Researchers develop way to better predict disease-causing mutations in human genes

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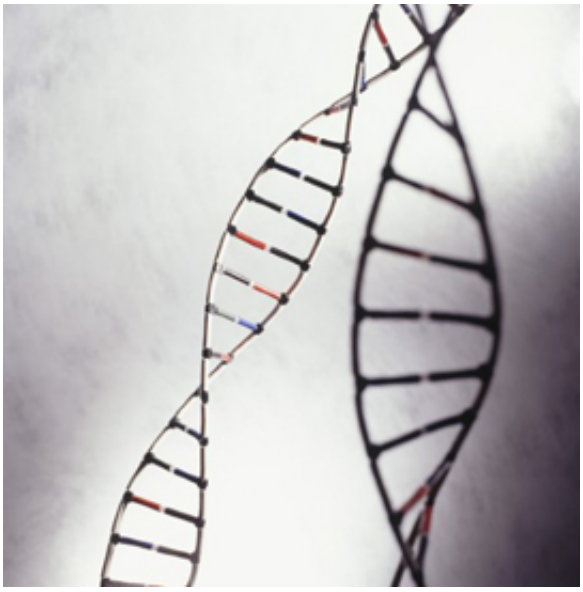

Credit: NIH

Two researchers at the University of Tennessee, Knoxville, have developed a method that could help clinicians and scientists better predict which mutations in people's genes could cause a disease and which would remain dormant.

Using a computational approach, Ogun Adebali and Igor Jouline have created a way to trace the evolutionary history of a human gene much more precisely.

Data gleaned from this method will help doctors know how to more effectively treat patients. It also could be used in the development of drugs to correct some of these mutations.

The study was published today in the journal *Genetics in Medicine* from the Nature Publishing Group.

Adebali, the paper's lead author, is a postdoctoral researcher and Jouline is a joint faculty professor at the UT-Oak Ridge National Laboratory's Joint Institute for Computational Sciences. They're both based in the UT Department of Microbiology.

"We're not a medical school, but this research shows that students who come to UT can tackle important medical problems," Jouline said.

Adebali and Jouline teamed up with two doctors—a physician geneticist and a specialist in lysosomal disorders. Together, they studied the Niemann-Pick disease type C, a rare inherited disorder in children that affects the way cholesterol is transported in the body.

Adebali noted that the disease can cause the spleen and liver to enlarge two to seven times their normal size. Cholesterol can also accumulate in the brain and affect speech and cognitive abilities. Most children with the disease don't survive beyond age 10. It affects one in 100,000 people in the world. For those of Jewish ancestry, the odds increase to one in 40,000.

Computational prediction of the mutation's role in disease is not a novel approach, Jouline said. What's new is the way scientists can improve the accuracy of predictions by tracing the behavior of mutations in the course of gene evolution. Adebali developed a website that will allow clinicians dealing with the Niemann-Pick disease to assess whether mutations seen in patients could be disease-causing.

"Using the same idea, we could potentially improve the diagnostics of other genetic diseases," Jouline said.

Provided by University of Tennessee at Knoxville