Gene mutation linked to cognition is found only in humans

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The human and chimpanzee genomes vary by just 1.2 percent, yet there is a considerable difference in the mental and linguistic capabilities between the two species. A new study showed that a certain form of neuropsin, a protein that plays a role in learning and memory, is expressed only in the central nervous systems of humans and that it originated less than 5 million years ago. The study, which also demonstrated the molecular mechanism that creates this novel protein, will be published online in *Human Mutation*, the official journal of the Human Genome Variation Society.

Led by Dr. Bing Su of the Chinese Academy of Sciences in Kunming, China, researchers analyzed the DNA of humans and several species of apes and monkeys. Their previous work had shown that type II neuropsin, a longer form of the protein, is not expressed in the prefrontal cortex (PFC) of lesser apes and Old World monkeys. In the current study, they tested the expression of type II in the PFC of two great ape species, chimpanzees and orangutans, and found that it was not present. Since these two species diverged most recently from human ancestors (about 5 and 14 million years ago respectively), this finding demonstrates that type II is a human-specific form that originated relatively recently, less than 5 million years ago.

Gene sequencing revealed a mutation specific to humans that triggers a change in the splicing pattern of the neuropsin gene, creating a new splicing site and a longer protein. Introducing this mutation into chimpanzee DNA resulted in the creation of type II neuropsin. "Hence,

the human-specific mutation is not only necessary but also sufficient in creating the novel splice form," the authors state.

The results also showed a weakening effect of a different, type I-specific splicing site and a significant reduction in type I neuropsin expression in human and chimpanzee when compared with the rhesus macaque, an Old World monkey. This pattern suggests that before the emergence of the type II splice form in human, the weakening of the type I splicing site already existed in the common ancestor of humans and chimpanzees, implying a multi-step process that led to the dramatic change of splicing pattern in humans, the authors note. They identified a region of the chimpanzee sequence that has a weakening effect on the splicing site that also probably applies to humans. "It is likely that both the creation of novel splice form and the weakening of the constitutive splicing contribute to the splicing pattern changes during primate evolution, suggesting a multi-step process eventually leading to the origin of the type II form in human," the authors state.

They note that further studies should probe the biological function of type II neuropsin in humans, as the extra 45 amino acids in this form may cause protein structural and functional changes. They note that in order to understand the genetic basis that underlies the traits that set humans apart from nonhuman primates, recent studies have focused on identifying genes that have been positively selected during human evolution. They conclude, "The present results underscore the potential importance of the creation of novel splicing forms in the central nervous system in the emergence of human cognition."

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