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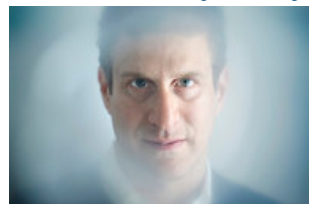
Bits of Mystery DNA, Far From 'Junk,' Play Crucial Role

By GINA KOLATA

Published: September 5, 2012 | 574 Comments

Among the many mysteries of human biology is why complex diseases like diabetes, high blood pressure and psychiatric disorders are so difficult to predict and, often, to treat. An equally perplexing puzzle is why one individual gets a disease like cancer or depression, while an identical twin remains perfectly healthy.

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Béatrice de Géa for The New York Times

"It is like opening a wiring closet and seeing a hairball of wires," Mark Gerstein of Yale University said of the DNA intricacies.

GRAPHIC: Rethinking 'Junk' DNA

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result of the work is an annotated road map of much of this DNA, noting what it is doing and how. It includes the system of switches that, acting like dimmer switches for lights, control which genes are used in a cell and when they are used, and determine, for instance,

Now scientists have discovered a vital clue to unraveling these riddles. The human genome is packed with at least four million gene switches that reside in bits of DNA that once were dismissed as "junk" but that turn out to play critical roles in controlling how cells, organs and other tissues behave.

The discovery, considered a major medical and scientific breakthrough, has enormous implications for human health because many complex diseases appear to be caused by tiny changes in hundreds of gene switches.

The findings, which are the fruit of an immense federal project involving 440 scientists from 32 laboratories around the world, will have immediate applications for understanding how alterations in the non-gene parts of DNA contribute to human diseases, which may in turn lead to new drugs. They can also help explain how the environment can affect disease risk. In the case of identical twins, small changes in environmental exposure can slightly alter gene switches, with the result that one twin gets a disease and the other does not.

As scientists delved into the "junk" — parts of the DNA that are not actual genes containing instructions for proteins — they discovered a complex system that controls genes. At least 80 percent of this DNA is active and needed. The

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whether a cell becomes a liver cell or a neuron.

“It’s Google Maps,” said Eric Lander, president of the Broad Institute, a joint research endeavor of Harvard and the Massachusetts Institute of Technology. In contrast, the project’s predecessor, the [Human Genome Project](#), which determined the entire sequence of human DNA, “was like getting a picture of Earth from space,” he said. “It doesn’t tell you where the roads are, it doesn’t tell you what traffic is like at what time of the day, it doesn’t tell you where the good restaurants are, or the hospitals or the cities or the rivers.”

The new result “is a stunning resource,” said Dr. Lander, who was not involved in the research that produced it but was a leader in the Human Genome Project. “My head explodes at the amount of data.”

The discoveries were published on Wednesday in [six papers in the journal Nature](#) and in 24 papers in Genome Research and Genome Biology. In addition, The Journal of Biological Chemistry is publishing six review articles, and Science is publishing yet another article.

Human DNA is “a lot more active than we expected, and there are a lot more things happening than we expected,” said Ewan Birney of the European Molecular Biology Laboratory-European Bioinformatics Institute, a lead researcher on the project.

In one of the Nature papers, researchers link the gene switches to a range of human diseases — multiple sclerosis, lupus, rheumatoid arthritis, Crohn’s disease, celiac disease — and even to traits like height. In large studies over the past decade, scientists found that minor changes in human DNA sequences increase the risk that a person will get those diseases. But those changes were in the junk, now often referred to as the dark matter — they were not changes in genes — and their significance was not clear. The new analysis reveals that a great many of those changes alter gene switches and are highly significant.

“Most of the changes that affect disease don’t lie in the genes themselves; they lie in the switches,” said Michael Snyder, a Stanford University researcher for the project, called [Encode](#), for Encyclopedia of DNA Elements.

And that, said Dr. Bradley Bernstein, an Encode researcher at Massachusetts General Hospital, “is a really big deal.” He added, “I don’t think anyone predicted that would be the case.”

The discoveries also can reveal which genetic changes are important in cancer, and why. As they began determining the DNA sequences of cancer cells, researchers realized that most of the thousands of DNA changes in cancer cells were not in genes; they were in the dark matter. The challenge is to figure out which of those changes are driving the cancer’s growth.

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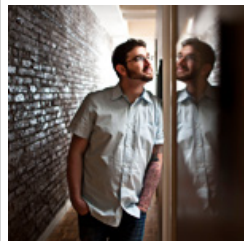
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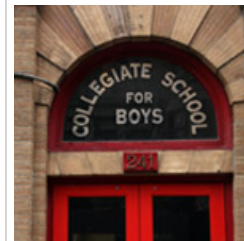
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