Genomic Journey of Polish Ancestors

Robert S. Sherins, M.D. Polish Genealogical Society of California January 2010

The advent of scientific investigations under the auspices of the Human Genomic Project made possible relatively inexpensive genetic testing, which was available to the public.¹ Several laboratories offered the services. The Family Tree DNA company was selected by the author to process his specimens, which were performed at the molecular biology laboratory under the direction of Professor, Dr. Michael Hammer at the University of Arizona. Dr. Hammer's laboratory has accumulated a genomic database of over 500,000 samples, which is currently the largest and foremost genomic testing laboratory for this purpose. The data are secured and indexed so that only the owner of the individual specimen can obtain his or her data by providing his/her sample kit number and a password. As new information is learned, the laboratory at no extra charge will provide updates to its participants. Additionally, Dr. Hammer's laboratory performs the DNA analyses on the specimens received from the National Geographic Genomic Project.

In 2001, "The Seven Daughters of Eve, the Science that Reveals our Genetic Ancestry," by Dr. Brian Sykes,² was published by W.W. Norton & Company, New York and London. This very readable and basic analysis of the female human genomic journey became a best seller, which created keen public interest in the use of DNA testing for the purpose of determining one's genetic ancestry.

Dr. Sykes determined that mutations in the genetic coding of our Y-DNA and mitochondrial DNA occur about once in every thousand years, although the mutations found in the male Y-DNA are more varied than the mitochondrial DNA (mtDNA) of females. The tests are performed and compared to the results in the database of samples taken from individuals living in other worldwide regions. Dr. Sykes identified and calculated the

¹ The "Human Genome Project" was organized and coordinated by the U.S. Department of Energy and the National Institutes of Health. The program was launched in 1990 in order to identify the approximately 20,000-25,000 genes in human DNA and to determine the sequences of the 3 billion chemical base pairs (nucleotides) that make up human DNA. By 2003, the results of the research investigations were released to the private sector.

² Oxford University Professor of Genetics at the Institute of Molecular Medicine.

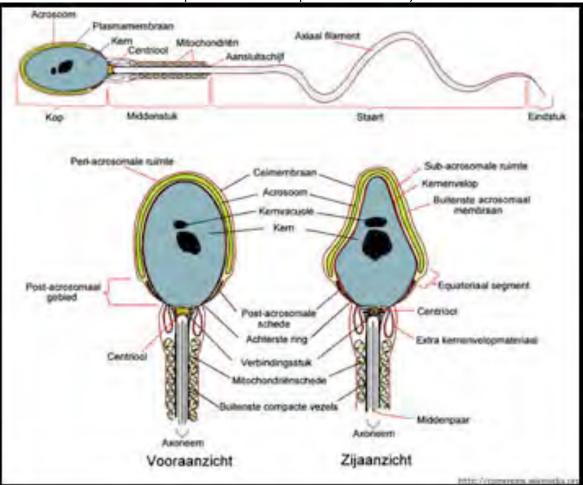
approximate dates of origin of the mutations. The results produced the estimated dates of the appearance of the most recent common female ancestors in Europe. Those females were called genetic "Eves". They were provided with fictitious given names, by which he identified the seven regional European genomic regions.

Mitochondrial DNA from those most recent genetic mothers was passed directly to every subsequent generation. By determining the chemical structures of the mitochondrial DNA, Dr. Sykes was able to establish the migration routes that were taken by the descendants of the "Eves" over the next tens of thousands of years. Each female Eve transferred her genes and her mutations to both female and male offspring. By sampling larger regional populations, a more accurate database was developed to predict the genomic identity of individuals, wherever they may have migrated in modern times.



Excerpted from Bryan Sykes, "The Seven Daughters of Eve"

Every male inherits both an X- and a Y-chromosome from his parents to produce the necessary final pairing of the XY-DNA that creates male gender. With maturation of the gametes (pre-sperm cells), the DNA of the sperm splits into two separate cells, one of which contains only the Ychromosome and the other that contains only the X-chromosome. Therefore, both the sperm (or ovum) contain only half of the original pair of chromosomes (called a haploid cell). In sexual union, both types of sperm compete to fertilize the ovum. With fertilization, however, the window of opportunity to gain access to the ovum is closed permanently. Thereafter, all other sperm are prevented from entering the same ovum.

