharan Africans are people who stayed inside of Africa is free from Neanderthal or Denisovan genes. As described earlier, all other people outside of Africa have about 2~2.5% of Neanderthal elements in their genomes. While Europeans do not carry Denisovan genetic elements, Asians carry about 1% of Denisovan DNAs. Interestingly about 3~5% of the DNA of Melanesians and Aboriginal Australians and around 6% in Papua New Guineans carry Denisovan genes21). This finding suggested anatomically modern human (AMH) interbred with (Siberian-) Denisovan soon after they interbred with Neanderthals. Region around Papua New Guinea might be a very likely place Denisovans had lived 50-60,000 years ago. David Reich designates this group of people Australo-Denisovan and suggested interbreeding with AMH occurred somewhere in Southeast Asia. Whatever the reason might be, AMH-Neanderthal-(Siberian-) Denisovan hybrid did not enter Europe.

In 2018, Browning et al.22) developed a new method detecting genetic introgression to 5,639 whole-genome sequences of people from Eurasia and Oceania and found the populations from East and South Asia and Papua New Guineans have two components with differing similarity to the sequences of the Siberian Denisovan.

This result implied that Denisovan admixture into modern humans occurred at least two different instances, and those Denisovan populations had different degree of relatedness to the Siberian Denisovan. Discovery of

---

21) Qing P, Stoneking M. "Denisovan ancestry in East Eurasian and Native American populations." *Mol Biol Evol.* 32:2665-2674, 2015.

22) Browning SR, Browning BL, Zhou Y. et al. "Analysis of human sequence data reveals two pulses of archaic Denisovan admixture." *Cell.* 73:53-61, 2018.

www.kci.go.kr

a 116,000 years old Denisovan jawbone with two molar teeth at Baishiya Karst cave at Tibet by Chen et al.[23], as discussed above, suggest that there are possibilities that yet-unidentified archaic genetic components might be present in genomes of Asians.

How they are related to Koreans or other populations of East remains to be clarified. Interestingly the people living deep in Amazon jungle, Mixe, Surui and other tribes, carry alleles Papua New Guineans have, indicating that a ghost population who originated in Southeast Asia, obtained (Australo-) Denisovan alleles, migrated up to Beringia and entered American continent. Denisovan genetic elements is not frequently found among native Americans. Extensive studies on the origins of Native American had established they entered American continent about 15,000 years ago through Bering strait, which was above sea level at that time and was a large tundra grass land, called Beringia. These people were from two regions, one from Siberia (Mal'ta boy genome is representative) and one from south Asia. This migration will be discussed further below.

6. Summary

Figure 3 shows the summary of this section.

---

[23] Chen F, Welker F, Shen CC, et al. “A late Middle Pleistocene Denisovan mandible from the Tibetan Plateau.” *Nature*. doi: 10.1038/s41586-019-1139-x. 2019.

www.kci.go.kr

Figure 3. Evolution of anatomically modern human (AMH) and influence of other hominins. AMH evolved and migrated out of Africa, circa 60,000 years ago, interbred with Neanderthals and Denisovans, who are offspring of Homo heidelbergensis. They diverged each other about 400,000 years ago, as they have been living in mainly in Europe and in Asia respectively. Note that maximum sea level during the ice age was low (for example, Yellow Sea level was 150 meter below the current level), such that people could easily migrate into Japanese islands. (Modified based on a picture by John D. Croft in English Wikipedia.)

Ⅲ. Ancient DNA revolution

DNA analyses of the people who lived more than thousands years ago revealed many surprising facts of human past histories and enforce anthropologists to rethink the facts they thought established. We were educated that people migrated out to the world in a way similar to diffusion of gas or flow of water, or “demic diffusion”. From the geology of Asia, such as Himalaya mountains, Tian-Shan mountains, Taklimakan and Gobi deserts and glaciers, I imagined natural barriers diverted demic diffusion and made people take one of two possible routes; southern route along the seashore (as beachcomber), through India, Malay peninsula to southern China or northern route through Siberian steppe.

However, analyses of genomes from several ancient individuals from Western Eurasia and Siberia revealed that some of these individuals have relationships to present-day Europeans, while others did not contribute their genetic elements to the present-day Eurasian populations. For example, presence of a human (Ust-Ishim man) in west Siberia 40,000 years ago did not show any genetic affinity with current Eurasians.

1. Ghost populations

Ancient DNA studies revealed that people migrated into an area, adapted to that locality (natural selection) and expanded, forming a unique

www.kci.go.kr

population, which could be identified only by genetic patterns. David Reich designated this group of populations, “ghost population”. This term is useful to understand the genetic patterns of the people separated across the continent without any archaeologic evidences linking them. For an example, genome of a tribe living in Amazon river area of south America carry same genes of New Guinea. We could imagine that ancestors carrying these genes migrate from Southeast Asia to America and then down to Amazon river valley. Consider another example of Ust-Ishim man who did not make any impact on the people living at north Siberia. He is one of ghost population who did not leave descendants.

Human migration was not smooth and continuous (simple demic diffusion), but is more like the punctuated equilibrium described by Eldridge and Gould[24], which posits that most social systems (populations in our case) exist in an extended period of stasis, which may be punctuated by sudden shifts leading to radical change. In other words, over the period of human evolution, new populations emerge rapidly and expand, replacing all the populations, persist for long period of time and then disappear completely without traces, except that could be found in their descendant’s genome.

2. Mal’ta boy – representative of Ancient North European (ANE) Ancestor population

In 2014, Raghavan et al.[25] at University of Copenhagen, Denmark, published the genetic analysis of the bones of a Siberian boy, whose

---

[24] Eldredge N, Gould JS. “Punctuated equilibria: an alternative to phyletic gradualism.” In Schopf TJM ed., *Models in Paleobiology*, Freeman Cooper. San Francisco, USA, 1972, pp. 82-115

[25] Raghavan M, Skoglund P, Graf KE et al. “Upper Palaeolithic Siberian genome reveals dual ancestry of Native Americans.” *Nature*. 505:87-91, 2014.

www.kci.go.kr

remains were found near the village of Mal'ta close to Lake Baikal in Siberia. The remains (named Mal’ta boy) were excavated in 1920s in a grave adorned with flint tools, pendants, a bead necklace and a sprinkling of ochre, indicating it was an Upper Paleolithic site. His Y chromosome belonged haplogroup R, and mitochondrial DNA to a haplogroup U, which are found almost exclusively in people living in Europe and regions of Asia west of the Altai Mountains. His DNA showed close ties to those of today's west Europeans and Native Americans.

He was an ancestor of both populations; about a third of the ancestry of today's Native Americans can be linked to him and ancestor ANE population. His genome met all the requirements as the “ghost” genome of ANE population. In 2019, Sikora et al.[26] reported Ancient Paleolithic Siberians are descendants of Mal’ta boy. This subject will be discussed further below.

3. Ancient Europeans

In 2014-15, the ancient DNA community, especially David Reich’s laboratory, published results dealing the DNAs obtained from 230 ancient individuals excavated from the graves in Spain, Germany, the steppe of far eastern Europe and Anatolia (which is now Turkey). By comparing these ancient individuals to West Eurasian people living today, they could trace the movement of Ancient West European DNAs. Some key concepts of their findings is illustrated in Figure 4. There is a common ancestral population that arose soon after the separation of Ancient North Eurasian from east Asians and sub-Saharan Africans. They named this ghost population, the Basal Eurasians. They also identified four basic ghost populations who contributed to current European gene pools; Levantine

---

[26] Sikora M, Pitulko VV, Sousa VC et al. “The population history of northeastern Siberia since the Pleitocene” *Nature*. doi: 10.1038/s41586-019-1279-z, 2019.

www.kci.go.kr

farmers (which was mixed with Basal Eurasians), Iranian farmers, Eastern Eurasian hunter-gatherers and Western European hunter-gatherers. They in turn became ancestors of two ghost populations; Anatolian and early European farmers and Steppe pastoralists. Western European hunter-gatherers, Levantine farmers and Iranian farmers are ancestors of Anatolian and early European farmer, and Steppe pastoralists are descendants of Iranian farmers and Eastern European hunter-gatherers. Steppe pastoralists then moved into West Europe with corded ware culture and Indo-European languages, and replaced Western European hunter-gatherers about 5,000 years ago.

