

## How could a modern man arise? A mini review.

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### Abstract

Usually, a modern man as a biological species means that he has conceptual thinking, large neocortex, speech, great physiological flexibility, the ability to adapt to different climatic conditions of the Earth, hairless skin, 46 rather than 48 chromosomes, the existence of purely human forms of pathologies (alcoholism, drug addiction, obesity, atherosclerosis, high altitude pulmonary edema), the ability to long-distance running etc. It is believed that all these unique features of a human are the result of the evolution of specific structural genes. The hypothesis that the broad variability of chromosomal heterochromatin regions, rather than structural genes, played a major role in the origin of modern man is discussed.

**Key words:** human origin; human adaptation; human evolution; human karyotype; heterochromatin regions; human body heat conductivity

### Introduction

It is commonly believed that modern man (*Homo sapiens sapiens*) originated around 150 000 - 200 000 years ago on the African continent. 30,000-50,000 years ago, some of them began to migrate from East Africa, began to populate the Eurasia, and eventually reached South America. This enterprise turned out to be so successful that currently only their descendants live on Earth and other hominins who began to explore Eurasia, Australia and Oceania even earlier than the ancestors of modern man, for unknown reasons, have not survived. By 40 000 years ago however modern people – *H. s. sapiens* - to have been the sole occupants of the Europe and Asia. Why other hominids failed to achieve such success is the subject of long-standing debate. Eventually, man took the top of the food chain and created a civilization.

From our point of view, out of all the existing hominin species, only one was lucky because only he managed to go this way: at first he acquired a hairless skin and a large brain → became the owner of 46 instead of 48 chromosomes, as in other higher primates → managed to adapt to more severe than the climatic conditions of East Africa → invented a tool, clothes and housing → carried out cultural evolution, formed states and created civilizations.

### How did it all start?

We believe that the beginning of human evolution was the acquisition of hairless skin and, as a consequence, the emergence of a large brain. Perhaps this process began at a time when one or more hominid populations found themselves in a very favorable climatic environment rich in high-calorie food.

Let's imagine such a situation; in search of food, some population of the ancestors of modern man found himself near a tropical lake rich in high-calorie food. However, such luck has confronted individuals in the population with a very serious adaptive problem. The fact is that the consumption of high-calorie food [2,3] will inevitably lead to the production of excess thermal energy, which must be removed outside the body in a timely manner so as not to disrupt the temperature homeostasis in the body in a tropical climate. This problem was especially aggravated due to the presence of a fur, which prevented the effective removal of excess heat outside the body. As we believe, this problem eventually led to the disappearance of the fur. Given this, the heat-protection function of hair passed over to the layer of subcutaneous fat and that in the process of evolution skin ceases to be only the barrier-protective organ, it also becomes the organ of sense (for details see [4-6]).

### From 48 to 46 chromosomes.

A fundamental feature of chromosomes in higher eukaryotes, including man, is the presence of two evolutionally consolidated types of genetic material: euchromatin and heterochromatin. Euchromatin, the conservative portion of the genome, contains transcribed structural genes, while heterochromatin, the variable portion of the genome, is predominantly composed of non-transcribed repeated DNA sequences. Heterochromatin is universally distributed in the chromosomes of all the eukaryotes - plants, animals and man, accounting for 10% to 60% of their genome. Chromosomal heterochromatin regions (HRs) account for about 15% - 20% of the human genome [7-11].

To-date two types of constitutive heterochromatin are recognized: C- and Q-heterochromatin. There are several significant differences between

them: C-heterochromatin is found in the chromosomes of all the higher eukaryotes, while Q-heterochromatin - only in man (*Homo sapiens*), the chimpanzee (*Pan troglodytes*) and gorilla (*Gorilla gorilla*) [11-15]. Although chromosomal Q-heterochromatin regions (Q-HRs) exist in the genome of three higher primates, their wide quantitative variability is characteristic only to human populations. Q-HRs variability can be found in man only on seven autosomes [3, 4, 13, 14, 15, 21 and 22], as well as on chromosome Y. Individuals in population differ in the number, location, size, and intensity of staining (fluorescence) of these specific chromosomal regions [7-15].