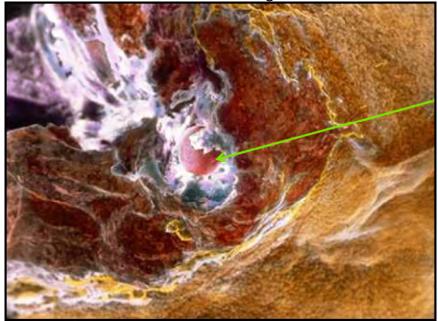


Graphic Chart of Sperm Anatomy

http://commons.wikimedia.org/wiki/File:Sperm_Dutch_text.png

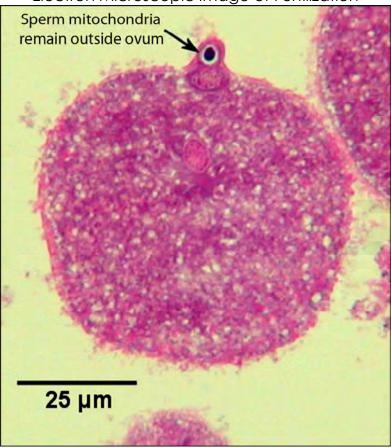
The mitochondrial DNA of the sperm is located in the neck of the cell, where the nucleus attaches to the tail. The neck of the sperm contains the mitochondrial DNA and does not enter the ovum in fertilization. Thus, when a father produces either a male or a female offspring, neither child can receive the mitochondrial DNA from him. It is the ovum that must produce both copies of the mtDNA in the "conceptus," whether or not that child was produced by a combination of the XY- or XX-DNA pairing. Therefore, mothers provide **both** copies of the mitochondrial DNA to **both** their sons and their daughters.

Ovulation Image



http://sprojects.mmip.mcgill.ca/gynecology/glostext.html

Electron Microscopic Image of Fertilization



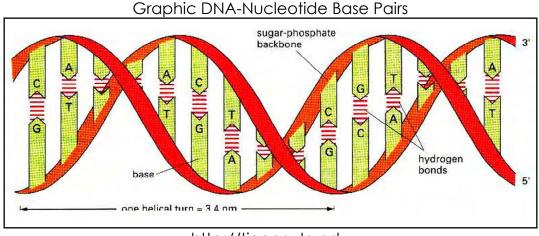
http://wiki.verkata.com/en/wiki/Spermatozoa

Submitting specimens for DNA testing:

This author submitted three tissue samples that were obtained by swabbing his mouth with sterile plastic-tipped probes that had been provided by the laboratory. The swabs were then mailed back to the laboratory in sealed pouches and analyzed. Tests were conducted for the Y-DNA that was inherited from Robert's Russian male ancestors, as well as for the mitochondrial-DNA material that he inherited from his mother's Romanian female ancestors.

In submitting a specimen, one can choose among several alternative tests. There is a simpler analysis of 12 or 25 targets at a lower cost, or more complex analyses of 37 or 67 targets. A laboratory report explained the results of Robert's Y-DNA analysis from 37 genetic molecular sites. The most complete testing of the entire genome is available at greater cost, but this was not necessary in the author's investigation. The entire process for ordering the tests and receipt of the results was provided on-line, which was simple and efficient. When the analysis was completed, a final certificate was sent by postal mail, which listed the molecular sites of the several mutations that were identified. If one doesn't have access to a computer, the tests may be ordered directly by telephone or by writing to the company.

Single mutations (SNPs) and repeated sequences of nucleotides were identified. The haplogroup type was identified by the constellation of all of the mutations, when compared to the same information that had been obtained from a varied group of samples taken from regional populations in the world. Knowing the DNA sequences from the global resources is essential in the process determining the genomic evolution from any individual sample. The substitution of a different nucleotide is called an **SNP**, specific nucleotide polymorphism. There are four types of nucleotide molecules, which combine as specific "base pairs": A-T and C-G (adenine with thymine or cytosine with Guanine).



http://tigger.ulc.edu

The author received e-mail notification when the results of the tests became available. A chart of those results is shown below, which appeared on the website, http://www.familytreedna.com. The Y-DNA samples submitted contained mutations that were positive for the "J" Haplogroup (shown below on the right side of the chart). With the more detailed analysis, Robert's Y-DNA subgroup was identified as "J2a."