Figure 4. Ancient genome analyses revealed 5 basic ghost populations (shown in blue circles) (Adapted from a figure by David Reich[27]).

---

[27] Reich D. *Who We Are and How We Got Here*. Pantheon Books, New York, USA, 2018.

www.kci.go.kr

4. Ust-Ishim man

Ust'-Ishim man is 45,000-year-old early modern human, whose bone was found in western Siberia. Genomic analysis showed no direct descendants of Ust-Ishim man's specific lineages among modern populations[28]. When compared to other ancient DNAs, he is more closely related to Tiányuán man, found near Beijing, China, dating from 42,000 to 39,000 years ago and Mal'ta boy than any others. He could be the first wave of anatomically modern humans who migrated out of Africa and diverged into distinct populations of Eurasia. According to a 2017 study, Siberian and East Asian populations were found to share 38% of their ancestry to Ust'-Ishim man. Genome of Ust-Ishim man provided precise information of an oldest human genome, but did not provide any new information. He is an outlier of evolutionary process of extant people.

5. Ancient Siberians

As discussed above, Martin Sikora at the University of Copenhagen, Denmark, and his collaborators reported the late Pleistocene population history of northeastern Siberia analyzing the results of 34 ancient genomes dating to between 31,000 and 600 years ago[29]. They documented complex population dynamics during this period, including three major migration events: 1) an initial peopling by a previously unknown Paleolithic population of 'Ancient North Siberians'(represented by men at Yana Rhinoceros Horn site or Yana RHS), who are distantly related to early West Eurasian hunter-gatherers; 2) the arrival of East Asian-related peoples,

---

[28] Fu Q, Li H, Moorjani P, et al. "Genome sequence of a 45,000-year-old modern human from western Siberia." *Nature*. 514: 445-449, 2014.

[29] Wong EH, Khrunin A, Nichols L, et al. "Reconstructing genetic history of Siberian and Northeastern European populations." *Genome Research*. 27: 1-14, 2017.

www.kci.go.kr

which gave rise to 'Ancient Paleolithic-Siberians', who are closely related to people living in far-northeastern Siberia (such as the Koryaks), as well as Native Americans; and 3) migration of other East Asian-related peoples during Holocene, who they name 'Neo-Siberians', and from whom many contemporary Siberians are descended. Each of these population expansions largely replaced the earlier inhabitants, and ultimately generated the mosaic genetic make-up of contemporary peoples who inhabit a vast area across northern Eurasia and the Americas (Figure 5a).

These authors also report the episodes of gene flow and local population replacements in recent times found in the Lake Baikal region in southern Siberia; here, the genomes from Ust' Belaya and neighboring Neolithic and Bronze Age sites show a succession of three distinct genetic ancestries over an approximately 6,000-year period. The earliest individuals show predominantly East Asian ancestry (represented by individuals from Devil's Gate Cave[30]) followed by a resurgence of Ancient Paleo-Siberian ancestry (up to about 50% ancestry) in the early Bronze Age, as well as the influence of West Eurasian steppe ancestry (about 10% ancestry from individuals associated with the Afanasievo culture). More importantly to us, authors showed geographical locations that are climatically suitable for human occupation in Siberia between 48 and 12 ka across temporal and spatial dimensions using Paleo-climatic niche modeling (Figure 5b). Climate change was a major driver of human population history across northern Eurasia and two possible scenarios could explain the gene flow during the formation of the early Native American and Ancient Paleo-Siberian gene pools; 1) early ANS-related groups occupying southern Beringia during the Last Glacial Maximum (LGM), and subsequently admixing with East Asian-related peoples who expanded northwards towards the end of the LGM, and 2) a more-southwesterly location (Lake Baikal region) for the admixture, with a

---

[30] Siska V, Jones ER, Jeon S, et al. "Genome-wide data from two early Neolithic East Asian individuals dating to 7,700 years ago." *Sci Adv. 3*:e1601877. doi: 10.1126/sciadv.1601877, 2017.

www.kci.go.kr

northward expansion after the LGM, which is supported by archaeological evidence for a movement toward south during the LGM. Genetic isolation of ancestral Native Americans after about 23 ka suggest maintenance of a structured population during the LGM, implying that Ancient Paleo-Siberians and ancestral Native Americans occupied different refugia.

I interpreted the scenario 2 or admixture at a more-southwesterly location (Lake Baikal region) is more likely because this region is highly climatically suitable for humans (Fig 5b) and this scenario is consistent with results of Jeong et al.31), which will be discussed further in section 3.8.

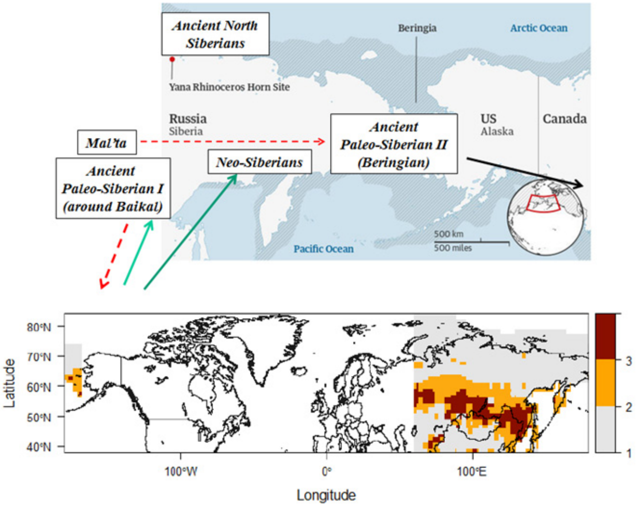

Figure 5. a. The population history of northeastern Siberia since the Pleitocene from the analysis of 34 ancient genomes analysis by Sikora et al32). b. Highly climatically suitable regions for human survival during the Last Glacial Maximum using Paleo-climatic niche modeling are shown in red; regions of low suitability are shown in grey; and regions with periods of both high and low suitability are shown in orange.

---

31) Jeong C, Wilkin S, Amgalantugs T et al. “Bronze Age population dynamics and the rise of dairy pastoralism on the eastern Eurasian steppe.” *Proc Natl Acad Sci. 115*:E11248-E11255, 2018.

32) Yang MA, Gao X, Theunert C, et al. “40,000-year-old individual from Asia provides insight into early population structure in Eurasia.” *Curr Biol. 27(20)*:3202-3208.e9. doi: 10.1016/j.cub.2017.09.030, 2017.

www.kci.go.kr

6. Tiányuán man

Tiányuán man is a 40,000-year-old individual from Tiányuán Cave, near Beijing, China. Tiányuán genome contained Papuan, Bengal, Jomon and South Asian (the ancient Hòabìnhians) genetic components, (Figure 1, lower panel) suggesting that he is more related to present-day and ancient Asians than he is to Europeans. However, he shared more alleles with a 35,000-year-old European individual (found at Kostenki site in Russia) than any other ancient Europeans, indicating that the separation between early Europeans and early Asians was not a single population split. About 1% of alleles were from Denisovans.

