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< Perfection Is Skin Deep: Everyone Has Flawed Genes

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AUDIE CORNISH, HOST:

From NPR News, this is ALL THINGS CONSIDERED. I'm Audie Cornish.

MELISSA BLOCK, HOST:

And I'm Melissa Block. Nobody's perfect. We've all heard those words and likely said them, offering comfort in times of failure. But it turns out it's not just a cliché to make us feel better. It's actually true. Scientists have now documented it on a genetic level. As NPR's Rob Stein reports, researchers discovered that even the genes of typical healthy people have a surprisingly large number of mutations.

ROB STEIN, BYLINE: Scientists have long known that everyone has flaws in their DNA. For the most part, the defects are harmless. But geneticist Chris Tyler-Smith says it's been unclear just how many mistakes are lurking in someone's genes.

CHRIS TYLER-SMITH: It's such an interesting question that people had been trying to make estimates from indirect approaches for a long time. So there were estimates

that ranged from just a handful up to 100 or more serious disease-associated mutations.

STEIN: But Tyler-Smith and his colleagues wanted to get a more precise estimate. So they analyzed the DNA of 179 people who had volunteered to have their entire genetic blueprints decoded. And the researchers are reporting a big surprise in a paper being published in the American Journal of Human Genetics.

TYLER-SMITH: We found quite amazingly large numbers of deleterious and known disease-causing mutations.

STEIN: According to their analysis, the average person has around 400 defects in their genes, including at least a couple that are directly associated with a disease. The weird thing is, none of the people whose DNA was studied seemed sick. They all were perfectly fine.

TYLER-SMITH: It could be that in some cases there's another copy of that gene or a similar gene that takes over. But it could also be that in other cases, the gene is just not needed. We can get by perfectly well, at least in modern society, without it.

STEIN: But about 1 in 10 had genetic mutations that should cause a disease.

TYLER-SMITH: It's a bit surprising that people should be walking around apparently healthy yet we're seeing known disease-causing mutations in their genomes. But the answer was that these tended to be for mild and very often late-onset conditions. Things like heart disease, an increased risk of developing cancer.

STEIN: Other researchers praised the analysis for trying to get a better handle on what scientists know about genetic mutations and disease. Robert Green in a Harvard geneticist.

ROBERT GREEN: I was very surprised by the total number and frequency that they

found. It's fascinating that they have identified so many potentially pathogenic variants.

STEIN: But Green and others say the research also raises big worries. It shows how easily widespread genetic sequencing could find mutations that scare people for what turns out to be no good reason.

GREEN: The critical take-home message here, one that needs to be reinforced over and over, is that as we enter the age of genomic medicine, simply having a mutation in a disease gene does not mean you have the disease or will get the disease. We just are going to have to figure that out gene by gene and disease by disease.

STEIN: So James Evans of the University of North Carolina says the new study shows we're all going to have to get used to that.

JAMES EVANS: Nobody is perfect. We're all mutants. The good news is that most of those mutations do not overtly cause disease, and we appear to have all kinds of redundancy and backup mechanisms to take care of that.

STEIN: And as doctors poke around more and more in their patients DNA, they're going to have to figure out which flaws matter and which are just part of being a normal healthy human. Rob Stein, NPR News.

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