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Essay Review

Adam, Eve, and Other Ancestors: A Story of Human Origins
Told by Genes*

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L. Luca Cavalli-Sforza, Paolo Menozzi, and Alberto Piazza, *The History and Geography of Human Genes*, Princeton, New Jersey: Princeton University Press, 1994, pp. xi + 518 (+ 526 map pages), \$90.00.

'Scenery is fine – but human nature is finer'.¹

John Keats, 1818

'It has often and confidently been asserted, that man's origin can never be known: but ignorance more frequently begets confidence than does knowledge: it is those who know little, and not those who know much, who so positively assert that this or that problem will never be solved by science'.²

Charles Darwin, 1871

Mankind is a biological species: it is a part of nature. Like other living things, humans have evolved from non-human ancestors. If we are to understand human nature, what we are, we must know whence we come, the story of our humbler beginnings. For a century after the publication of Darwin's *On the Origin of Species* in 1859, the story of evolution was reconstructed with evidence from paleontology (the study of fossils), biogeography (the study of the geographical distribution of organisms), and from the comparative study of living organ-

* Parts of this essay are slightly modified from a review prepared for publication in the *Journal of Molecular Evolution*.

¹ Taken from a letter dated 13 March 1818 to Benjamin Bailey.

² Taken from Darwin's *The Descent of Man*.

isms: their morphology, development, physiology, and the like. The fusion of biochemistry and genetics in mid-twentieth century has yielded molecular biology, the most dependable source for reconstructing the ancestral relationships among living species.

An important recent contribution to the evolutionary literature is *The History and Geography of Human Genes* by L. Luca Cavalli-Sforza, Paolo Menozzi, and Alberto Piazza. This massive volume analyzes gene frequencies in indigenous living populations in order to reconstruct the history of mankind since the emergence 100,000 years ago of 'anatomically modern humans', that is, people with morphological traits essentially indistinguishable from living humans.

The hominid lineage diverged from the chimpanzee lineage 5-7 million years ago (Mya) and it evolved exclusively in the African continent until the emergence of *Homo erectus*, somewhat before 1.8 Mya. The first known hominid, *Ardipithecus ramidus*, lived 4.4 Mya, but it is not certain that it was bipedal or in the direct line of descent to modern humans. The recently described *Australopithecus anamensis*, dated 3.9-4.2 Mya, was bipedal and has been placed in the line of descent to *Australopithecus afarensis*, *Homo habilis*, *H. erectus*, and *H. sapiens*. Other hominids, not in the direct line of descent to modern humans, are *Australopithecus africanus*, *Paranthropus aethiopicus*, *P. boisei*, and *P. robustus*, who lived in Africa at various times between 3 and 1 Mya, a period when three or four hominid species lived contemporaneously in the African continent.

Shortly after its emergence in tropical or subtropical eastern Africa, *H. erectus* spread to other continents. Fossil remains of *H. erectus* are known from Africa, Indonesia (Java), China, the Middle East, and Europe. *H. erectus* fossils from Java have been dated 1.81 ± 0.04 and 1.66 ± 0.04 Mya, and from Georgia between 1.6 and 1.8 Mya. Anatomically distinctive *H. erectus* fossils have been found in Spain, deposited before 780,000 years ago, the oldest in southern Europe.

The transition from *H. erectus* to *H. sapiens* occurred around 400,000 years ago, but it is uncertain whether some fossils are *erectus* or 'archaic' forms of *sapiens*. *H. erectus* persisted for some time in Asia, until 250,000 years ago in China and perhaps until 100,000 years ago in Java. The subspecies *H. sapiens neanderthalensis* appeared in Europe around 200,000 years ago and persisted until thirty to forty thousand years ago. The Neanderthals were thought to be ancestral to anatomically modern humans, but now we know that modern humans appeared at least 100,000 years ago, much before the disappearance of the Neanderthals. Moreover, in caves in the Middle East, fossils of modern humans have been found dated 120,000-100,000 years ago, as

well as Neanderthals dated at 60,000 and 70,000 years ago, followed by modern humans dated at 40,000 years ago. It is unclear whether the two forms repeatedly replaced one another by migration from other regions, or whether they coexisted, or indeed whether interbreeding may have occurred.

