Study finds value in 'junk' DNA

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For about 15 years, scientists have known that certain "junk" DNA -- repetitive DNA segments previously thought to have no function -- could evolve into exons, which are the building blocks for protein-coding genes in higher organisms like animals and plants. Now, a University of Iowa study has found evidence that a significant number of exons created from junk DNA seem to play a role in gene regulation.

The findings, which increase understanding of how humans differ from other animals, including non-human primates, appear Oct. 17 in the open-access journal *PLoS Genetics*.

Nearly half of human DNA consists of repetitive DNA, including transposons, which can "transpose" or move around to different positions within the genome. A type of transposon called retrotransposons are transcribed into RNA and then reintegrated into the genomic DNA. The most common form of retrotransposons in the human genome are Alu elements, which have more than one million copies and occupy approximately 10 percent of the human genome.

"Alu elements are a major source of new exons. Because Alu is a primate-specific retrotransposon, creation of new exons from Alu may contribute to unique traits of primates, so we want to better understand this process," said the study's senior author Yi Xing, Ph.D., assistant professor of internal medicine and biomedical engineering, who holds a joint appointment in the University of Iowa Carver College of Medicine and the UI College of Engineering.

To study the impact of Alu-derived exons on human gene expression, the researchers used a high-density exon microarray. The technology has nearly six million probes for monitoring the expression patterns of all human exons. Using data generated by these microarrays, the scientists analyzed 330 Alu-derived exons in 11 human tissues. The team then identified a number of exons with interesting expression and functional characteristics.

"Hundreds of exons in the human genome were created from Alu elements. The whole-genome exon microarray allowed us to quickly identify exons that most likely contribute to the regulation of gene expression and function," said Lan Lin, Ph.D., University of Iowa postdoctoral fellow in internal medicine and the lead author of this study.

Analysis of one human gene, SEPN1, which is known to be involved in a type of muscular dystrophy, along with comparative data from chimpanzee and macaque tissues, suggested that the presence of a muscle-specific Alu-derived exon resulted from a human-specific change that occurred after humans and chimpanzees diverged evolutionarily.

"In this case, this exon is only expressed at a high level in the human muscle but not in any other human or non-human primate tissue, so this implies that the exon plays a functional role in muscle, and this role is human-specific," said Xing, who is also affiliated with University of Iowa Center for Bioinformatics and Computational Biology.

Source: University of Iowa

https://phys.org/news/2008-10-junk-dna.html