Family Tree DNA – Haplogroup Results Webpage

In order to identify the genome of the author's <u>maternal</u> grandfather, an additional specimen was obtained from a male first cousin. It was submitted separately for Y-DNA analysis. The results are presented below as a chart of the mutations that were identified. Each mutation is called an "SNP" – single nucleotide polymorphism, which means that a single molecular site of a nucleotide had changed (mutation). Several mutations were identified. In this case, the results indicated the "E" haplogroup.

Table 2. Sample Y-DNA SNP Results

Your Y-DNA Belongs to Haplogroup E	
Your SNP results are: M96+ P2+ M2- M35+ M78- M183+ M81+ M107- M165- M1	23-

http://www.familytreedna.com

The results of the tests confirmed that the paternal and maternal grandfathers had different genomic identities. Their ancestral origins were not the same. In comparison, the family history and genealogical documents obtained from vital records proved that the two families had origins in Poland during the 18th century. The Polish ancestors of the paternal grandfather became "Russianized" as a result of the First Partition of Poland in 1772, when the border between Poland and Russia was simply moved westward as a result of the Russian invasion and occupation of Eastern Poland. The ancestors of the *maternal grandfather*, however, had origins in Galicia, Southern Poland, which became part of the Austro-Hungarian Empire after the Partitions. The earlier family journey to Poland prior to the discovery of any legal documents (1790's) remains a mystery. One can speculate about the possible earlier routes taken by the ancestors, in Poland, but only the DNA analyses confirmed a location of the most recent common genetic ancestors. The "E" haplogroup population appeared earlier out of Africa in the Arabian peninsula about 35,000 years ago, while the ancestors of the "J" haplogroup appeared in Central Asia about 25, 000 years ago. A sub-group (subclade) "J2a" appeared in the region of the Fertile Crescent about 13,000 years ago and further mutations have been identified (M41), which confirmed that the founder ancestor of M410 actually lived in Georgia or North Ossetia (Caucasus) about 8,000 years ago. This information confirmed that a great geographic distance had separated the ancient paternal and maternal family lines. The genomic journey shows great differences in the path to Poland. However, the ancient ancestors lived millennia before our Patriarch Abraham was born (Sanli Urfa, Turkey, 18,000 years BCE).



http://irrationalgeographic.files.wordpress.com

Since the author inherited the mitochondrial DNA from his mother, the specimen also was submitted for analysis of the mtDNA. As explained above, sperm cannot deliver any mitochondria to an ovum; the female must produce both copies of the mtDNA. Because male gender is determined by the XY-DNA combination, the specimen revealed the genomic identity of the author's mother and all of her ancient female genomic ancestors. By the same technical methods, the mtDNA of the author's sample was identified as "H" Haplogroup. According to Dr. Bryan

Sykes³, the letter "H" was assigned to the most recent common female ancestor, "Helena," who appeared in Southern France about 25,000 years ago. However, the mtDNA represented by the "H" haplogroup "Eve" had an ancestor, who originated much earlier and elsewhere in Western-Central Asia about 35,000 years ago. She was identified with a mitochondrial DNA designation of "R" haplogroup. The "R" ancestral "Eve" was the descendant from another female identified from the "N" haplogroup population, who had arrived in Central Asia about 80,000 years ago.

CRS – CAMBRIDGE REFERENCE SEQUENCE – mtDNA Results

mtDNA Results
Your Haplogroup and mutations relative to the Cambridge Reference Sequence (CRS) are shown below. A value of CRS indicates no mutations. High resolution (HVR2) results are shown only if you have requested the mtDNAPlus or mtDNA Refine test. If you ordered a Mega mtDNA the Coding Region (CR) will be displayed below.
As you go through your mtDNA results, we strongly encourage you to read the mtDNA Results Tutorial that we have put together in the form of frequently asked questions about mtDNA results.
Haplogroup - H HVR1 differences from CRS HVR2 differences from CRS 160803 16183C 16189C 16356C 1636 2633 309.1C 309.2C 16360T 315.1C

http://www.familytreedna.co

Human Genomic Journey – A Summary:

