

A new study suggests that so called “junk DNA” might be what separates apes and man - Stuart Fox

<http://www.popsoci.com/stuart-fox/article/2008-09/how-human-got-his-thumbs>

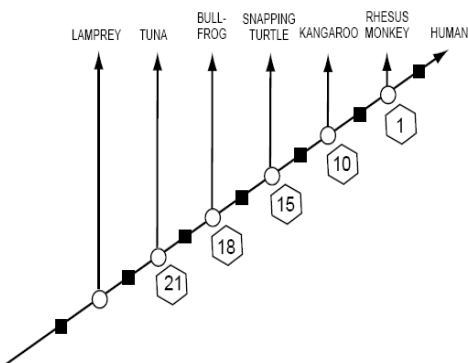
For decades, people referred to the non-coding bits of DNA between genes as junk DNA. Then, in the eighties scientists discovered that some of that junk DNA served an important purpose. The DNA attracted or repelled transcription factors and RNA, greatly enhancing or inhibiting the potency of adjacent genes. Now scientists have just found that one of those gene enhancers may be what separates humans and chimps.

Researchers from U.S. Department of Energy's Lawrence Berkeley National Laboratory, publishing online in the most recent issue of the journal *Science*, placed human, chimpanzee and macaque versions of the enhancer into mouse DNA. The scientists also added a gene that would release a blue dye to show where in the mouse fetus the enhancer was most active. When the mice developed, the researcher saw that it was active in the hands, feet and throat. Additionally, the mice with the human version showed the most activity, with the chimp version producing some activity, and the macaque version producing very little.

The researchers then showed that the difference between the human and chimp versions of the enhancer result from a difference of only 13 nucleotides, a far larger number of changes than would be expected had the mutations been the result of drift rather than selection. The location of enhancer activity highlights the importance of the difference. Our hands, with their opposable thumbs, our feet, evolved for bipedal locomotion, and our throats, which allow us to speak, make up three key differences between humans and all other apes. Because of its role enhancing the genes that regulate the development of those regions, the evolution of this gene enhancer must have been a key step in the evolutionary separation of the human/chimpanzee common ancestor. Furthermore, by following the presence of this particular gene enhancer, researchers should be able to locate which genes are responsible for our differences from chimps and when they evolved. Mapping out humanity's divergence from apes? Not bad for a bunch of DNA once thought of as junk.

The article made one mistake according to what we learned earlier. We learned that not all genes code for proteins. Explain, citing examples from prokaryotes and eukaryotes:

The region of human DNA and Chimpanzee DNA the scientists injected into mouse foetuses is called HACSN1 (take my word for it). This region of DNA has a particular function. Explain



Here is a review of some “cytochrome c” amino acid sequence differences as compared to the human amino acid sequence.

The human “cytochrome c” amino acid sequence is identical chimpanzee’s. Why would that be? _____

Why do the numbers get progressively larger? _____

Here is an 81 nucleotide snapshot of HACSN1; the squares represent the 13 of the total 16 base pair variations mentioned in the article.

		■		■		■		■	■	■	■	■	■	■	■	■	■	■	■	
Human	GCAGCCTTGGGTTCCGCA	AATAGGGGC	ACCCACAGTA	AACG	TGTGGGCGCG	ACCCCGCGG	TGGCGCAATCGGGGCTT	TATAC												
Chimpanzee	A	G		T		T	A	A		T	A	TA								G
Rhesus	A	G		T		T	A	A		T	A	TA								G
Mouse	GT		C	T		T	A	CA		T	A	TA								C
Rat	GT		T			T	A	CA		T	A	TA								C
Dog	GT		T			T	G	A		A	A	TA								C
Chicken	GG	G		TA		T		A		A		T	A	TA						G

Compare the Chimpanzee sequence to the Rhesus sequence. What do you observe? _____

Now compare Chimpanzee and Mouse sequences: _____

Now compare Chimpanzee and Rat sequences: _____

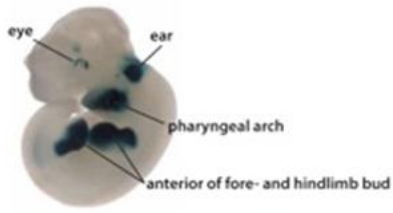
Now compare Chimpanzee and Dog sequences: _____

Now compare Chimpanzee and Chicken sequences: _____

Would scientists expect to see more differences with “junk DNA” nucleotide sequence than with amino acid sequences? _____

Does this region of DNA demonstrate the same pattern as the cytochrome c data? _____

Now compare Chimpanzee and Human sequences. Are there any surprises? _____



No human embryos were used in this experiment, only mouse embryos. Here is a photo of a “transgenic mouse embryo”. Remember that every cell in the mouse embryo has been transformed with the blue dye gene linked to one of the three versions of the enhancer sequence we are studying.

What exactly is a “transgenic mouse embryo”? Why do some (and not all) regions of this embryo stain blue?



Here are three typical “transgenic mouse embryos”. Which embryo has the human HACSN1 and which embryos have the chimpanzee and rhesus versions.



How does the human version of this gene behave differently than the chimpanzee version?



Here is a mouse forelimb expressing the human version of the DNA. Why is this photo remarkable?

How could minor differences at the enhancer level explain how humans differ from chimpanzees?

Taking this line of thinking one level further – How could chromosome rearrangements, including translocations such as Robertsonian Fusions result in Human-Chimpanzee differences.

Why would Darwin be surprised at these results? Why would Niles Eldredge and Stephen Jay Gould NOT be surprised at all?

Most certainly, these scientists will be pursuing their line of experimentation even further. What interesting question will they be addressing next? _____