As the amount of food decreases, populations began to expand the territory of their habitat to more severe, including cold areas. Perhaps, just at this stage in the life history of our ancestors, the adaptive changes appeared in their karyotype, which, ultimately, resulted to the emergence of the modern human. We think it highly probable, that the causes of origin of individuals with 46 chromosomes were climate in the East Africa. Middle and Late Miocene ecology was far from being uniform, and such climatic changes as cooling, aridity, seasonal and diurnal temperature fluctuations gradually became dominant environmental factors [16].

It is old-established fact that the chromosome number in the chimpanzee, gorilla and orangutan was 48 and not 46 as in man. However, cause and effect of such chromosome rearrangement is unknown. We believe that the transition from 48 to 46 chromosomes, as well as changes in the composition, localization and amount of chromosomal heterochromatin regions in the karyotype of the ancestors of modern man turned out to be crucial in his formation as a biological species.

A hypothesis has been proposed that natural selection caused merger of two pairs of autosomes carrying Q-HRs into one chromosome. With the fusion of two pairs of autosomes into a single chromosome, apparently, carried on their short arms of Q-HRs with a very high frequency, the number of autosomes bearing the Q-HRs reduced from nine to seven pairs, as in the modern human. Such chromosome rearrangement resulted in two important consequences: a) chromosomal Q-HRs distributed into seven Q-polymorphic autosomes, so that it was possible to give birth to the individuals with different, including the low, number of Q-heterochromatin; b) in the population individuals with low number of Q-HRs appeared, able to adapt to new, harsher climatic conditions. In the changed climate of the East Africa, individuals with less amount of chromosomal Q-HRs in genome were the most adapted. With the lapse of time, these individuals formed a new population in the new territory, where individuals with the number of 46 chromosomes in the genome began to dominate. Thus individuals with different, including the low, number of Q-HRs, got the adaptive advantage and to colonize new ecological zones of the East Africa and Eurasia (for more details see [17-19]).

It is hard to say why the ancestors of *P. troglodytes* and *G. gorilla* are unable to use this path. However, it can be assumed that due to the high frequency of chromosomal Q-HRs at all Q-polymorphic loci of these two higher primates, the probability of birth in a population of individuals with different, and especially with a low number of Q-HRs in the genome is simply excluded. Moreover, in the karyotype of gorillas and chimpanzees found a special type of Q-heterochromatin, located on the distal ends of some chromosomes [7, 11, 20 and 23 – in gorillas; 20, 21, 22, 23 – the chimpanzee], which excludes the possibility of birth of individuals with different numbers of Q-HRs in the karyotype. As for other hominins, contemporaries of the ancestors of *Homo s. sapiens*, it is difficult to say anything, since we do not know what their karyotype was and how the chromosomal HRs were distributed. Of course, we are far from thinking that in the karyotype of our ancestors the other chromosomal rearrangements did not take place, including those, which related to chromosomes without Q-HRs. They, apparently, took place. However, in

the process of adaptation only those rearrangements preserved that were not eliminated by natural selection.

### The consequences of the new karyotype.

In our opinion, the main consequences of all these complex chromosomal rearrangements are their effect on cell thermoregulation (CT). Based on investigations of chromosomal HRs variability in human populations, condensed chromatin (CC), interphase nucleus and redundant non-coding DNAs in the genome, an attempt is made to justify the view of possible participation of CC in CT. CC, being the densest domains in a cell, apparently conducts heat between the cytoplasm and nucleus when there is a difference in temperature between them.

The mechanism of CT is presented to us as follows. Chromosomes have both internal (repair, recombination, rearrangement, modification, restriction) and external (replication, transcription, packaging, organized movement) molecular activities, which are accompanied, inter alia, by some heat output. If for any reasons the temperature in a nucleus begins to exceed than in cytoplasm there is a need for dissipation of surplus heat outside the nucleus. To do this the nucleus has two options: increasing its volume or increasing the heat conductivity of the nuclear membrane. The first option is limited for obvious reasons. The second option is the more promising one should the heat conductivity of the nuclear membrane be increased somehow. Since the nuclear envelope consists of double-membraned extension of the rough endoplasmic reticulum, the nuclear membrane cannot essentially change its structure. However, it is necessary to remove the surplus heat from the nucleus somehow. Apparently, Nature chose a very simple and effective solution: it increased heat conductivity of the nuclear areas by forming temporary structures in the form of CC around the nucleus, chromocenters and nucleoli. The essence of the proposed hypothesis is the assumption that the CC, nucleoli and chromocenters participate in cell thermoregulation. Namely, they are involved in the removal of excess heat from the "hot" areas of the interphase nucleus through a dense layer of peripheral condensed chromatin in the cytoplasm [20-26].

