ANGELL*BIO 150* SPRING 2025
Recent Human Evolution

Questions to think about as you read….

1. Why do you think we used to imagine that humans would not experience natural selection?
2. Pick out and describe all the examples of recent evolution in humans (what is up with earwax-was it selected for?).
3. How did they detect the occurrence of selection?
4. If a trait was selected for a very long time ago, how would the regions around that gene differ from whether it was more recently selected for?
5. They point out that there are at least 25 genes that play a role in skin pigmentation. How accurate do you think this number is?
6. Describe the skin color example.
7. The authors note that ear wax differs between populations, but are not sure what survival advantage different kinds of earwax might confer. Instead they think the difference exists due to…..?
8. Africans show the highest levels of "neutral" or "background" variation while Europeans and East Asians seem to show more genes under recent selection. Why might both these patterns exist?
9. Hard vs. soft sweeps? What does this mean?

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Adventures in Very Recent Evolution
By NICHOLAS WADE

Ten thousand years ago, people in southern China began to cultivate rice and quickly made an all-too-tempting discovery — the cereal could be fermented into alcoholic liquors. Carousing and drunkenness must have started to pose a serious threat to survival because a variant gene that protects against alcohol became almost universal among southern Chinese and spread throughout the rest of China in the wake of rice cultivation.

The variant gene rapidly degrades alcohol to a chemical that is not intoxicating but makes people flush, leaving many people of Asian descent a legacy of turning red in the face when they drink alcohol.

The spread of the new gene, described in January by Bing Su of the Chinese Academy of Sciences, is just one instance of recent human evolution and in particular of a specific population's changing genetically in response to local conditions. Scientists from the Beijing Genomics Institute last month discovered another striking instance of human genetic change. Among Tibetans, they found, a set of genes evolved to cope with low oxygen levels as recently as 3,000 years ago. This, if confirmed, would be the most recent known instance of human evolution.

Many have assumed that humans ceased to evolve in the distant past, perhaps when people first learned to protect themselves against cold, famine and other harsh agents of natural selection. But in the last few years, biologists peering into the human genome sequences now available from around the world have found increasing evidence of natural selection at work in the last few thousand years, leading many to assume that human evolution is still in progress. "I don't think there is any reason to suppose that the rate has slowed down or decreased," says Mark Stoneking, a population geneticist at the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany.

So much natural selection has occurred in the recent past that geneticists have started to look for new ways in which evolution could occur very rapidly. Much of the new evidence for recent evolution has come from methods that allow the force of natural selection to be assessed across the whole human genome. This has been made possible by DNA data derived mostly from the Hap Map, a government project to help uncover the genetic roots of complex disease. The Hap Map contains samples from 11 populations around the world and consists of readings of the DNA at specific sites along the genome where variations are common.

One of the signatures of natural selection is that it disturbs the undergrowth of mutations that are always accumulating along the genome. As a favored version of a gene becomes more common in a population, genomes will look increasingly alike in and around the gene. Because variation is brushed away, the favored gene’s rise in popularity is called a sweep. Geneticists have developed several statistical methods for detecting sweeps, and hence of natural selection in action.

About 21 genome-wide scans for natural selection had been completed by last year, providing evidence that 4,243 genes — 23 percent of the human total — were under natural selection. This is a surprisingly high proportion, since the scans often miss various genes that are known for other reasons to be under selection. Also, the scans can see only recent episodes of selection — probably just those that occurred within the last 5,000 to 25,000 years or so. The reason is that after a favored version of a gene has swept through the population, mutations start building up in its DNA, eroding the uniformity that is evidence of a sweep.

Unfortunately, as Joshua M. Akey of the University of Washington in Seattle, pointed out last year in the journal Genome Research, most of the regions identified as under selection were found in only one scan and ignored by the 20 others. The lack of agreement is “sobering,” as Dr. Akey put it, not least because most of the scans are based on the same Hap Map data. From this drunken riot of claims, however, Dr. Akey believes that it is reasonable to assume that any region identified in two or more scans is probably under natural selection. By this criterion, 2,465 genes, or 13 percent, have been actively shaped by recent evolution. The genes are involved in many different biological processes, like diet, skin color and the sense of smell.

