



# Reanimated 'Junk' DNA Is Found to Cause Disease

By GINA KOLATA  
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The human genome is riddled with dead genes, fossils of a sort, dating back hundreds of thousands of years — the genome's equivalent of an attic full of broken and useless junk.

Some of those genes, surprised geneticists reported Thursday, can rise from the dead like zombies, waking up to cause one of the most common forms of [muscular dystrophy](#). This is the first time, geneticists say, that they have seen a dead gene come back to life and cause a disease.

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“If we were thinking of a collection of the genome’s greatest hits, this would go on the list,” said Dr. [Francis Collins](#), a human geneticist and director of the [National Institutes of Health](#).

The disease, facioscapulohumeral muscular dystrophy, known as FSHD, is one of the most common forms of muscular dystrophy. It was known to be inherited in a simple pattern. But before [this paper](#), published online Thursday in [Science](#) by a group of researchers, its cause was poorly understood.

The culprit gene is part of what has been called junk DNA, regions whose function, if any, is largely unknown. In this case, the dead genes had seemed permanently disabled. But, said Dr. Collins, “the first law of the genome is that anything that can go wrong, will.” David Housman, a geneticist at [M.I.T.](#), said scientists will now be looking for other diseases with similar causes, and they expect to find them.

“As soon as you understand something that was staring you in the face and leaving you clueless, the first thing you ask is, ‘Where else is this happening?’” Dr. Housman said.

But, he added, in a way FSHD was the easy case — it is a disease that affects every single person who inherits the genetic defect. Other diseases are more subtle, affecting some people more than others, causing a range of symptoms. The trick, he said, is to be “astute enough to pick out the patterns that connect you to the DNA.”

FSHD affects about 1 in 20,000 people, causing a progressive weakening of muscles in



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the upper arms, around the shoulder blades and in the face — people who have the disease cannot smile. It is a dominant genetic disease. If a parent has the gene mutation that causes it, each child has a 50 percent chance of getting it too. And anyone who inherits the gene is absolutely certain to get the disease.

About two decades ago, geneticists zeroed in on the region of the genome that seemed to be the culprit: the tip of the longer arm of chromosome 4, which was made up of a long chain of repeated copies of a dead gene. The dead gene was also repeated on chromosome 10, but that area of repeats seemed innocuous, unrelated to the disease. Only chromosome 4 was a problem.

“It was a repeated element,” said Dr. Kenneth Fischbeck, chief of the neurogenetics branch at the National Institute of Neurological Disorders and Stroke. “An ancient gene stuck on the tip of chromosome 4. It was a dead gene; there was no evidence that it was expressed.”

And the more they looked at that region of chromosome 4, the more puzzling it was. No one whose dead gene was repeated more than 10 times ever got FSHD. But only some people with fewer than 10 copies got the disease.

A group of researchers in the Netherlands and the United States had a meeting about five years ago to try to figure it out, and began collaborating. “We kept meeting here, year after year,” said Dr. Stephen J. Tapscott, a neurology professor at the [University of Washington](#).

As they studied the repeated, but dead, gene, Dr. Tapscott and his colleagues realized that it was not completely inactive. It is always transcribed — copied by the cell as a first step to making a protein. But the transcriptions were faulty, disintegrating right away. They were missing a crucial section, called a poly (A) sequence, needed to stabilize them.

When the dead gene had this sequence, it came back to life. “It’s an if and only if,” Dr. Housman said. “You have to have 10 copies or fewer. And you have to have poly (A). Either one is not enough.”

But why would people be protected if they have more than 10 copies of the dead gene? Researchers say that those extra copies change the chromosome’s structure, shutting off the whole region so it cannot be used.

Why the reactivated gene affects only muscles of the face, shoulders and arms remains a mystery. The only clue is that the gene is similar to ones that are important in development.

In the meantime, says Dr. Housman, who was not involved in the research but is chairman of the scientific advisory board of the FSHD Society, an advocacy group led by patients, the work reveals a way to search for treatments.

“It has made it clear what the target is,” he said. “Turning off that dead gene. I am certain you can hit it.”

The bigger lesson, Dr. Collins said, is that diseases can arise in very complicated ways. Scientists used to think the genetic basis for medical disorders, like dominantly inherited diseases, would be straightforward. Only complex diseases, like [diabetes](#), would have complex genetic origins.

“Well, my gosh,” Dr. Collins said. “Here’s a simple disease with an incredibly elaborate mechanism.”

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“To come up with this sort of mechanism for a disease to arise — I don’t think we expected that,” Dr. Collins said.

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