There is considerable controversy about the origin of modern humans. Some anthropologists argue that the transition from *H. erectus* to archaic *H. sapiens* and later to anatomically modern humans occurred consonantly in various parts of the Old World. Proponents of this 'multiregional model' emphasize fossil evidence showing regional continuity in the transition from *H. erectus* to archaic and then modern *H. sapiens*. In order to account for the transition from one to another species, something which cannot happen independently in several places, they postulate that genetic exchange occurred from time to time between populations, so that the species evolved as a single gene pool, even though geographic differentiation occurred and persisted, just as geographically differentiated populations exist in other animal species. This explanation depends on the occurrence of persistent migrations and interbreeding between populations from different continents, of which no direct evidence exists. Moreover, it is difficult to reconcile the multiregional model with the contemporary existence of different species or forms in different regions, such as the persistence of *H. erectus* in China and Java for more than one hundred thousand years after the emergence of *H. sapiens*.

Other scientists argue instead that modern humans first arose in Africa or in the Middle East somewhat prior to 100,000 years ago and from there spread throughout the world, replacing elsewhere the pre-existing populations of *H. erectus* or archaic *H. sapiens*. Some proponents of this 'African replacement' model claim further that the transition from archaic to modern *H. sapiens* was associated with a very narrow bottleneck, consisting of only two or very few individuals who are the ancestors of all modern mankind. This claim is supported, erroneously as I will soon show, by the investigation of a peculiar small fraction of our genetic inheritance, the mitochondrial DNA (mtDNA).

The genetic information we inherit from our parents is encoded in the linear sequence of the DNA's four nucleotide components (represented by A, C, G, T) in the same fashion as semantic information is encoded in the sequence of letters of a written text. Most of the DNA is contained in the chromosomes inside the cell nucleus. The total amount of DNA in a human cell nucleus consists of six thousand million nucleotides, half in each set of 23 chromosomes inherited from

each parent. A relatively small amount of DNA, about 16,000 nucleotides, exists in the mitochondria, cell organelles outside the nucleus. The mtDNA is inherited in a peculiar manner, that is, exclusively along the maternal line. The inheritance of the mtDNA is a gender mirror image of the inheritance of the family name. Sons and daughters inherit their mtDNA from their mother but only the daughters transmit it to their progeny, just as sons and daughters receive the family name of the father, but only the sons transmit it to their children.

Analysis of the mtDNA from more than 100 ethnically diverse individuals has shown that the mtDNA sequences of modern humans coalesce to one ancestral sequence, the 'mitochondrial Eve' that existed in Africa about 200,000 years ago.³ This Eve, however, is not the one mother from whom all humans descend, but an mtDNA molecule (or the woman carrier of that molecule) from whom all modern mtDNA *molecules* descend.

Some scientists and many science writers have drawn the inference that all humans descend from only one, or very few women, but this is based on a confusion between gene genealogies and individual genealogies. Gene genealogies gradually coalesce towards a unique DNA ancestral sequence (in a similar fashion as living species, such as humans, chimpanzees, and gorillas, coalesce into one ancestral species). Individual genealogies, on the contrary, increase by a factor of two in each ancestral generation: an individual has two parents, four grandparents, and so on.⁴ Coalescence to one ancestral gene, originally present in one individual, does not disallow the contemporary existence of many other ancestors from whom we have inherited the other genes.