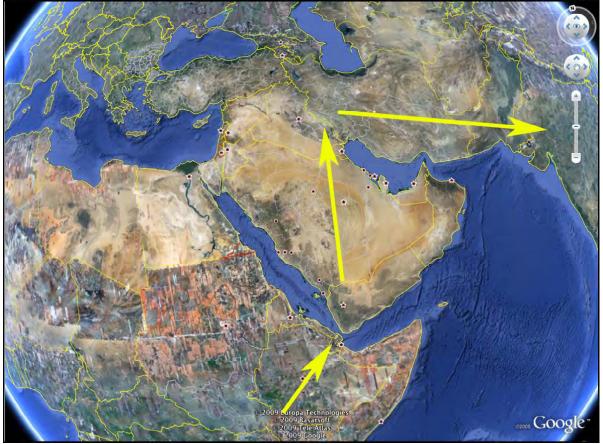
Based upon analysis of female mtDNA, the appearance of modern humans was estimated 150,000 years ago in the Rift Valley, which is located in the Kenya-Ethiopia region of Northeast Africa. Recent studies have identified a considerably older male genome that is calculated to be 348,000 years ago.

³ Sykes, Bryan: "The Seven Daughters of Eve," W.W. Norton & Company, New York and London, 2001.

Table of the Human Genomic Journey

Approximate Date	Region	Haplogroup
348,000 years ago	Oldest Human male Y-DNA	
150,000 years ago	Human Females: African Rift Valley	L1
100,000-90,000	Migrated within Africa	L1, L2, L3
80,000	Migration to Arabian Peninsula	L3
80,000	First founding mutation out of Africa	DE
65,000-55,000	Middle East and West Eurasia	Ν
60,000	East Eurasian	Μ
60,000	Southern Asia, Indonesia and Australia	DE
60,000 – 45,000	Large sub-set of the N Haplogroup; East and West Central Asia and the Middle East. R was derived from Haplogroup "N"	R
45,000	Sub-Set of "R" Haplogroup appeared	M89
40,000	Sub-set of M89; Mainly Middle East	J and J-1
35,000	Middle East and Levant	E
25,000	Southern Mediterranean and North Africa	E
13,000	Sub-Set of Haplogroup "J" - Located	J-2
	in the Fertile Crescent Region of Mesopotamia	M-172
8,000	Y-DNA haplogroup located in the Caucasus, Georgia and North Ossetia	J2a/M410

An aerial map view shows the principle migration routes of the earliest human migrations out of Africa.



"D" Haplogroup - First Out of Africa, 90,000 Years Ago

http://earth.google.com/

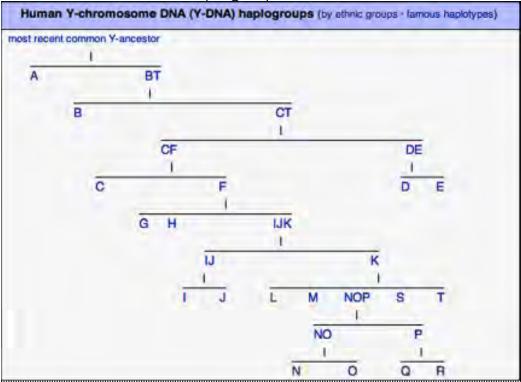


"N" Haplogroup in Central Asia, 80,000 Years Ago

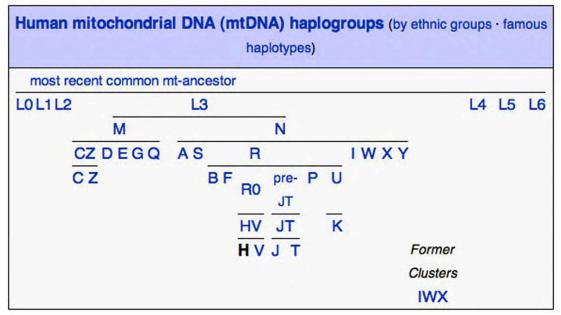
http://www.google.com/maps

Subsequent to an ice-age maximum about 45,000 years ago, sea levels dropped and grasslands emerged in Europe. Herds of animals migrated to the better pasturelands. Tracking their food sources, bands of modern humans migrated westward from Central Asia and entered Europe for the first time. These individuals have been named, CroMagnon. About 35,000 years ago, other human bands migrated eastward to Asia. Humans were still hunters and gathers and they followed the herds of animals into the newly accessible lands. After a later ice-age maximum, herds migrated to Siberia about 20,000 years ago. When a land bridge opened between Siberia and Alaska, and the height of the seas dropped, modern humans entered the North American continent for the first time – approximately 18,000 years ago.

