

Human genetic 'deserts' are teeming with significant life

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Many of the areas of the human genome previously thought to be deserts are in fact teeming with life, a scientist will tell the annual conference of the European Society of Human Genetics today.

Most known human genes in the genome map are still incompletely annotated, says Professor Alexandre Reymond, from the Centre for Integrative Genomics, University of Lausanne, Switzerland and the Department of Genetic Medicine, University of Geneva, Geneva, Switzerland. “We found that the vast majority of the protein coding genes we studied utilised often in a tissue-specific manner previously unknown set of exons [the regions of DNA within a gene that are transcribed to messenger RNA] outside the current boundaries of the annotated genes”, Professor Reymond will say.

He and his team are among the many international collaborators working together on a pilot project to use as a ‘reference set’ by the ENCODE consortium, launched in 2003 by the National Human Genome Research Institute, part of the US National Institutes of Health, with the aim of identifying all functional elements in the human genome sequence. The pilot phase is focusing on 44 regions, totalling approximately 1% of the human genome. By testing and comparing existing methods for annotating the human genome the consortium members hope to find the most effective ways of analysing it in its entirety.

Although the finished sequence of the human genome announced in 2003 was a great achievement, it left a number of questions unanswered.

Before the information contained in the sequence can be used to its best effect, the identity and precise location of all the protein-encoding and non protein-encoding genes in the genome will have to be determined. “The notion that mammalian transcriptomes [the set of all messenger RNA molecules produced in a population of cells] are made of a swarming mass of different overlapping transcripts, together with our findings that suggest we have only uncovered a portion of its complexity, has important implications for medicine, says Professor Reymond. “They increase the size of the genomic regions that might harbour disease-causing mutations, and they could impair cloning strategies that try to find genes implicated in these pathologies.”

They also suggest that extra caution should be used when associating a genetic phenotype with a gene knock-out or knock-in, where genes are added or deleted from a model organism in order to study the effect of therapies, he says. “It appears that the same nucleotide [the structural unit of DNA and RNA] on the genome can carry out a number of different, sometimes simultaneous, functions.”

Professor Reymond’s team, in collaboration with other laboratories in Switzerland, the UK, the US and Spain, now intends to carry out the same kind of analysis for two complete human chromosomes. “I will be surprised if we do not find the same kind of variety in these structures”, he says. “Our work has shown that the human genome is far more complex than anyone could have imagined, even ten years ago. Understanding these complexities is essential to the development of effective and safe genetic medicine in the future.”

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