Deep analysis showed Tiányuán individual is not from a population that is directly ancestral to any group of present-day East or Southeast Asians. He was an outlier, like Ust-Ishim man (Figure 6). In other words, his genome suggests there was a ghost population that contributed to both present-day East and Southeast Asians, including Jomon. Interestingly Tiányuán individual shared alleles with some Native American groups in South America than with Native Americans elsewhere. In a computer model, Amazonians turned out to be a mixture of other Native Americans, the Tiányuán individual, and the Papua New Guineans33). He might be a representative of early migrant population through southern route, who went all the way to Beringia and beyond, ultimately to south America.

7. People at Devil's gate cave

In 2017 genome-wide data of two early Neolithic East Asian individuals from Devil's Gate, an early Neolithic cave site, was reported34). These

---

33) Siska V, Jones ER, Jeon S, et al. "Genome-wide data from two early Neolithic East Asian individuals dating to 7,700 years ago." *Sci Adv*. 3:e1601877. doi: 10.1126/sciadv.1601877, 2017.

www.kci.go.kr

individuals, dating to 7,700 years ago, are genetically most similar to geographically close modern Tungstic language speakers living around the Amur Basin, Ulchi tribe. The similarity to nearby modern populations and the low levels of additional genetic material in the Ulchis implied a high level of genetic continuity in this region[35]. It is interesting to see Jomon share many alleles with Ulchis and individuals of Devil’s Gate. Because Ulchis are one of Korean ancestors.

8. Khövsgöl burials in Mongolia – Ghost Population of Ancient Mongolian?

Genomic and proteomic data by Jeong et al.[36] of 22 directly dated Late Bronze Age burials, putatively associated with early pastoralism (confirmed by milk eating) in northern Mongolia (ca. 1380–975 BCE) showed the main cluster of Khövsgöl individuals are largely descended from a population represented by Early Bronze Age hunter-gatherers in the Baikal lake region. The genetic affinity between the main Khövsgöl clusters and world-wide populations (with Central African Mbuti as an outgroup), top signals were observed with earlier ancient populations from the Baikal region, such as the early Neolithic individuals from the Shamanka II cemetery, followed by present-day Siberian and northeast Asian populations from the Amur River basin, such as Ulchi’s, ancient persons at Devil's gate, and Neghidals and Nganasans from the Taimyr peninsula in deep

---

[34] Jeong C, Wilkin S, Amgalantugs T et al. “Bronze Age population dynamics and the rise of dairy pastoralism on the eastern Eurasian steppe.” *Proc Natl Acad Sci.* 115:E11248–E11255, 2018.

[35] Yang MA, Gao X, Theunert C, et al. “40,000-year-old individual from Asia provides insight into early population structure in Eurasia.” *Curr Biol.* 27(20):3202-3208.e9. doi: 10.1016/j.cub.2017.09.030, 2017.

[36] Gakuhari T, Nakagome S, Rasmussen S et al. “Jomon genome sheds light on East Asian population history.” https://www.biorxiv.org/content/biorxiv/early/2019/03/15/579177.full.pdf

www.kci.go.kr

north Siberia. Interestingly, Upper Paleolithic Siberians from nearby Afontova Gora (AG-2 subject, dated 17,000 years ago) and Mal'ta (24,000 years ago) had the highest extra genetic affinity with the main cluster of Khövsgöl individuals, when compared with other groups. AG-2 subject and Mal'ta boy had very similar genetic structures. Assuming that the early Neolithic populations of the Khövsgöl region resembled those of the nearby Baikal region, authors concluded that the Khövsgöl main cluster obtained ～11% of their ancestry from an Ancient North Europeans (shown in Figure 3) source during the Neolithic period and 4~7% from West Siberian herders ancestry (of Sintashta culture, descendants of Steppe Pastoralists) ancestry in the early Bronze Age. Authors also noted that Khövsgöls and other ancient Siberians share more ancestry with Native American-related gene pools than modern populations in the region do. These data suggest ~18% of Khövsgöl people at the time of beginning of Bronze age may be from Central European Steppe, and were Indo-European language speakers.

These features are depicted in Figure 6, which was developed by Gakuhuri et al.[37] to show evolutionary relationship of Jomon. The main cluster Khövsgöl individuals mostly belonged to Y chromosome Q1a (except one N1c1a) haplogroup, and mitochondrial A, B, C, D, and G, which are the major genetic types of Native Americans (except Y chromosome N and mtDNA G types). Dominance of Y chromosome haplogroup Q suggests west Siberian males became dominant, while females were mostly southern Asian descendants. I interpreted these data demonstrate that Indo-European speakers came to peri-Bikal region and Mongolia, in early Neolithic age. If the genetic markers of this Ghost Population of Ancient Mongolian could be found among Koreans, they will be difficult to identify, because this proto-Mongolian was formed by repeated mixture with people from southern Asian origin (probably more females) and west Siberian (probably more males), and then diluted further during their expansion.

---

[37] 방민규, "북극해 연안 소수 원주민의 치아인류학 특징." 『한국 시베리아연구』, 제22권 2호(배재대학교 한국-시베리아센터, 2018).

www.kci.go.kr

Recent studies by Bang[38] on the anthropological characteristics based on the dental data suggest an interesting way to explore further. He showed minority natives in the Arctic coast, including the Nenets, Komis, Eskimos, Aleuts, Yakuts, and Chukchis revealed that Aleuts and Eskimos had more similar teeth patterns of Koreans than others, while Carabelli's cusp, a characteristic index of Europeans, was highest among Komis, suggesting that dental data might be useful tool to study formation of Korean people.

9. Jomon, Ainu and Japanese

Ancient DNA analysis of a Jomon, IK002, by Gakuhari et al.[39] shows that this person clusters between present-day Southeast and East Asians and the Upper-Paleolithic human remain (40 kya) from Tiányuán Cave. IK002 also clustered with the Ainu, supporting previous findings that they are direct descendants of the Jomon people. The genetic component unique to Jomon and Ainu is also shared with present-day mainland Japanese as well as Ulchi (9.8% and 6.0%, respectively). They also note that IK002 can be modelled as a basal lineage to East Asians, Northeast Asia/East Siberians, and Native Americans, supporting a scenario in which their ancestors arrived through the southern route and migrated from Southeast Asia towards Northeast Asia. Analysis with the TreeMix program strongly supported that IK002 is the direct descendant of the people who brought the Upper Paleolithic stone tools 38,000 years ago into the Japanese archipelago. This interpretation is consistent with Browning et al.[40], who showed Chinese and Japanese have two identical Denisovan components.

---

38) 방민규, "북극해 연안 소수 원주민의 치아인류학 특징." 『한국 시베리아연구』, 제22권 2호(배재대학교 한국-시베리아센터, 2018).

39) Gakuhari T, Nakagome S, Rasmussen S et al. "Jomon genome sheds light on East Asian population history." https://www.biorxiv.org/content/biorxiv/early/2019/03/15/579177.full.pdf

40) Browning SR, Browning BL, Zhou Y. et al. "Analysis of human sequence data reveals two pulses of archaic Denisovan admixture." *Cell.* 2018,73:53-61

www.kci.go.kr

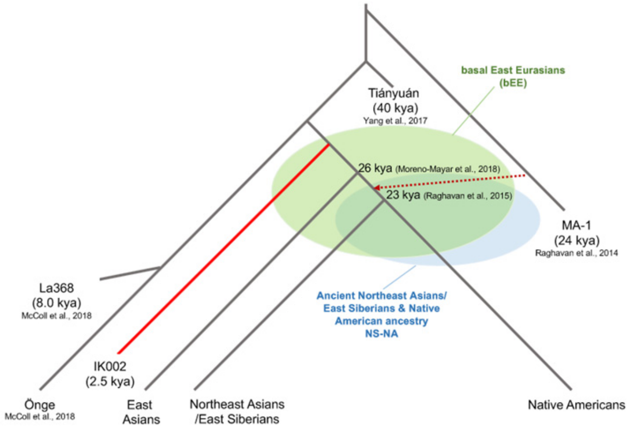

Figure 6. The evolutionary relationships of Asian populations analyzed with TreeMix program. Representative genetic datasets were made for different ancestries of East Eurasians and Native Americans; IK002 (Jomon), East Asians [Han, Ami, Japanese and Devils Cave, Northeast Siberians (Lokomotiv and Shamanka, the ancient Siberians), Native Americans (Clovis and USR1, the ancestry of Native American); ancient DNAs [Tiányuán, Mal'ta (MA-1) and Ust'Ism] of Upper Paleolithic period. Adopted from Gakuhari et al.41)