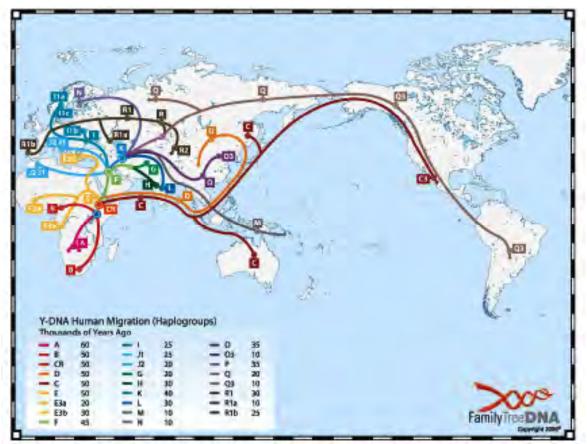
All living humans have evolved either from direct African ancestry or from the ancestors of the first migration out of Africa ("DE" haplogroup) and the second migration to Central Asia ("N" haplogroup). With the exception of the "D" and "E" haplogroups, most people living today are descendants of the humans who migrated to Central Asia about 45,000 years ago. The following two graphic tables demonstrate the evolution of of the male Y-DNA and female, mtDNA haplogroups. Two additional graphic maps are positioned below to demonstrate the wide distribution of both Y-DNA and mtDNA haplogroups in modern times.



Y-DNA Evolution http://en.wikipedia.org/wiki/Haplogroup



mtDNA Distribution/http://en.wikipedia.org/wiki/Haplogroup_H_(mtDNA)



http://www.familytreedna.com/

Survey of the Polish Genealogical Society of California Membership:

Polish people could be simply defined as those Slavic individuals who lived in the territory of Polonia since the 10th century. The senior group of Dukes elected their first king, Miesko I, from among their several tribes. Polonia was pagan until the German king announced that he would destroy Polonia unless it became Christian. The problem was resolved when Miesko I, married the Catholic Moravian princess Dobrawa in 965. Miesko was baptized in 966. Following this marriage, the entire country became Catholic.

Since the 10th - 13th centuries, the overwhelming majority of Poles are Roman Catholic, with a minority of Orthodox, Uniate and Protestant populations. A smaller group of Jewish inhabitants lived in Poland from the 13th century, having grown to about 2 million individuals by the 17th century. There are a few additional groups of other faiths in Poland. Poles generally identify themselves as Slavic people, but their diverse origins were previously unknown. With the advent of inexpensive DNA testing, the genomic origins of Poles can be determined. A survey of the membership of the Polish Genealogical Society of California was conducted. Thirtyseven members responded by sending the results of their DNA tests to fellow member Robert S. Sherins, M.D. The group divided into almost equal cohorts of males and females. Since Poles were forced to inhabit Prussia, Russia, and Galicia after the Partitions, the list of participants included several members with Kashubian, Galician, and Russian ethnic identities.

The following table was constructed from the results of DNA testing of PGSCSA members. Most members showed Central Asian origins, with the exception of the few male members, who demonstrated an "E" haplogroup identity.

DCSCA: DNA Haplogroup Distribution

HAPLOGROUP-SUB-CLADES	Origin	Y-DNA	mtDNA
outhwestern Arabian Peninsul:	35,000	E1b1b1	
Arabian Peninsula:	35,000	E1b1b1	-
outhwestern Arabian Peninsula:	35,000	E1b1b1	-
Southwestern Arabian Peninsula:	35,000	E1b1b1	-
5, Branch of F Caucasus	16,000	G	
outheastern Europe; Balkan:	15,000	I-2b1	
Fertile Crescent:	10,000	12	-
Fertile Crescent	10,000	J2a	
European:	12,000	K	-
East Asia:	20,000	N	
East Asia	18,000	N-LLY22	-
Siberia; Descendant of P:	20,000	0	
R1a1-Y-DNA: Central Asia	30,000	Rial	
Central Asia	30,000	8101	-
R1a1-Y-DNA: Central Asia	30,000	Rial	-
R1a1-Y-DNA: Central Asia	30,000	Rial	1
Central Asia:	25,000	R1b1b2	-
Western Europe:	25,000	Ribic	
Central & South Asia:	30,000	-	R1 - 010
Family from Northern Italy	30,000	1	н
Helena mtDNA:	33,000	1	н
Helena mtDNA:	33,000	-	H
Helena:	33,000	1	н
Origin Southwestern Europe; Distributed hroughout Europe:	35,000	1.	н
Drigin Southwestern Europe ; Distriuted Widely in Europe:	35,000	1.000	н
Distributed throughout Europe: years ago throughout Europe	35,000	1	H
Drigin Southwestern Europe ; Distriuted Widely in Europe:	35,000		HVR1
Western Asia; Descendant of JT:	45,000		3
ntDNA; 40% Ashkenasi Women:	4,000	1	Klaibia
Western Eurasia; Descendant of RO:	30,000	1	R-HV0
mtDNA-T: Central Asia/N. Iran about	8,000		T1
Western Asia; Descendant of "R":	55,000		U
Western Asia; Descridant of R to U; Spawned U1-U8	55,000	1	U4
Descendant of Haplogroup R:	45,000		W