We have checked this hypothesis on the level of human organism assuming that CT is the basis for heat conductivity of whole cell part of body. It turned out that the level of human body heat conductivity (BHC) really depends on the amount of chromosomal HRs in the genome. Results obtained show that individuals in population truly differ from each other in BHC and its level depends on the amount of chromosomal HRs in human genome [23].

Relevant studies have shown that: a) individuals in a population differ from each other on the level of BHC; b) on the average BHC of males is higher than that of females; c) natives of low altitude regions differ on the average by higher BHC than population of high altitude ones; d) natives of low latitudes differ on the average by higher BHC than populations of high latitudes [22-29].

### The consequences of the loss of hair for human.

What is unusual about the human brain is that we are the only largish mammal whose brain size kept pace with our growth in body size. The plausible reason of this phenomenon is his skin, when after having lost its hair it became the largest and almost universal organ of sense, which begins functioning as early as in the prenatal period of human development.

In the heterochromatin part of genome in the direct ancestors of modern human some changes occurred; in addition to Q-HRs, on three pairs of autosomes [1, 9,16], and on the Y chromosome unusually large C-heterochromatin regions appeared, which do not exist in karyotypes of chimpanzee and gorilla. Thus by the total amount of chromosomal HRs the genome of the *H. sapiens* turned out to be richest one. We assume that

assemblage of the greatest amount of chromosomal HRs in the *H. sapiens* karyotype among the higher primates was the turning point in human evolution, as exactly this circumstance has led to disappearance of hairy cover on his skin. The latter turned out to be the main factor responsible for increase of the brain size during the first years of life of the *H. s. sapiens* [5,6].

Apparently, the main reasons for appearance of hairless skin were the following factors: 1) increase of BHC because of high Q-HRs and C-HRs content in the genome of the direct ancestor of modern human; 2) quantitative and qualitative changes of the diet composition which lead to increase of heat production in the organism demanding efficient heat loss from the body for preservation of temperature homeostasis; 3) climate of Africa, where the ancestors of the *H. sapiens* inhabited, had a strong selective influence on such organisms because their bodies have changed towards high heat conductivity and heat production.

It is believed that the loss of hair was beneficial for human in the sense that if he were not naked, he would not begin to manufacture clothes, which is impossible without availability of fine instruments like awl, needle, and other instruments, production of which is connected with fine coordination of hands' movements. Fire striking and dwelling construction is also impossible without skillful hands even with availability of high intellect. Naked skin made the human not only improve his labor; it also promoted the brain development in another direction. It is possible to say that with appearance of skin a vast area of body has been formed provided with highly differentiated and fine receptors of different signals both from the outer and inner environment. Brain became more complex and started to develop [5,6].

A human, instead of his hair that he has lost, acquired clothes, which is more important in sense that in addition to body protection from cold (physiology) it has an important ethnical and cultural, psycho-emotional, social and economic importance that promotes the technical and scientific progress. In addition, the latter facilitates functioning of the temperature homeostasis in human. Therefore, in reply to the old question, namely; 'has a human survived because he became intelligent, or, he became intelligent only because he managed to survive', we have chosen the second version [17,18].

Humans tend to tropicalize their environment virtually everywhere they go. They do this by hands for the most part (clothing, tools, housings, heating). Apparently, transformation of the frontal extremities into skillful hands, bipedality and erect walking are connected with appearance of hairless skin. All the above, in its turn and to a variable degree they have an influence on the human brain size. Thus, the whole evolution of the BHC, skin and brain size is a result of adaptation to constantly changing temperature conditions of the environment. Therefore, we believe that the increase of the human brain size was not the result of drastically changes of the structural genes. Most likely, it was the consequence of more ordinary events, such as evolution of constitutive heterochromatin in chromosomes, BHC and skin.