Ⅳ. After the Last Ice Age-understanding Chinese genetic patterns

From many archaeological evidences, we now know that people had reached east Asia soon after they migrated out of Africa, as exemplified by Tiányuán individual. Wang et al.42) showed the genome of Koreans, Chinese and Japanese are comprised by three dominant ancestral components; Northern East Asian, Southern East Asian, and Ryukyuan (=Jomon) components (Figure 1). My interpretation is that Mongolian

---

41) Gakuhari T, Nakagome S, Rasmussen S et al. "Jomon genome sheds light on East Asian population history." https://www.biorxiv.org/content/biorxiv/early/2019/03/15/579177.full.pdf

42) Wang Y, Dongsheng L, Chung Y, Xu S. "Genetic structure, divergence and admixture of Han Chinese, Japanese and Korean populations." *Hereditas* 155:19. DOI: 10.1186/s41065-018-0057-5, 2018.

www.kci.go.kr

genetic component (shown in red in the middle panel of Figure 1) is from Mongolian Ghost Population formed in early Neolithic Age at east Siberian steppe around the Baikal lake (Shamanka II and Lokomotiv in Fig. 6) and Mongolia, as discussed above. I suspect the Ryukyuan component shown in Figure 1 descended from ancient south Asians. Report of Takeuchi et al.[43] is consistent with this notion in that they showed Ryukyuan clusters have more components from the Southeast Asian and south Asians (both 4-6% vs. 0-1%) than the main Japanese component, whose major components of ancestry profile were from the Korean (87-94%), followed by Han Chinese (0-8%) distributed predominantly in northern China. As shown already above, results of Gakuhari et al.[44] were also consistent.

1. Two Ghost Populations inside of China, a conjecture of David Reich

David Reich hypothesizes two ghost populations in East Asia about 9,000 years ago (Figure 7); the farmers near the Yellow River in northern China growing millet and other crops; another in the south China near the Yangze River, growing crops including rice. He developed this hypothesis mainly from language patterns; languages of the mainland of East Asia comprise at least eleven major families; Sino-Tibetan, Tai-Kadai, Austronesian, Austroasiatic, Hmong-Mien, Japonic, Indo-European, Mongolic, Turkic, Tungustic, and Koreanic. Citing Diamond and Bellwood[45], he identified the first six groups correspond to expansions of East Asian farmers from the Yangze River disseminating their languages. Then Reich

---

[43] Takeuchi F, Katsuya T, Kimura R, et al. "The fine-scale genetic structure and evolution of the Japanese population." *PLoS One*. 12:e0185487. doi: 10.1371/journal.pone.018548, 2017.

[44] Marks J. *What it means to be 98% chimpanzee,* Berkeley: University of California Press. pp. 202-7. USA. 2002.

[45] Diamond J, Bellwood P. "Farmers and their languages: the first expansions." *Science*. 300:597-603, 2003.

www.kci.go.kr

support his hypothesis with genetic evidences that (1) many populations in Southeast Asia and Taiwan derive most of their genetic ancestry from a homogenous ancestral population, and (2) location of these populations strongly overlap where the rice farming expanded from the Yangze River valley. He thought this "Yangze River Ghost Population" contributed the overwhelming majority of ancestry to present day Southeast Asians. He pointed out, however, genetic patterns of Han Chinese is not consistent with descending directly from Yangze River Ghost population, but also have a large proportion of ancestry from another deeply divergent East Asian lineage found in northern Han. He provided no specific reference, but suggested ancient DNA research is ongoing in China.

Reich reasoned the presence of "Yellow River ghost Population" is necessary to accommodate Shang dynasty in Chinese history, as well as there should be a population who developed agriculture in the north while spreading Sino-Tibetan languages. They acknowledge, however, this conjecture is impossible to discern based on the genetic analysis of populations living today. Reich's conjecture is consistent with the genetic data. Xu et al.46) showed Han Chinese population is complicatedly sub-structured, with the main observed clusters roughly corresponding to N-Han (NHC), C-Han (CHC), and S-Han (SHC) with the greatest genetic differentiation between the NHC and the SHC. Citing previous studies based on analyses of archeological, anatomical, linguistic, and genetic data, Xu et al. suggested the presence of a significant boundary between the northern and southern populations in China, and the population differentiation could have resulted from isolation due to a geographical barrier such as the Yangze River.

---

46) Xu S, Yin X, Li S, et al. "Genomic dissection of population substructure of Han Chinese and its implication in association studies." *Am J Hum Genet*. 85:762-74, 2009.

www.kci.go.kr

Figure 7. Two Ghost Populations hypothesis of David Reich[47]. I added the Liao River Ghost Population to indicate the people associated with Hongshan culture. It is believed to have played critical role in the development of early Korean culture and Chinese civilization as well. Y chromosome haplogroup O2b and O2b1 might have arose here and expanded after they move into Korean peninsula and Japanese islands (see Figure 5). If this population spoke proto-Korean language, a branch of Altaic languages, Liao River Ghost Population could be descendant of Ancient Mongolian Ghost Population, as discussed in section of Khövsgöl burials.

2. Liao River Ghost Population

In 2009 Andrew Lawler, a contributing correspondent of journal Science, wrote an article entitled “Beyond the Yellow River: How China became China?”[48]. I think the title of the paper send clear message. He wrote “no one doubts that the plains around the middle Yellow river are where Chinese civilization coalesced around the middle of the 2nd millennium B.C.E., during what historians call the Shang dynasty”. However he also tell us “the origin of Chinese civilization is scattered all over the present-day country”, and “the very notion of [a single] Chinese civilization will probably have to be jettisoned”, and pointed out that “two cultures in particular - the Hongshan in the northeast, which flourished from 4,500~2,250 B.C.E., and the Liangzhu, which lasted from 3,500~2,250 B.C.E. - were setting the pace

---

[47] Reich D. *Who We Are and How We Got Here*. Pantheon Books, New York, USA, 2018.

[48] Lawler A. “Beyond the Yellow River: How China became China.” *Science*. 325: 930-935. 2009.

www.kci.go.kr

many centuries before the Shang", and finally the "excavations at Hongshan sites such as Niuheliang in the Liao river valley northeast of Beijing demonstrated 'a level of cultural sophistication that is not duplicated elsewhere at this time in early China' ". There are four key features relevant to this paper in the Lawler's article; 1) Neolithic culture began in China around 10,000 ago; 2) cultures in Hongshan and Liangzhu were among the first complex cultures in East Asia, possibly independent of other later cultures developed in Yellow river valley (*Xinglongwa culture, which precede culture, was 6,200~5,200 B.C.E.)*; 3) people developed "a level of cultural sophistication that is not duplicated elsewhere at this time in early China. They produced finely carved jade, like a phoenix and dragon – animals that later become central symbols in Chinese mythology; 4) the millet cultivation had started far north than Yellow river valley and it preceded rice cultivation in the south around Yangtze river, which began by 7,000 B.C.E. or earlier. I interpreted these four features are evidences for an arrival of new people who brought Neolithic culture into China about 10,000 years ago. And they point people came from the north. This subject is hot topic in the People's Republic of China and vast amount of literature was published on.