A new approach to identifying selected genes has been developed by Anna Di Rienzo at the University of Chicago. Instead of looking at the genome and seeing what turns up, Dr. Di Rienzo and colleagues have started with genes that would be likely to change as people adopted different environments, modes of subsistence and diets, and then checked to see if different populations have responded accordingly.

She found particularly strong signals of selection in populations that live in polar regions, in people who live by foraging, and in people whose diets are rich in roots and tubers. In Eskimo populations, there are signals of selection in genes that help people adapt to cold. Among primitive farming tribes, big eaters of tubers, which contain little folic acid, selection has shaped the genes involved in synthesizing folic acid in the body, Dr. Di Rienzo and colleagues reported in May in the Proceedings of the National Academy of Sciences.

The fewest signals of selection were seen among people who live in the humid tropics, the ecoregion where the ancestral human population evolved. “One could argue that we are adapted to that and that most signals are seen when people adapt to new environments,” Dr. Di Rienzo said in an interview. One of the most visible human adaptations is that of skin color. Primates have unpigmented skin beneath their fur. But when humans lost their fur, perhaps because they needed bare skin to sweat efficiently, they developed dark skin to protect against ultraviolet light.

Coloring the skin may sound simple, but nature requires at least 25 different genes to synthesize, package and distribute the melanin pigment that darkens the skin and hair. The system then had to be put into reverse when people penetrated the northern latitudes of Europe and Asia and acquired lighter skin, probably to admit more of the sunlight required to synthesize vitamin D.

Several of the 25 skin genes bear strong signatures of natural selection, but natural selection has taken different paths to lighten people's skin in Europe and in Asia. A special version of the golden gene, so called because it turns zebrafish a rich yellow color, is found in more than 98 percent of Europeans but is very rare in East Asians. In them, a variant version of a gene called DCT may contribute to light skin. Presumably, different mutations were available in each population for natural selection to work on. The fact that the two populations took independent paths toward developing lighter skin suggests that there was not much gene flow between them.

East Asians have several genetic variants that are rare or absent in Europeans and Africans. Their hair has a thicker shaft. A version of a gene called EDAR is a major determinant of thicker hair, which may have evolved as protection against cold, say a team of geneticists led by Ryosuke Kimura of Tokai University School of Medicine in Japan.

Most East Asians also have a special form of a gene known as ABCC11, which makes the cells of the ear produce dry earwax. Most Africans and Europeans, on the other hand, possess the ancestral form of the gene, which makes wet earwax. It is hard to see why dry earwax would confer a big survival advantage, so the Asian version of the gene may have been selected for some other property, like making people sweat less, says a team led by Koh-ichiro Yoshiura of Nagasaki University.

Most variation in the human genome is neutral, meaning that it arose not by natural selection but by processes like harmless mutations and the random shuffling of the genome between generations. The amount of this genetic diversity is highest in African populations. Diversity decreases steadily the further a population has migrated from the African homeland, since each group that moved onward carried away only some of the diversity of its parent population. This steady decline in diversity shows no discontinuity between one population and the next, and has offered no clear explanation as to why one population should differ much from another. But selected genes show a different pattern: Evidence from the new genome-wide tests for selection show that most selective pressures are focused on specific populations.

One aspect of this pattern is that there seem to be more genes under recent selection in East Asians and Europeans than in Africans, possibly because the people who left Africa were then forced to adapt to different environments. "It's a reasonable inference that non-Africans were becoming exposed to a wide variety of novel climates," says Dr. Stoneking of the Max Planck Institute.

The cases of natural selection that have been tracked so far take the form of substantial sweeps, with a new version of a gene being present in a large percentage of the population. These hard sweeps are often assumed to start from a novel mutation. But it can take a long time for the right mutation to occur, especially if there is a very small target, like the region of DNA that controls a gene. In the worst case, the waiting time would be 300,000 generations, according to a calculation by Jonathan Pritchard, a population geneticist at the University of Chicago. And indeed, there are not many hard sweeps in the human genome.

