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Humans, chimpanzees and monkeys share DNA but not gene regulatory mechanisms, scientists report at ASHG 2012

Humans share over 90% of their DNA with their primate cousins. The expression or activity patterns of genes differ across species in ways that help explain each species' distinct biology and behavior.

DNA factors that contribute to the differences were described today (Nov. 6) at the American Society of Human Genetics 2012 meeting in a presentation by Yoav Gilad, Ph.D., associate professor of human genetics at the University of Chicago.

Dr. Gilad reported that up to 40% of the differences in the expression or activity patterns of genes between humans, chimpanzees and rhesus monkeys can be explained by regulatory mechanisms that determine whether and how a gene's recipe for a protein is transcribed to the RNA molecule that carries the recipe instructions to the sites in cells where proteins are manufactured.

In addition to improving scientific understanding of the uniqueness of humans, studies such as the investigation conducted by Dr. Gilad and colleagues could have relevance to human health and disease.

“Through inter-species' comparisons at the DNA sequence and expression levels, we hope to identify the genetic basis of human specific traits and in particular the genetic variations underlying the higher susceptibility to certain diseases such as malaria and cancer in humans than in non-human primates,” said Dr. Gilad.

Dr. Gilad and his colleagues studied gene expression in lymphoblastoid cell lines, laboratory cultures of immortalized white blood cells, from eight humans, eight chimpanzees and eight rhesus monkeys.

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They found that the distinct gene expression patterns of the three species can be explained by corresponding changes in genetic and epigenetic regulatory mechanisms that determine when and how a gene's DNA code is transcribed to a messenger RNA (mRNA) molecule.

Dr. Gilad also determined that the epigenetics process known as histone modification also differs in the three species. The presence of histone marks during gene transcription indicates that the process is being prevented or modified.

“These data allowed us to identify both conserved and species-specific enhancer and repressor regulatory elements, as well as characterize similarities and differences across species in transcription factor binding to these regulatory elements,” Dr. Gilad said.

Among the similarities among the three species were the promoter regions of DNA that initiated transcription of a particular gene. In all three species, Dr. Gilad's lab found that transcription factor binding and histone modifications were identical in over 67% of regulatory elements in DNA segments that are regarded as promoter regions.

The researchers presentation is titled, “Genome-wide comparison of genetic and epigenetic regulatory mechanisms in primates.”

About ASHG

The American Society of Human Genetics is the primary professional membership organization for nearly 8,000 human genetics specialists worldwide. The ASHG Annual Meeting is the world's largest gathering of human genetics professionals and a forum for renowned experts in the field.

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