However crucial data are missing, particularly ancient DNA data, as China want to establish their own laboratory. I think it was inevitable, however, for "China Civilization Origin Search Process" (中华文明探源工程) of Chinese government to accept the early Liao river civilization as one of the roots of Chinese civilization. Therefore we need a third, the Liao River Ghost Population, to accommodate people who made culture. This population had contributed significant genetic input to Koreans and Japanese, which is the reason why we see Y chromosome haplogroup O2b and O2b1 among Koreans and Japanese49).

---

49) Wang Y, Dongsheng L, Chung Y, Xu S. "Genetic structure, divergence and admixture of Han Chinese, Japanese and Korean populations." *Hereditas* 155:19. DOI: 10.1186/s41065-018-0057-5, 2018.

www.kci.go.kr

Ⅴ. Languages and cultures

In this section, I will elaborate differences between the Koreans and Chinese people, especially their languages and letters. I will not discuss the difference (or identity) between the Korean and the Japanese languages, because I already provided evidences that Japanese are descendants of Liao River Ghost Population. I discussed this controversial issue in some detail in my book (1). I found contents of a recent book, 『Language Dispersal Beyond Farming』, edited by Robbeets and Savelyev[50] are supportive of common origin of Korean and Japanese languages. Particularly a chapter written by George van Driem, was quite supportive in that he argued that linguistic dispersals were, in most parts of the world, posterior to initial human colonization and that many linguistic dispersals were predominantly later male-biased intrusions, citing *Father Tongue hypothesis* of Poloni et al.[51]

1. Korean calligraphy, Hangul, and language

King Sejong of Yi dynasty knew the Korean language is different from the Chinese language and invented Korean Alphabet or Hangul. This fact every Korean knows. According to World Atlas of Langauge Structure (WALS), which is made linguistically maximally independent from each other[40], Korean language is within the Eurasian language macroarea, while China belongs to Southeastern language macroarea among the six macroareas defined (Figure 7). One of the major differences between

---

[50] Robbeets M and Savelyev A. *Language Dispersal Beyond Farming,* John Benjamins Publishing Company, 2017.

[51] Poloni ES, Ray N, Schneider S, Langaney A. “Languages and genes: Modes of transmission observed through the analysis of male-specific and female-specific genes.” In *Proceedings: Evolution of Language, 3rd International Conference 3–6 April 2000*, Jean-Louis Dessalles & Laleh Ghadakpour (eds), 185–186. Paris: École Nationale Supérieure des Télécommunications. 2000.

www.kci.go.kr

Korean and Chinese languages is of their use of tones. Thousands of languages in the world fall into one of two categories (notable exceptions being Japanese, some dialects of Scandinavia and northern Spain's Basque), tone or non-tone languages. While Korean is a non-tone language, Chinese use intonation in addition to using pitch for distinguishing words; for instance, the Chinese word "huar" said in a high pitch means 'flower(花)', but in a dipping pitch means 'picture (畵)'.

Tone languages are distributed in southeast Asia, as well as in Africa, suggesting that tone is kept in speaking language while people migrate via southern route after their migration out of Africa41). This pattern suggest early migrants might have used tone language. There are much more differences between Chinese and Korean languages other than their tonicities, such as agglutination of pronunciation, which will be discussed further below.

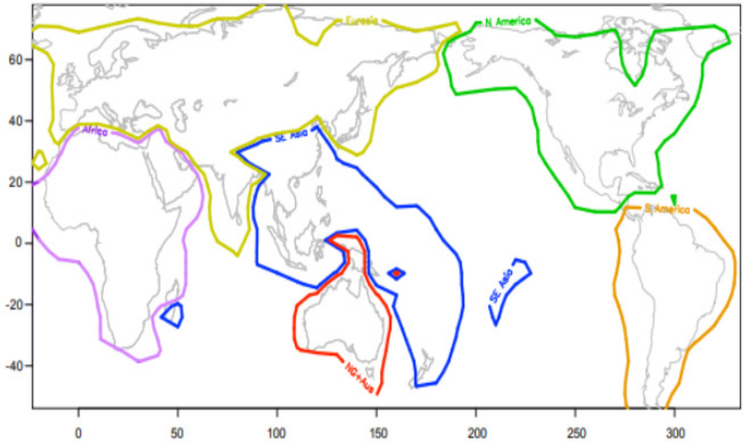

Figure 7. Geographic distribution of six macroareas pre-defined by World Atlas of Langauge Structure. Africa (purple); Eurasia (yellow); Southeast Asiaand Oceanic (blue); Australia and New Guinea (red); North America (green); South America (orange). Cited from Cysouw, Dediu and Moran52).

---

52) Cysouw M, Dediu D, Moran S. Comment on "Phonemic diversity supports a serial founder effect model of language expansion from Africa." *Science*. 335:657, 2012.

www.kci.go.kr

2. Language genes

There is an interesting report that use of tone in laguanges is tightly associated with the variations of two genes controlling brain function, ASPM and Microcephalin53). The mutations were absent in populations that speak tone languages, but abundant in nontone speaking populations. A significant correlation between the load of the derived alleles of ASPM (ASPM-D) and tone perception was observed in a group of European Americans (but not with Microcephalin in any measure of the brain function), suggesting that ASPM-D might have played an important role in language evolution by affecting brain function54). Didieu55) noted that the computer models suggest that when enough such individuals exist in a population for long enough time, language will indeed be influenced by their combined biases." It is interesting that ASPM-D variant evolved about 5,800 (~14,000) years ago56), but has since swept to become high frequency in the populations, suggesting this allele is under strong positive selection. Frequency was much higher among Europeans and Middle Easterners (highest among the Kalashi people of Pakistan) than Chinese (Yizu and Dai people have highest with 25%, but other tribes have less than 10%) or sub-Saharan Africans. ASPM-D frequency was unusually high among the Papua New Guineans, with a 59.4%. It is tempting to speculate that ASPM-D emerged somewhere near the Pakistan border and that

---

53) Dediu D, Ladd DR. "Linguistic tone is related to the population frequency of the adaptive haplogroups of two brain size genes, ASPM and Microcephalin." *Proc Natl Acad Sci U S A*. 104:10944-9, 2007

54) Wong PC, Chandrasekaran B, Zheng J. "The derived allele of ASPM is associated with lexical tone perception." *PLoS One*. 7:e34243. 10.1371/journal.pone.0034243, 2012

55) Didieu D. "Genes: Interactions with language on three levels - inter-individual variation, historical correlations and genetic biasing." in Binder PM and Smith K (eds.), *The Language Phenomenon*, The Frontiers Collection, DOI: 10.1007/978-3-642-36086-2_7, Springer-Verlag, Berlin Heidelberg. Germany. 2013.

56) Mekel-Bobrov N, Gilbert SL, Evans PD, et al. "Ongoing adaptive evolution of ASPM, a brain size determinant in Homo sapiens." *Science*, 309:1720–1722, 2005.

www.kci.go.kr

person (may be an Iranian farmer) became ancestor of Steppe pastoralist (as defined by David Reich, in Figure 4), a proto-Indo-European language speaking tribe. Dan Didieu of Max Planck Institute for Psycholinguistics, Nijmegen, opined that “people with the derived haplogroups of ASPM and Microcephalin might indeed turn out to be different from those without them in ways relevant to learning, processing or producing linguistic tone”[57].

3. Genomic history of India and spread of language – ‘father tongue hypothesis’ in action

Most Indian groups are mixed descendants of two genetically divergent populations: Ancestral North Indians (ANI), who are genetically related to Central Asians, Middle Easterners, Caucasians, and remotely Europeans. Ancestral South Indians (ASI) are not closely related to any groups outside the Indian subcontinent. Genome-wide study of the 73 Indian groups revealed all of them have West Eurasian influences, except an aboriginal Indians, i.e. Onge, living in Little Andaman island in Indian sea. These latter people are more closely related to present day East Asians, such as southern Chinese, separated more than 10,000 years ago. This evidence suggests how AMH migrated through southern route.