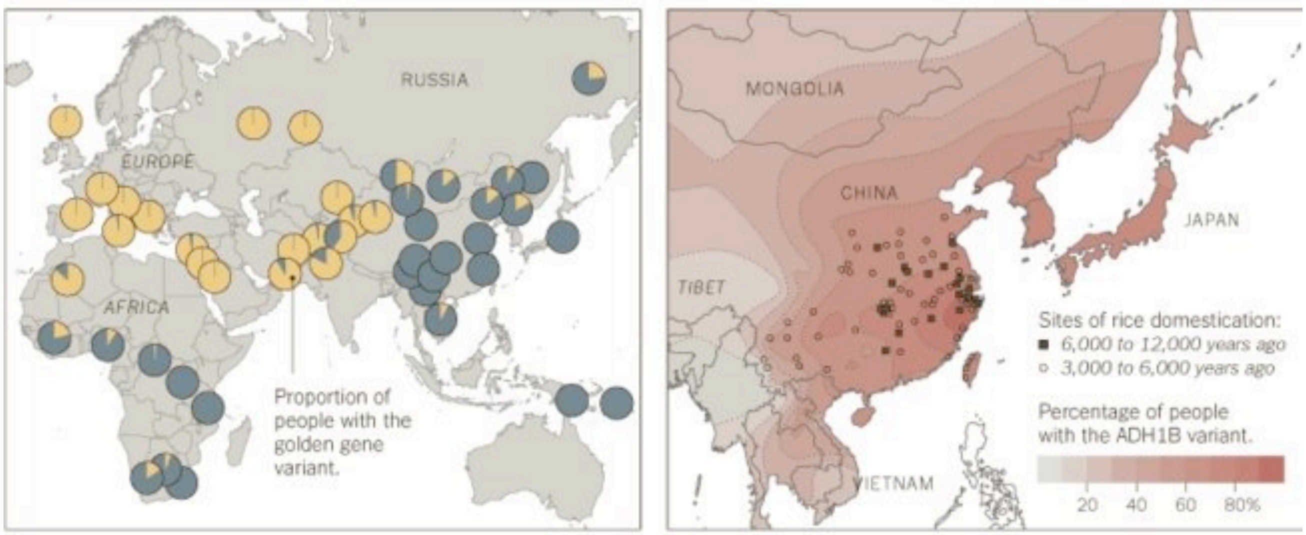
But the new evidence that humans have adapted rapidly and extensively suggests that natural selection must have other options for changing a trait besides waiting for the right mutation to show up. In an article in Current Biology in February, Dr. Pritchard suggested that a lot of natural selection may take place through what he called soft sweeps.

Soft sweeps work on traits affected by many genes, like height. Suppose there are a hundred genes that affect height (about 50 are known already, and many more remain to be found). Each gene exists in a version that enhances height and a version that does not. The average person might inherit the height-enhancing version of 50 of these genes, say, and be of average height as a result.

Suppose this population migrates to a region, like the Upper Nile, where it is an advantage to be very tall. Natural selection need only make the height-enhancing versions of these 100 genes just a little more common in the population, and now the average person will be likely to inherit 55 of them, say, instead of 50, and be taller as a result. Since the height-enhancing versions of the genes already exist, natural selection can go to work right away and the population can adapt quickly to its new home.

Genetic Changes

Researchers have found increasing evidence of recent human evolution in response to local changes in diet, disease and climate.



SKIN COLOR Europeans and Asians probably acquired lighter skin to better synthesize vitamin D. A variant known as the golden gene is found in more than 98 percent of Europeans but is rare in East Asia, where lighter skin is thought to derive from a different set of genes.

ALCOHOL A variation in a gene called ADH1B protects against intoxication, making the skin flush when a person drinks. The variant became almost universal among southern Chinese after rice cultivation and fermentation began about 10,000 years ago.

Sources: Molecular Biology and Evolution; BMC Evolutionary Biology

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