By the uniqueness of the human karyotype, we mean not so much the number of chromosomes, but something more. In fact, nothing special is hidden behind the number 46. With so many chromosomes, there are animals and plants. Here, in our opinion, the composition and broad quantitative variability of chromosomal HRs in the karyotype are important. The uniqueness of the human karyotype is as follows: 1) unlike other higher eukaryotes, only humans and two other higher primates contain both types of constitutive heterochromatin – C- and Q-heterochromatin; 2) among the higher primates, the largest amount of chromosomal C-HRs is in the human karyotype, and they are localized on his three autosomes (1, 9 and 16) and on Y chromosome. It is to this circumstance that a human owes the highest BHC; 3) unlike chimpanzees and gorillas, the number of chromosomal Q-HRs in individuals in a

population is different and it ranges from 0 to 10; 4) such a wide quantitative variability is associated with an uneven distribution of the number of chromosomal Q-HRs on seven Q-polymorphic autosomes; 5) the phenotypic manifestation of such biological variability is the difference between individuals in the population from each other in the levels of BHC, with all the ensuing consequences for the body.

## Discussion

It is difficult to imagine that all the known biological features of man – conceptual thinking, large neocortex, speech, great physiological flexibility, the ability to adapt to different climatic conditions of the Earth, hairless skin, 46 rather than 48 chromosomes, the existence of purely human forms of pathologies (alcoholism, drug addiction, obesity, atherosclerosis, high altitude pulmonary edema), the ability to long-distance running, etc. – originated on the basis of the evolution of structural genes. The simplest theoretical calculations suggest that this is impossible. It is known that there is no direct relationship between the number of structural genes and the complexity of the structure of organisms. So, for example, it was found that by the number of genes in the genome, such different higher eukaryotes as the *Arabidopsis* plant, mouse and human do not differ significantly and they have about 20,000 genes each. The emergence of such complex traits would require the participation of many genes and their modifications, which are not present in other mammals.

There is nothing unexpected in this. The fact is that a man does not have a single organ, protein or enzyme, which in one form or another would be absent from other living beings. Apparently, genes already existing in mammals were involved in the emergence of unique human features. If there was anything unusual in all this, it was the wide quantitative variability of chromosomal HRs in the human genome. Thus, a species arose that had no analogues in the history of evolution.

If the loss of the hair cover led to the emergence of a large brain with all the ensuing consequences, then the emergence of 46 instead of 48 chromosomes made it possible for the birth of children in the population with different, including a low number of chromosomal Q-HRs. This made it possible for individuals to be able to adapt to different, including cold climatic conditions. However, the mechanisms of human adaptation to climatic conditions other than the tropics of Africa were fundamentally different from those of animals. The latter have adapted to new climatic conditions for hundreds and millions of years with the involvement of structural genes. The uniqueness of human adaptation also lies in the fact that only he managed to populate the entire surface of the Earth, including such extreme natural environments as the Far North and high-altitude areas, while remaining a single, tropical biological species. Moreover, all this happened in a very short period (about 30 000-50 000 years), an event unprecedented in the history of evolution [1]. We believe that such a high rate of adaptation became possible because a human used a mobile non-conservative part of his genome to adapt to different natural environments (for more details see [4,26,28]).

However, the existence of individuals with different BHC in the population and the settlement of temperate and northern geographical latitudes by humans also had negative consequences; purely human forms of pathology (obesity, alcoholism, drug addiction and atherosclerosis) arose, which are not characteristic of other mammals in natural conditions.

Thus, modern man owes his origin and position in the world of living beings on Earth to three factors: hairless skin, large neocortex and wide variability of the BHC in individuals in the population. They, in turn, are the result of the evolution of chromosomal HRs, and not structural genes. The main driving force that led to the emergence of man was the need to adapt to changing environmental conditions. If the formation of modern man was mainly influenced by the broad variability of chromosomal HRs,

rather than structural genes, then our evolutionary history was very prosaic: no drama, no purpose, no Creator. It is difficult to get rid of the feeling of dissatisfaction, as if the “mouse gave birth to a mountain”.

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