Dravidian language speakers of India have more Ancestral Southern Indian (ASI) ancestry, while Indo-European speakers have more Ancestral North Indian (ANI) ancestry. Genetic pattern changed continuously from north to south. After ANI and ASI had mixed, further population mixture within and between the ethnic groups has been very rare, even if they are very closely related, because of caste system and endogamy[58]. Around

---

[57] Mekel-Bobrov N, Gilbert SL, Evans PD, et al. “Ongoing adaptive evolution of ASPM, a brain size determinant in Homo sapiens.” *Science*, 309:1720–1722, 2005.

[58] Moorjani P, Thangaraj K, Patterson N, et al. “Genetic evidence for recent population mixture in India.” *Am J Hum Genet*. 93:422-38, 2013.

www.kci.go.kr

20~40% of Indian men and 30~50% of eastern European men have a R1a related Y chromosome types. Underhill et al.[59)] estimated that they descended from a male ancestor living in the vicinity of Iran and Eastern Turkey about 5,800 years ago (possibly one of Iranian farmers of Figure 4). In contrast, mtDNA types are almost entirely restricted to India, suggesting that they may have come all from ASI. These phylo-geographic data suggest the possibility that R1a lineages accompanied demic expansions of ANI into India initiated the Copper Age, slowly replacing previous Y-chromosome strata of ASI until Iron Age. Males with this Y chromosome types, speaking Indo-European language, were extraordinarily successful at leaving offspring while female immigrants made far less of genetic contribution. However Indo-European language somehow did not propagate beyond the Indian sub-continent via southern route of people's migration. Consistent with somatic genomic analysis, Y chromosome type analysis by Wang and Li[60)] (Figure 5) showed NO haplogroup appeared about 35,000-40,000 years ago in the east of the Aral sea and a branch of O (i.e. O2*) moved into South Asia via Indian subcontinent and another sub- branches (N and O) spread to North Asia. Koreans, Japanese and Manchurian have significant frequencies of Y chromosome haplogroup O2b and O2b1 (left panel)[61)], suggesting that they have originated inside of Manchuria.

[59)] Underhill PA, Poznik GD, Rootsi S et al. "The phylogenetic and geographic structure of Y-chromosome haplogroup R1a." *Eur J Hum Genet*. 23:124-31, 2015.

[60)] Wang C-C, Li H. "Inferring human history in East Asia from Y chromosomes." *Invest Genet*. 4:11. doi: 10.1186/2041-2223-4-11, 2013.

[61)] Kim SH, Kim KC, Shin DJ, et al. "High frequencies of Y-chromosome haplogroup O2b-SRY465 lineages in Korea: a genetic perspective on the peopling of Korea." *Investig Genet*. 4:10. doi: 10.1186/2041-2223-2-10, 2011.

www.kei.go.kr

4. Y chromosome types of Koreans emerged recently from the Hongshan culture

In 2015 Balaresque et al.[62] had reported the variations of Y chromosome of more than 5,000 men and found 11 descendant clusters among Eurasians. In this paper they noted O2b is unique Korea, which coincides with proto three kingdom period. They also showed Korean O2b have two centers; one inside of the Korean peninsula and another one at Liao river basin (Figure 8), where the Hongshan culture had flourished

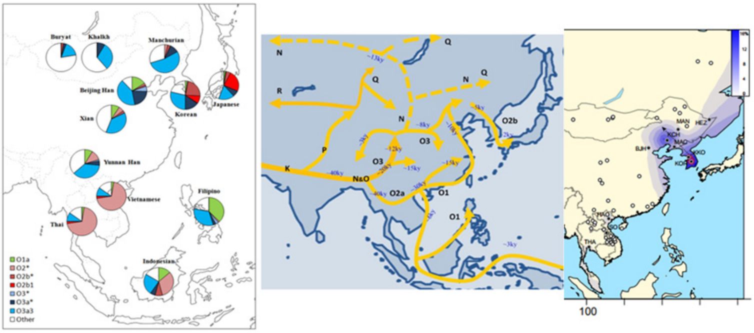

Figure 8. Koreans, Japanese and Manchurian have significant frequencies of Y chromosome haplogroup O2b and O2b1 (left panel), suggesting that they have originated inside of Manchuria[63][64]. They entered Korean peninsula about 5,000 years ago and moved to Japanese island about 1,500 years ago (middle panel). Frequency of O2b show two dense focuses[65], one in Liao river valley, another in south Korea, suggesting this haplogroup emerged from the Hongshan culture and entered Korean peninsula.

---

[62] Balaresque P, Poulet N, Cussat-Blanc S, et al. “Y-chromosome descent clusters and male differential reproductive success: young lineage expansions dominate Asian pastoral nomadic populations” *Eur J Hum Genet.* 23:1413-22, 2015.

[63] 권중혁. 『유라시아어의 기원과 한국어』 (in Korean). 퍼플출판, 2013.

[64] Pagel M, Quentin D. Atkinson QD, et al. “Ultraconserved words point to deep language ancestry across Eurasia.” *PNAS*. 110: 8471- 8476, 2013.

[65] Sagart L, Jacques G, Lai Y, et al. “Dated language phylogenies shed light on the ancestry of Sino-Tibetan”. *Proc Natl Acad Sci USA*. pii: 201817972.
doi: 10.1073/pnas.1817972116, 2019.

www.kci.go.kr

They report the analysis of the geographical patterns and ages of these descendant clusters (DCs) and their associated cultural characteristics show that the most successful lineages are found both among sedentary agriculturalists and pastoral nomads, and expanded between 2100 BCE and 1100 CE. However, those with recent origins in the historical period, including O2b, are almost exclusively found in Altaic-speaking pastoral nomadic populations, which may reflect a shift in political organization in pastoralist economies and a greater ease of transmission of Y-chromosomes through time and space facilitated by the use of horses. This observation reinforces an idea that males carrying Y chromosome haplogroup O2b, who entered Korean peninsula and Japanese archipelago were horse riding nomad speaking Altaic language.

Mr. Kwon JH Kwon JH, an independent researcher, published a book66), in which he demonstrated strong affinities between the Sanskrit and Korean. He found it during his long period of reading Buddhist bible written in Sanskrit. This proposition is consistent with a report from Santa Fe Institute and its collaborators, which showed Altai and Indo-European languages originated from a common language67).

5. Spread of Chinese languages

Recent reports provide additional evidences that the Sino-Tibetan languages arose in northern China about 5,900 to 7,200 years ago, associated with either the late Cishan and the early Yangshao cultures and millet cultivation, in contrast to the current view that Sino-Tibetan language arose from Yangtze river basin in association with rice cultivation68).

---

66) 권중혁.『유라시아어의 기원과 한국어』(in Korean). 퍼플출판, 2013.

67) Pagel M, Quentin D. Atkinson QD, et al. “Ultraconserved words point to deep language ancestry across Eurasia.” *PNAS.*110: 8471- 8476, 2013.

68) Sagart L, Jacques G, Lai Y, et al. “Dated language phylogenies shed light on the ancestry of Sino-Tibetan”. *Proc Natl Acad Sci USA*. pii: 201817972. doi: 10.1073/pnas.1817972116, 2019.

www.kci.go.kr

Directions of the spread are consistent with basic tenets of this paper in that Chinese culture arose by arrival of new people from the north. I regret that these studies did not include the languages spoken by northern Asia, including Korean, Japanese or Mongols.

