3200 genes essential to all humans

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Fiddle just a little bit with any one of about 3,200 genes in the human body and you could be toast. That's the conclusion of a new study, which finds that about 15 percent of our 20,000 genes are so critical to our livelihood that even minor mutations can kill us before we're born. The findings should help researchers better track down the genes that cause human disease.

Given how useful any insights into gene function are for understanding the genetic basis of disease, the data are "priceless," says Kári Stephánsson, a geneticist at deCODE in Reykjavík.

The new study, which was recently posted to a preprint repository but has not yet been published in a peer-reviewed publication, was the result of researchers comparing the parts of the genome known as exomes, which code for proteins, from 60,000 people — 10 times more than had ever been attempted. Researchers led by Daniel MacArthur, a geneticist at the Broad Institute of the Massachussetts Institute of Technology and Harvard University in Cambridge, Massachusetts, achieved this feat by reaching out to teams around the world who had collected their own sets of exomes. Among all those genes, the researchers found 10 million variants — places within genes that varied from person to person.

Whenever one of these genes mutates, the embryo usually dies or the person is too sick to reproduce — so the variation disappears.

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