98% of the human genome: We are finally beginning to understand the mystery of ‘dark matter’ junk DNA

Less than 2% of the three billion letters of the human genome are dedicated to proteins. Only around 20,000 distinct protein-coding genes were found to exist in the long lines of molecules known as base pairs that make up our DNA sequences.

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The remaining 98% of our DNA became known as dark matter, or the dark genome, a mysterious melee of letters with no obvious meaning or purpose. Initially some geneticists suggested that the dark genome was simply junk DNA or the rubbish bin of human evolution – the remnants of broken genes which had long ceased to be relevant.

For others though, it was always obvious that the dark genome was crucial to our understanding of humanity. “Evolution has absolutely no tolerance for junk,” says Kári Stefánsson, chief executive of the Icelandic company deCODE genetics...

Now, two decades on, we have the first inklings of the role of the dark genome. Its primary function appears to be regulating the decoding process, or expression, of protein-making genes. It helps to control how our genes behave in response to all the environmental pressures our bodies face throughout our lives, ranging from diet to stress, pollution, exercise, and how much we sleep, a field known as epigenetics.

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