Conclusion:

Since the first modern human coupling, human migrations have reached every continent on Earth and successfully adapted to the highly varied conditions from Polar regions to the hot tropical environments.

DNA is the chemical instruction code for building the structures of the cells of our bodies and the means by which all of our traits are inherited. The molecular structure of DNA can be accurately determined. There are 23pairs of human chromosomes, which determine our inheritance. One of those pairs specifically determines the gender of offspring. Those special chromosomes are called the sex-linked chromosomes and are identified by the letters "X" and "Y." An individual possessing the XY-DNA will become male; an individual inheriting the XX-DNA chromosome pair will become female. Because males can only receive their Y-DNA from the father, all living males can trace their genetic lineage back to the first human coupling. Similarly, females and males only inherit their mitochondrial DNA (mtDNA) from their mothers; so all living individuals can trace their maternal genetic evolution back to the first female of the coupling. By determining the locations and rate of changes in the molecular chemistry of the sex-linked chromosomes, an evolutionary pathway has been demonstrated for humans.

Modern humans first appeared in the Rift Valley of northeastern Africa about 348,000 years ago. For about the first 250,000 years, three identifiable mutations in the DNA appeared among the first groups of humans. Those small bands of individuals migrated, but remained within Africa. The people of the San-Bushman tribe are the modern descendants of the first humans. Previously the oldest female mtDNA was 150,000 to 200,000 years ago and the oldest male Y-DNA was about 60-90,000 years ago. Recent evidence of older DNA was obtained from an African-American man living in Georgia, whose Y-DNA mutations have been dated to 348,000 years ago. The mutations have been classified as Haplogroup A00. Two additional males have been identified in Chad and Algeria, who matching Y-DNA was calculated to 348,000 years ago. These remarkable findings push back our knowledge of the appearance of modern humans.

San Busman Village



http://nissan4x4.co.za

About 90,000 years ago, a band or bands of humans migrated out of Africa and crossed over to the Southern Arabian Peninsula, where the lands of the Horn of Africa and Arabia meet. The routes of migration out of Africa have been determined by the measuring molecular changes (mutations) of the gender-determining chromosomes, the Y-DNA and the mitochondrial DNA, (mtDNA).

This article has outlined the methods and results of published research that demonstrated the human journey since the first coupling in Africa about 348,000 years ago. The first group out of Africa has been identified as the "DE" haplogroup, which appeared in Arabia 90,000 years ago. Descendants of those first individuals continued to migrate along the coastline of South Asia, reaching India, Indonesia and Australia by 60,000 years ago. About 10% of every human on earth is a descendant of that group.

A second wave of humans left Africa later, about 80,000 years ago. They migrated northward into the Middle East. Some of those individuals remained in the Middle East, but others continued their journey northward reaching Central Asia about 80,000-60,000 years ago. About 90% of all humans on earth are descendants of this early wave of humans. By 45,000 years ago, an ice-age maximum occurred, which opened new grasslands to Europe. Early modern humans followed the herds of animals into Europe for the first time. By 30,000 to 25,000 years ago, other territories in Asia became accessible to wildlife. Modern humans migrated as far as Siberia. By 18,000 a Siberian land bridge to North America appeared and small bands of humans entered North America for the first time. When the seas rose again, those humans, who had migrated to North America, were unable to return to Siberia. Instead, they continued to move southward. Within a relatively short time, descendants from those early Americans reached Central and South America.

The earliest genetic ancestors of all living individuals have been identified. Every person on Earth is related to the first humans in Africa. Almost everyone is related to the earliest human inhabitants of Central Asia, with the exception of modern Africans, who are the descendants of the earliest bands of humans and the individuals who have inherited the DNA identified by both the "D" and "E" haplogroups. The history and demonstration of the routes of human migration have been discussed.