Genetic mutations that boost individual's adaptability have greater chances of getting through to X chromosomes

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One of the most important questions for evolution researchers is how a species develops and adapts during the course of time. An analysis of the genes of twelve chimpanzees has now demonstrated that the chimpanzee X chromosome plays a very special role in the animal's development. The analysis was carried out by researchers at the Bioinformatics Research Centre, Aarhus University, the Section of Bioinformatics, University of Copenhagen, the Copenhagen Zoo and the sequencing centre at the Beijing Genomics Institute (BGI), China. The results have been published online in the *Proceedings of the National Academy of Sciences*.

The background for evolution is found in our genome – in DNA. Evolution is driven by mutations that create changes in the genome on an ongoing basis. These mutations are most often deleterious, but they are sometimes beneficial for bearers in the environment in which they live. Their survivability can be improved, which in turn increases the likelihood of having more offspring. These beneficial variants then increase in number until all the individuals in a species have the new variant. This process is called natural selection.

One X is enough

By sequencing all the genes in twelve chimpanzees from Central Africa, the researchers demonstrated that beneficial variants are accumulated on

the X chromosome in particular.

Why does this chromosome behave in such a special way? The gender of the individual is determined by the X chromosome along with the Y chromosome. Males have one X chromosome and one Y chromosome, while females have two X chromosomes.

A new beneficial variant on one X chromosome in the female can 'hide itself' if it is not expressed as strongly as the old variant sitting on the other copy of the X chromosome. In this case, the new variant is called recessive, i.e. it is suppressed by the other more dominant gene. This means that a new beneficial recessive variant does not immediately provide a benefit for the females. On the other hand, the males only have one X chromosome and it is expressed immediately, thus enabling natural selection to 'catch sight' of it. This does not apply to the remaining twenty-two chromosomes in the genome, because both males and females have two copies of each of these.

One third are good changes

The researchers found that about a third of all the changes that have taken place on the X chromosome since humans and chimpanzees diverged approximately four to six million years ago have been beneficial for the chimpanzee. This is far from the case for the chimpanzee's remaining twenty-two chromosomes. The researchers therefore conclude that most of the new beneficial mutations must be recessive. This was already known regarding deleterious mutations on the X chromosome, e.g. colour blindness as a recessive trait and therefore only occurring in men. However, the new results show that this must also be the case for the more interesting beneficial mutations, i.e. those that develop a species over a period of time.

Disease leads to development

The new results make it probable that corresponding activity takes place in the human X chromosome. They also provide an explanation of previous studies in which it was observed that the X chromosome behaved strangely during the speciation process of humans and chimpanzees, in that it was considerably less variable than the remaining chromosomes. This lack of variation can be explained by the fact that natural selection, which actually eliminates variation, works stronger on the X chromosome than on the remaining chromosomes.

By closely studying all the chimpanzee genes, the researchers found examples of natural selection in the remaining chimpanzee chromosomes all being associated with genes that are important for the immune system, including a gene that gives partial resistance to HIV in humans. This indicates that diseases are one of the most important factors in the evolutionary development and adaptability of the chimpanzee.

Provided by Aarhus University

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