This conclusion can be illustrated with an analogy. My family name is shared by many people, who live in Spain, Mexico, the Philippines, and other countries. An historian of our family name has concluded that all Ayalas descend from Don Lope Sánchez de Ayala, grandson of Don Vela, vassal of King Alfonso VI, who established the domain ('señorío') de Ayala in the year 1085, in the now Spanish Basque province of Alava. Don Lope is the Adam from whom we all descend on the paternal line, but we also descend from many other men and women who lived in the eleventh century, as well as earlier and later.

The inference warranted by the mtDNA analysis is that the mitochondrial Eve is the ancestor of modern humans in the *maternal line*.

³ A.C. Wilson and R.L. Cann, 'The Recent African Genesis of Humans', *Scientific American*, April 1992, pp. 68-73.

⁴ The theoretical number of ancestors for any one individual becomes enormous after some tens of generations, but 'inbreeding' occurs: after some generations, ancestors appear more than once in the genealogy.

Any person has a single ancestor in the maternal line in any given generation. Thus, a person inherits the mtDNA from the mother, from the maternal grandmother, from the great grandmother on the maternal line, and so on. But the person also inherits other genes from other ancestors. The mtDNA that we have inherited from the mitochondrial Eve represents one-four-hundred-thousandth of the DNA present in any modern human. The rest of the DNA, 400,000 times more than the mtDNA, we have inherited from other contemporaries of the mitochondrial Eve.

From how many contemporaries? Before I answer this question I want to describe the genetic history of another peculiar component of our biological inheritance. This is the so-called Y chromosome, which is transmitted from father to sons, and thus exclusively in the paternal line, just as the mtDNA is inherited through the maternal line. A small DNA segment of the Y chromosome, part of the *ZFY* gene (involved in the maturation of the testes) has been analyzed in 38 men of diverse geographic origins, representative of major ethnic groups.⁵ Comparison with the *ZFY* gene from chimps and gorillas allows one to calculate the rate of evolution of this gene, which in turn leads to the conclusion that the last common ancestor of the *ZFY* gene of modern humans lived 270,000 years ago.

The carrier of the ancestral *ZFY* gene predicted by these calculations is the ancestor of all modern humans in the *paternal line*. As it was the case for the mitochondrial Eve, this 'ZFY Adam' is the individual from which all humans have inherited the *ZFY* gene, but not our only ancestor in his generation. We have inherited the other thousands of genes from many other contemporaries of this Adam.

The issue of how many human ancestors we had in the past has been elucidated by investigating the genes of the human immune system.⁶ The genes of the human leucocyte antigen (HLA) complex exist in multiple versions, which provide people with the diversity necessary to confront bacteria and other pathogens that invade the body. The evolutionary history of some of these genes shows that they coalesce into ancestral genes 30-60 Mya, that is, much before the divergence of humans and apes. (Indeed, humans and apes share many of these genes.) The genetical theory of gene coalescence makes it possible to estimate the number of ancestors that must have lived in any one generation in

⁵ R.L. Dorit, H. Akashi, and W. Gilbert, 'Absence of Polymorphism at the *ZFY* Locus on the Human Y Chromosome', *Science*, vol. 268, pp. 1183-1185, 1995.

⁶ F.J. Ayala, A. Escalante, C. O'Huigin, and J. Klein, 'Molecular Genetics of Speciation and Human Origins', *Proceedings of the National Academy of Sciences of the United States of America*, 91 (1994), 6787-6794.

order to account for the preservation of so many diverse genes through hundreds of thousands of generations. The estimated *effective* number is about 100,000 individuals per generation. This 'effective' number of individuals is an average rather than a constant number, but it is a peculiar kind of average (a 'harmonic mean'), compatible with much larger but not much smaller numbers of individuals in different generations. Thus, through millions of years our ancestors existed in populations that were 100,000 individuals strong, or larger.

The authors of *The History and Geography of Human Genes* are distinguished scientists who pioneered some years ago the methods they now apply and develop in large scale. The book is copiously documented and illustrated; with numerous tables, including two that occupy 9 and 75 pages; with 526 full-page maps collected in the back pages of the book; and with dozens more maps as well as many other illustrations scattered throughout the text.