Ⅵ. Summary

There are two dominant genetic components in East Asian populations; Northern East Asian and Southern East Asian (Figure 1). I conjectured the Northern East Asian or proto-Mongolian people evolved in peri-Baikal to Mongolia region, who provided basic Northern East Asian genetic component, and then they mixed with people who migrated up to this region taking southern route (Southern East Asian component). Being adapted to the cold environment of the Ice Age, the proto-Mongolian should have evolved many cold adapted features. I took the emergence of Neolithic culture in Manchurian plain and northern China after the end of Ice Age indicate their southern migration which was hindered by a natural barrier (probably glacier) between northern East Asia and Manchuria. Then they expanded in several regions where foods are rich; Liao river and Yellow river valley, where they established Hongshan culture and Yangshao culture, respectively. Y chromosome haplogroup O2b and O2b1 show some of them went to Korean peninsula and Japanese archipelago. I named this population the Liao River Ghost Population, to compare them with the Yangtze and the Yellow River Ghost Populations, a term David Reich used. Many unsolved questions on genetic and cultural phenomena, such as differences between Korean language and Chinese, could be solved if Liao River Ghost Population is characterized. Further studies are definitely needed to establish this conjecture, particularly analysis of DNAs from bones excavated in those regions. As this study is mostly based on the genetic studies, additional studies and analyses on Transbaikal region,

www.kci.go.kr

Siberia, as well as East Asia in large, will be need to fully understand formation of Koreans, as well as American populations. Son[69] claimed there is close affinity between Mexican language and Koreans, which might be interpreted as a result of their shared origins described in this paper. As recommended by Bang[70], further studies of ancient environment, the size of population, hereditary affinities, living strategies, and physical conditions including the cultural aspects of the entire area of Holocene Siberia in the middle age will be needed.

Ⅶ. Postscript

At the time when this paper was submitted, three major works on the ancient DNAs of Chinese, Siberians and North Asian were published[71][72][73] Several new observations made supported the conclusions described in this article.

---

[69] 손성태, “우리민족의 이동 흔적(2),” 『한국 시베리아연구』, 제20권 1호(배재대학교 한국-시베리아센터, 2016).

[70] 방민규, “고인골 자료로 본 시베리아 연바이칼 지역의 신석기시대 생계체계의 변화 양상에 대한 연구.” 『한국 시베리아연구』, 제24권 1호(배재대학교 한국-시베리아센터, 2020).

[71] Yang MA, Fan X, Sun B, et al. “Ancient DNA indicates human population shifts and admixture in northern and southern China.” *Science.* May 14;eaba0909. doi: 10.1126/science.aba0909, 2020.

[72] Yu Y, Spyrou MA, Karapetian M. et al. “Paleolithic to Bronze Age Siberians reveal connections with first Americans and across Eurasia.” May 17;S0092-8674(20)30502-X. doi: 10.1016/j.cell.2020.04.037, 2020.

[73] Ning C, Li T, Wang K, et al. “Ancient genomes from northern China suggest links between subsistence changes and human migration.” *Nat Commun.* 11:2700, 2020.

www.kci.go.kr

〈References〉

권중혁.『유라시아어의 기원과 한국어』(in Korean). 퍼플출판. 2013.

방민규, “고인골 자료로 본 시베리아 연바이칼 지역의 신석기시대 생계체계의 변화양상에 대한 연구.”『한국 시베리아연구』, 배재대학교 한국-시베리아센터, 제 24권, 1호, 2020.

방민규, “북극해 연안 소수 원주민의 치아인류학 특징.”『한국 시베리아연구』, 배재대학교 한국-시베리아센터, 제 22권, 2호, 2018.

손성태, “우리민족의 이동 흔적(2),”『한국 시베리아연구』, 배재대학교 한국-시베리아센터, 제 20권, 1호, 2016.

‘ASHG Denounces attempts to link genetics and racial supremacy.*’ Am J Hum Genet* 103:636, 2018.

Anikovich MV, Sinitsyn AA, Hoffecker JF et al. “Early Upper Paleolithic in Eastern Europe and implications for the dispersal of modern humans.” *Science*. 315:223-6, 2007.

Balaresque P, Poulet N, Cussat-Blanc S, et al. “Y-chromosome descent clusters and male differential reproductive success: young lineage expansions dominate Asian pastoral nomadic populations*” Eur J Hum Genet*. 23:1413-22, 2015.

Browning SR, Browning BL, Zhou Y. et al. “Analysis of human sequence data reveals two pulses of archaic Denisovan admixture.” *Cell*. 73:53-61, 2018.

Cann RL, Stoneking M, Wilson AC. “Mitochondrial DNA and human evolution.” *Nature*. 325:31-6, 1987.

Chen F, Welker F, Shen CC, et al. “A late Middle Pleistocene Denisovan mandible from the Tibetan Plateau.” *Nature*. doi: 10.1038/s41586-019-1139-x. 2019.

Cysouw M, Dediu D, Moran S. Comment on “Phonemic diversity supports a serial founder effect model of language expansion from Africa.” *Science*. 335:657, 2012.

Dediu D, Ladd DR. “Linguistic tone is related to the population frequency of the adaptive haplogroups of two brain size genes, ASPM and

www.kci.go.kr

Microcephalin.*”* *Proc Natl Acad Sci U S A*. 104:10944-9, 2007.

Dennell R. “Dating of hominin discoveries at Denisova.” *Nature*. 565:571-572, 2019.

Diamond J, Bellwood P. “Farmers and their languages: the first expansions.” *Science.* 300:597-603, 2003.

Didieu D. “Genes: Interactions with language on three levels - inter-individual variation, historical correlations and genetic biasing.” in Binder PM and Smith K (eds.), “『The Language Phenomenon』, The Frontiers Collection”, DOI: 10.1007/978-3-642-36086-2_7, Springer-Verlag, Berlin Heidelberg. Germany. 2013.

Douka K, Slon V, Jacobs Z. “Age estimates for hominin fossils and the onset of the Upper Palaeolithic at Denisova Cave.” *Nature*. 565:640–644, 2019.

Dryer MS. “Large Linguistic Areas and Language Sampling” *Studies in Language*. 13:257- 92, 1989.

Eldredge N, Gould JS. “Punctuated equilibria: an alternative to phyletic gradualism.” In Schopf TJM ed., *Models in Paleobiology*, Freeman Cooper. San Francisco, USA, 1972.

Fu Q, Li H, Moorjani P, et al. “Genome sequence of a 45,000-year-old modern human from western Siberia.*”* *Nature*. 514: 445–449, 2014.

Gakuhari T, Nakagome S, Rasmussen S et al. “Jomon genome sheds light on East Asian population history.”
https://www.biorxiv.org/content/biorxiv/early/2019/03/15/579177.full.pdf

Goebel T. “Anthropology; The missing years for modern humans.” *Science*. 315:194-196, 2007.

Jeong C, Wilkin S, Amgalantugs T et al. “Bronze Age population dynamics and the rise of dairy pastoralism on the eastern Eurasian steppe.” *Proc Natl Acad Sci*. 115:E11248–E11255, 2018.

Kibisild T. “The study of human Y chromosome variation through ancient DNA.” *Hum Genet*. 136:529–546, 2017.

Kim SH, Kim KC, Shin DJ, et al. “High frequencies of Y-chromosome

www.kci.go.kr

haplogroup O2b-SRY465 lineages in Korea: a genetic perspective on the peopling of Korea." *Investig Genet.* 4:10. doi: 10.1186/2041-2223-2-10, 2011.

Krause J, Fu Q, Good JM, et al. "The complete mitochondrial DNA genome of an unknown hominin from southern Siberia." *Nature.* 2010, 464:894-7

Lawler A. "Beyond the Yellow River: How China became China." *Science.* 325: 930-935. 2009.

Lee HK. *The Origin of Koreans*. Woori History Research Foundation, Seoul, Korea. 2011.

Magalhães TR, Casey JP, Conroy J, et al. "HGDP and HapMap analysis by Ancestry Mapper reveals local and global population relationships." *PLoS One*. 7:e49438. doi: 10.1371*/journal.pone*.0049438. 2012.

