19. Genetic development of the human brain

Humans have extraordinarily large and complex brains, even when compared with non-human primates. An adult human brain is estimated to contain about 86 billion neurons and at least 100 trillion synapses linking neurons.

We know that at least a third of approximately 20,000 different genes that make up the human genome are primarily active in the brain. Aproximately 80–95% of protein-coding genes are expressed in at least one brain region. These genes influence the development and function of the brain, and ultimately control how we move, think, feel, and behave.

What is so characteristic about the development of the human brain is the presence of about 1,100 new human specific genes which do not exist in chimpanzees.

Scientists have identified clusters of genes in the brain that could determine human intelligence. There are two clusters of hundreds of individual genes that are thought to influence all our cognitive functions - including memory, attention, processing speed and reasoning, but we do not know how.

A significant insight into the development of the brain was provided by the Bruce Lahn team at the Howard Hughes Medical Institute at the University of Chicago. Lahn investigated two genes, microcephalin and ASPM across different human populations. He assumed that these genes played a role in our cerebral development since the mutation of either of these genes leads to the severe condition called microcephaly - where the brain is much smaller than normal. Both genes are known to regulate brain size.

The Lahn group found that for both the microcephalin and ASPM genes one predominant variant exists. The team determined that the prevalent microcephalin variant emerged approximately 37,000 years ago, while the dominant ASPM variant appeared about 5,800 years ago. By sequencing the versions of these genes carried by 1,200 people from across the globe they found that the preferred microcephalin variant was common in all but sub-Saharan Africa.

The emergence of the microcephalin variant coincides with archaeological estimates of the movement of Cro-Magnons into Europe about 40,000 years ago and the development of more sophisticated society, rituals, beliefs and art. The appearance of the ASPM variant coincides with the later development of the Sumerian, Egyptian and Chinese civilizations.

One of the study's major surprises is the relatively large number of genes that have contributed to human brain development. They came to the conclusion that the development of the human brain could be the result of many thousands of mutations in thousands of genes. Lahn’s research confirms that the human brain developed in large steps and much faster than could

be accounted for by normal mutation rates. He believes that this was the result of accelerated evolution, but he is unable to explain how this acceleration happened and what the mechanism behind it was.

Several other scientists also observed more rapid alterations of the human genome than would ordinarily have resulted from random mutations. These unexplained changes were estimated to be a hundred times above known mutation rates. The mechanisms of mutations are well researched and known, therefore they could not account for these high rates.