The genetic information gathered from 971 published papers and monographs consists of the frequencies of more than 120 genes from 1,950 aboriginal populations widely representative of the world's ethnic diversity. For analysis at the continental level, the populations are reduced to 491 by combining some of them. For discerning worldwide patterns, the populations are further collapsed into 42. The authors use traditional genetic methods of analysis, such as the reconstruction of evolutionary (phylogenetic) trees, principal-component analysis, genetic distances, and so on. The results are presented at different levels of detail, from maps of individual genes in particular continents or regions, to synthetic maps that integrate all the genetic data.

The analysis manifests that there is greater genetic differentiation between African and non-African populations than between any other groups. This deep dichotomy indicates, according to the authors, that the origin of modern humans was in Africa, whence modern humans expanded to the rest of the world starting about 100,000 years ago. Nevertheless, they cautiously note that it 'is not yet possible, however, to exclude completely a partial participation of archaic *H. sapiens* from the Old World' in the origin of modern humans (p. 155).

If we accept the prevailing evidence that modern humans dispersed from Africa to the other continents, we need a hypothesis to account for the replacement of the pre-existing *H. erectus* and archaic *H. sapiens* populations by modern humans. Outright extinction of ancient human populations in the various continents previous to colonization by modern humans from Africa is an *ad hoc* possibility, but seems hardly compatible with evidence showing previous long-term persistence of *H. erectus* for well over a million years in different

continents. Moreover, the evidence of alternating Neanderthals and *H. sapiens* populations in Middle East caves, which I cited above, invalidates the generality of such presupposition. *The History and Geography of Human Genes* gives credit for the success of modern humans to greater skills in communicating by means of symbolic language. Increased ability to communicate would have strategically improved exploration and travel, and facilitated the development of technical improvements, which in turn would have led to success in colonizing other continents.

This is a sensible account of what might have been. But is there any empirical support for the hypothesis? It seems that there is such support, even if tenuous. It comes from the phylogenetic congruence between genetic trees and linguistic trees. Languages evolve very fast, which makes the reconstruction of language phylogenies prone to error. Not even the question whether human languages have one or multiple origins has been definitely settled, although a monogenetic origin is gaining favor. Direct 'fossil' evidence of languages comes from written records, but goes no deeper than 5,000 years. Nevertheless, major linguistic families have been identified and the authors show considerable congruence between sets of populations in the genetic tree and linguistic families. This congruence, although fuzzy at the margins, is particularly meaningful because it is unanticipated, since gene replacement and linguistic replacement can occur independently from each other, as indeed historical events have confirmed. For example, considerable gene admixture has occurred in the United States over the last three centuries with virtually complete adoption by all of the English language. It may very well be the case that, during the Neolithic and later, genetic and linguistic dispersal may have prevalently gone hand-in-hand.

The genetic trees of 42 worldwide populations consistently show, as noted above, a first split between African and non-African populations.⁷ This first split is statistically robust. The African/non-African split is also deep, so that the genetic distance between the first split and the next one (North Eurasians versus Southeast Asians + Oceanians) is about one-half of the average genetic distance between the most differentiated of the 36 non-African populations.

Branchings among non-African populations are statistically less robust than African versus non-African. Northeast and Arctic Asians cluster with each other and then with Amerinds, and their cluster

⁷ The Berbers of North Africa join the Caucasoids but this would be expected since North Africans are prevalently Caucasoid.

associates with Caucasoids. Pacific Islanders associate with Southeast Asians, and their cluster is remotely associated with Oceanians (New Guineans + Australians).