Marks J. *What it means to be 98% chimpanzee,* Berkeley: University of California Press. USA. 2002.

Mekel-Bobrov N, Gilbert SL, Evans PD, et al. "Ongoing adaptive evolution of ASPM, a brain size determinant in Homo sapiens." *Science*, 309:1720–1722, 2005.

Moorjani P, Thangaraj K, Patterson N, et al. "Genetic evidence for recent population mixture in India." *Am J Hum Genet*. 93:422-38, 2013.

Ning C, Li T, Wang K, et al. "Ancient genomes from northern China suggest links between subsistence changes and human migration." *Nat Commun*. 11:2700, 2020.

Pääbo S. *Neanderthal Man: In Search of Lost Genomes*. *Basic Books*, New York, USA, 2014.

Pagel M, Quentin D. Atkinson QD, et al. "Ultraconserved words point to deep language ancestry across Eurasia." *PNAS*.110: 8471- 8476, 2013.

Poloni ES, Ray N, Schneider S, Langaney A. "Languages and genes: Modes of transmission observed through the analysis of male-specific and female-specific genes." In *Proceedings: Evolution of Language, 3rd International Conference 3–6 April 2000*, Jean-Louis Dessalles & Laleh

www.kci.go.kr

Ghadakpour (eds), 185–186. Paris: École Nationale Supérieure des Télécommunications. 2000.

Qing P, Stoneking M. “Denisovan ancestry in East Eurasian and Native American populations.” *Mol Biol Evol.* 32:2665–2674, 2015.

Raghavan M, Skoglund P, Graf KE et al. “Upper Palaeolithic Siberian genome reveals dual ancestry of Native Americans.” *Nature.* 505:87-91, 2014.

Rasmussen M, Guo X, Wang Y, et al. “An Aboriginal Australian genome reveals separate human dispersals into Asia.” *Science.* 334:94-8, 2011.

Reich D. *Who We Are and How We Got Here,* Pantheon Books, New York, USA, 2018.

Robbeets M and Savelyev A. *Language Dispersal Beyond Farming*, John Benjamins Publishing Company, 2017.

Roebroeks W. “Time for the Middle to Upper Paleolthic transition in Europe.” *J Hum Evol.* 55:918-26, 2008.

Rogers AR, Bohlender RJ, Huff CD. “Early history of Neanderthals and Denisovans.” *Proc Natl Acad Sci U S A.* 114:9859-9863. doi: 10.1073/pnas.1706426114, 2017.

Røyrvik EC, Burgstaller JP, Johnston IG. “mtDNA diversity in human populations highlights the merit of haplotype matching in gene therapies.” *Mol Hum Reprod.* 22:809-817, 2016.

Sagart L, Jacques G, Lai Y, et al. “Dated language phylogenies shed light on the ancestry of Sino-Tibetan”. *Proc Natl Acad Sci USA.* pii: 201817972. doi: 10.1073/pnas.1817972116, 2019.

Sikora M, Pitulko VV, Sousa VC et al. “The population history of northeastern Siberia since the Pleitocene”, *Nature.*
doi: 10.1038/s41586-019-1279-z, 2019.

Siska V, Jones ER, Jeon S, et al. “Genome-wide data from two early Neolithic East Asian individuals dating to 7,700 years ago.” *Sci Adv.* 3:e1601877. doi: 10.1126/sciadv.1601877, 2017.

Takeuchi F, Katsuya T, Kimura R, et al. “The fine-scale genetic structure

www.kci.go.kr

and evolution of the Japanese population." *PLoS One*. 12:e0185487. doi: 10.1371/journal.pone.018548, 2017.

Underhill PA, Poznik GD, Rootsi S et al. "The phylogenetic and geographic structure of Y-chromosome haplogroup R1a." *Eur J Hum Genet*. 23:124-31, 2015.

Wang C-C, Li H. "Inferring human history in East Asia from Y chromosomes." *Invest Genet*. 4:11. doi: 10.1186/2041-2223-4-11, 2013.

Wang Y, Dongsheng L, Chung Y, Xu S. "Genetic structure, divergence and admixture of Han Chinese, Japanese and Korean populations." *Heredita*s 155:19. DOI: 10.1186/s41065-018-0057-5, 2018.

Wong EH, Khrunin A, Nichols L, et al. "Reconstructing genetic history of Siberian and Northeastern European populations." *Genome Research*. 27: 1−14, 2017.

Wong PC, Chandrasekaran B, Zheng J. "The derived allele of ASPM is associated with lexical tone perception." *PLoS One*. 7:e34243. 10.1371/journal.pone.0034243, 2012.

Xu S, Yin X, Li S, et al. "Genomic dissection of population substructure of Han Chinese and its implication in association studies." *Am J Hum Genet*. 85:762-74, 2009.

Yang MA, Fan X, Sun B, et al. "Ancient DNA indicates human population shifts and admixture in northern and southern China." *Science*. May 14;eaba0909. doi: 10.1126/science.aba0909, 2020.

Yang MA, Gao X, Theunert C, et al. "40,000-year-old individual from Asia provides insight into early population structure in Eurasia." *Curr Biol*. 27(20):3202-3208.e9. doi: 10.1016/j.cub.2017.09.030, 2017.

Yu Y, Spyrou MA, Karapetian M. et al. "Paleolithic to Bronze Age Siberians reveal connections with first Americans and across Eurasia." May 17;S0092-8674(20)30502-X. doi: 10.1016/j.cell.2020.04.037, 2020.

Zhang M, Yan S, Pan W, Jin L. "Phylogenetic evidence for Sino-Tibetan origin in northern China in the Late Neolithic." *Nature*. 569:112-115, 2019.

www.kci.go.kr

〈Abstract〉

Origin of Korean people and DNA tracking

Lee, Hong Kyu

In the book, “The Origins of Koreans” published in 2011, I had argued that mainstream Koreans came down from Siberia, and they spoke proto-Altaic language, from the archaeological and genetic anthropological perspectives. I revised its main conclusion in this paper to adopt new observations made in the ancient DNA field. There are two dominant ancestral components are found among East Asian populations; Northern East Asian and Southern East Asian components and they are all descendants of anatomically modern Homo sapiens originated in Africa. I maintained a conjecture that people evolved in peri-Baikal lake to Mongolia region during the last Ice Age provide basic Northern East Asian genetic component to explain ‘cold adapted feature of Mongolian’, and then they had mixed with people who migrated up to this area after taking southern route (Southern East Asian component). After the end of Ice Age, they expanded into the several regions, including the Liao river valley, where they established Hongshan culture. Y chromosome haplogroup O2b and O2b1 show a branch of them evolved there and invaded Korean peninsula and Japanese archipelago, where Jomon were living since 38,000 years ago. I propose to name the population who developed Hongshan culture, the Liao River Ghost Population, to compare them with the Yangtze River Ghost Population and the Yellow River Ghost Population, the terms David

www.kci.go.kr

Reich coined to explain Chinese population history. Many unsolved questions on genetic and cultural phenomena, such as differences between Korean language and Chinese, could be solved if Liao River Ghost Population is characterized genetically. Further studies are needed to justify this conjecture, particularly by analysis of ancient DNAs from bones excavated from sites of Hongshan culture.

〈Key Word〉

Origin of Koreans, ancient DNA, Hongshan culture, Liao River Ghost Population, Yellow River Ghost Population. Chinese.

저자 정보
이홍규(을지대학 을지병원, 석좌교수, 서울대학교 의과대학 명예교수)
hkleemd@eulji.ac.kr

논문 접수일: 2020. 5. 20
논문 심사일: 2020. 6. 5
게재 확정일: 2020. 6. 9

www.kci.go.kr