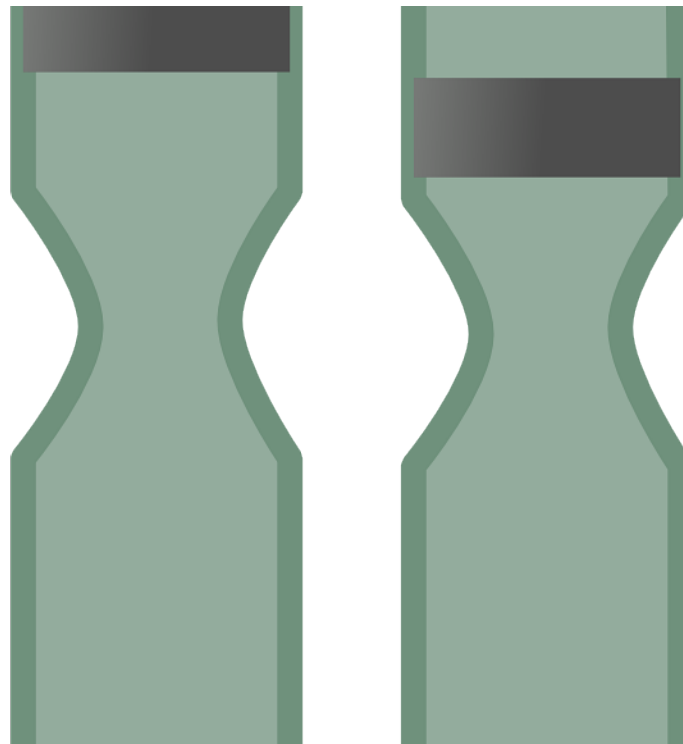


New limits to functional portion of human genome reported

July 14 2017, by Jeannie Kever



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An evolutionary biologist at the University of Houston has published new calculations that indicate no more than 25 percent of the human genome is functional. That is in stark contrast to suggestions by scientists with the ENCODE project that as much as 80 percent of the genome is functional.

In work published online in *Genome Biology and Evolution*, Dan Graur reports the functional portion of the human [genome](#) probably falls between 10 percent and 15 percent, with an upper limit of 25 percent. The rest is so-called junk DNA, or useless but harmless DNA.

Graur, John and Rebecca Moores Professor of Biology and Biochemistry at UH, took a deceptively simple approach to determining how much of the genome is functional, using the deleterious mutation rate - that is, the rate at which harmful [mutations](#) occur - and the replacement fertility rate.

Both [genome size](#) and the rate of deleterious mutations in functional parts of the genome have previously been determined, and historical data documents human population levels. With that information, Graur developed a model to calculate the decrease in reproductive success induced by [harmful mutations](#), known as the "mutational load," in relation to the portion of the genome that is functional.

The functional portion of the genome is described as that which has a selected-effect function, that is, a function that arose through and is maintained by natural selection. Protein-coding genes, RNA-specifying genes and DNA receptors are examples of selected-effect functions. In his model, only functional portions of the genome can be damaged by deleterious mutations; mutations in nonfunctional portions are neutral since functionless parts can be neither damaged nor improved.

Because of deleterious mutations, each couple in each generation must produce slightly more children than two to maintain a constant population size. Over the past 200,000 years, replacement-level fertility rates have ranged from 2.1 to 3.0 children per couple, he said, noting that global population remained remarkably stable until the beginning of the 19th century, when decreased mortality in newborns resulted in fertility rates exceeding replacement levels.

If 80 percent of the genome were functional, unrealistically high birth rates would be required to sustain the population even if the deleterious mutation rate were at the low end of estimates, Graur found.

"For 80 percent of the human genome to be functional, each couple in the world would have to beget on average 15 children and all but two would have to die or fail to reproduce," he wrote. "If we use the upper bound for the deleterious mutation rate (2×10^8 mutations per nucleotide per generation), then ... the number of children that each couple would have to have to maintain a constant population size would exceed the number of stars in the visible universe by ten orders of magnitude."

In 2012, the Encyclopedia of DNA Elements (ENCODE) announced that 80 percent of the genome had a biochemical function. Graur said this new study not only puts these claims to rest but hopefully will help to refocus the science of human genomics.

"We need to know the functional fraction of the [human genome](#) in order to focus biomedical research on the parts that can be used to prevent and cure disease," he said. "There is no need to sequence everything under the sun. We need only to sequence the sections we know are functional."

Provided by University of Houston

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