Modern humans are represented in the Middle East by fossils dated as early as 100,000 years ago, perhaps reflecting that the expansion of modern humans into tropical and subtropical east Africa and the neighboring Middle East region may have happened shortly after their emergence. The expansion into the Far East happened probably by two routes, one through Central Asia and one through South Asia into Southeast Asia and Australasia. The Far East was already colonized about 60,000 years ago. New Guinea and Australia were reached shortly afterwards, before 40,000 years ago. Polynesia was colonized by populations from Southeast Asia much later, within the last 3,000-3,500 years, although the process probably started around 6,000 years ago with the improvement of the nautical skills of South East Asians.

America was colonized by nomadic Siberian hunters during the last glaciation, which occurred 30,000-13,000 years ago, when the Bering strait had disappeared because the coast line was much lower than now or at earlier times (owing to retention of water in the polar ice cap). Some archaeologists place the earliest entry into North America around thirty thousand years ago; others date it around fifteen thousand years ago, after the glaciation peak that occurred at eighteen thousand years ago. The oldest archaeological sites are in Latin America, but their dates are controversial. Oldest claims are for some sites in Mexico at more than 30,000 years ago, the Piki-machay Caves in Peru are dated at 20,000 years, and in Brazil the Alice Boer Site is dated at 20-40 thousand years and the Pedra Furada at 32,000 years. None of the reliable sites (including Monteverde in Chile) are thought to be older than 14-16 thousand years by some archaeologists. In the United States, the oldest known sites are Wilson Butte Cave, in Idaho at 13-14.5 thousand years; Fort Rock Cave, in Oregon at 13,000 years; plus sites in Pennsylvania and Florida, dated 12,000 years. Numerous other known sites, dated at 11-11.5 thousand years, are characterized by arrowhead points of the Clovis culture. Sites in Alaska and Canada have been dated as early as 27,000 years ago, but more reliable dates do not go farther than 13.9-15.5 thousand years.

Archaeological and linguistic evidence indicates that at least three separate migrations occurred from Asia. First came the Paleo-Indians, ancestral to all indigenous populations in Mexico, and Central and South America. The Na Dene, named after the family of languages with that name, followed shortly afterwards and settled in Alaska and

the northwestern coast of North America until the beginning of the present millennium, when their descendants migrated farther south and east to parts of present Canada and the United States. Their southernmost representatives are the Apache and Navajo, who settled in the region that is now the US Southwest around A.D. 1200. The third migration about 10,000 years ago brought the Eskimo-Aleut, who settled in the Aleutian islands, Alaska, and the northern coast of North America.

The genetic data are not altogether consistent with the archaeological and linguistic picture. A genetic tree (Figure 6.9.1) deeply splits into two clusters, one fairly compact that joins the southern Na Dene with the Amerinds of Mexico, Central and South America. The other cluster is also deeply split, one branch joining the north Na Dene with Canadian, Alaskan and Greenland Eskimo; and the other branch including the Russian Eskimo and the Siberian Chukchi and Koryak. Cavalli-Sforza and coauthors conclude that the Apache-Navajo must have had considerable admixture with northern Amerinds. 'The attachment of Na-Dene to other Amerinds', they write, 'indicates that the component of the mixture due to the latter is, on the average, strong enough that it outweighs an original, unknown component responsible for the difference between Amerind and Na-Dene' (p. 324).

But other explanations are possible. Andrew Merriwether⁸ has investigated the mtDNA of 1300 native Americans from more than 40 populations, as well as ancient DNA from bone fragments and mummified tissue remains of 300 individuals from three burial sites in North, Central, and South America, dated between 5000 B.C. and 500 A.D. The different mtDNA types are broadly distributed among the native populations in a pattern, says Merriwether, that is much more consistent with a single wave (or series of waves) rather than with two distinct, Amerind and Na Dene, migrations. Other geneticists, but not all, interpret Merriwether's and other recent data similarly.

The earliest hominids from North America are anatomically modern humans. Not so in Europe, where *H. erectus* as well as Neanderthals and other archaic *H. sapiens* are known from multiple sites, preceding the presence of modern humans by as much as one million years. The European colonization of modern *H. sapiens* is however relatively recent, posterior even to the colonization of New Guinea and Australia, geographically much more remote from the African origins of modern humans.

⁸ See Nigel Williams, 'The Trials and Tribulations of Cracking the Prehistoric Code', *Science*, 269 (1995), 923-924.

Anatomically modern humans most likely colonized Europe in a population wave expanding westward from somewhere in southwest Asia, starting around 40,000 years ago and reaching western Europe by 30,000 years ago. From the last part of the middle Paleolithic until about 40,000 years ago, only Neanderthal *H. sapiens* lived in Europe, but they completely disappeared at that time. *The History and Geography of Human Genes* favors the view that the westward expansion of modern *H. sapiens* happened without any admixture with Neanderthals. Whether the expanding modern humans caused the extinction of the Neanderthals or, rather, it was the extinction of the latter that made possible the expansion of the former, cannot at present be settled for lack of evidence.

The genetic trees show Lapps and Sardinians as the two most differentiated European populations. Greeks and Yugoslavians, although much closer, are also outside of the main cluster that includes most European populations. Within this inclusive cluster of European populations, the Basques are the most distinctive group. The first two 'principal components' of genetic diversity show Sardinians, Lapps, and Basques as outliers. The distinctiveness of Lapps and Sardinians is due to admixture with populations of non-European lineage.

Basque is an enigmatic and the most distinctive of European languages. Four European spoken languages (Hungarian, Finnish, Lapp, and Estonian) belong to the Uralic family; all others but Basque are Indo-European, which may have originated in either present-day Turkey or the Ukraine. Basque cannot be associated with any existing family of world languages. It may have been, according to the authors, the language spoken in western Europe until the arrival of Neolithic colonizers, some ten thousand years ago, who brought the first Indo-European language into the area. Indeed, 'Basques are the only European people who can aspire to the privileged position of proto-Europeans' (p. 301).

I would like to add that the area now occupied by the Basques, in the western Pyrenees and in the adjacent Cantabric region of northern Spain, is also the geographic center of Paleolithic wall-cave art. Figurines of bone, ivory, or stone representing women, animals, and enigmatic creatures appear in Europe (and curiously in no other part of the World) around 30,000 years ago, during the early Upper Paleolithic, and last for about 20,000 years. Items of this 'miniature' art have been found in caves from eastern and central Europe, the Italian peninsula, and most abundantly in southern France and northern Spain. Painted walls occur in caves of southern France and northern Spain, in some 80 major and as many minor caves, scattered over an

area around the region now occupied by the Basques. Magnificent exemplars of artistic expression are Altamira in Spain and, in France, Lascaux and Vallon-Pont-d'Arc in the Ardèche, discovered in 1995.

Between 10,000 and 6,000 years ago, Europe was deeply transformed by the gradual expansion of agricultural techniques, crops, and domesticated animals, introduced by Neolithic farmers from the Middle East in waves that may have most often involved cultural expansion at the wave's front rather than extensive population migrations.

In 1863, four years after the publication of Darwin's *On the Origin of Species*, Thomas H. Huxley wrote: 'The question of questions for mankind – the problem which underlies all others, and is more deeply interesting than any other – is the ascertainment of the place which man occupies in nature and of his relations to the universe of things. Whence our race has come; what are the limits of our power over nature, and of nature's power over us; to what goal we are tending; are the problems which present themselves anew and with undiminished interest to every man born into the world'.⁹ Molecular biology and evolutionary studies shed light on the issue of human origins, Huxley's starting point for ascertaining the place of humans in nature and their relations to the universe of things. But, I hasten to add, the complete answer to the 'question of questions', of man's place in the universe of things, requires much more than biology.

⁹ T.H. Huxley, *Evidence As to Man's Place in Nature*, London: Williams and Norgate, 1863.