From DNA to Disease: A Diagnosis Made in Under a Day

By Julianna LeMieux — February 13, 2018

In the old days, genetic disease diagnoses were identified by the disease first, using the trait that the person had. If you couldn't stop bleeding, it was hemophilia. Black urine meant alcaptonuria. It was not until recently that genetic diseases could be identified by sequencing the DNA first, finding the mutation, and then notifying the person of their disease. (1)

In today's world, genetic diseases can be identified far more quickly and well before someone is exhibiting the associated trait. In fact, they can be made in under a day.

How quickly can a genetic disease be diagnosed?

Scientists at the Rady Children’s Institute for Genomic Medicine [2] (RCIGM) in San Diego have set a record (a Guinness Book of World Record, actually) for how quickly a genome was sequenced and a genetic diagnosis made.

The Rady team, using Illumina’s NovaSeq 6000 Sequencing System and a new high-density flow cells Illumina sequencer, sequenced and analyzed an entire human genome in under 24 hours (it was 19.5 hours.)

That's quicker than some basic blood or other common diagnostic tests. But, that is their dream, says Shimul Chowdhury, who directs the clinical lab at Rady. "That doctors would be able to order genetic analysis with the same routine they use for their other standard labs and get the results back in pretty much the same time frame," he said.

Does genomic sequencing have to be that fast?
Rady's sequencing facility can turn around a DNA sequence plus genetic diagnosis in about 37 hours even though it is not uncommon for this kind of work to take weeks. Does it really need to be done so quickly? It does, if you want to remove weeks of guesswork by physicians.

Rady's Dr. Stephen Kingsmore shares a story of a child suffering from severe liver disease who was heading into the operating room when genetic information showed a different diagnosis and stopped the operation in the nick of time.

Clinical lab director Shimul Chowdhury says that in kids who come in suffering from seizures, time is precious because the longer the seizures last, the more damage to the brain is done. “For these uncontrollable-seizure kiddos, every hour makes a difference, for sure,” he said.

But, speed is not the only goal. The team also is excited by how many sequences can be done at a time. They want to be able to sequence every child who comes in, not just those in a crisis state.

Even though this technology is not at the bedside quite yet, it is a good example of how fast DNA sequencing, something that used to take days, is becoming. And, faster sequencing will, inevitably, lead the way in precision medicine.

Note: (1) Hemophilia is a genetic mutation that causes a lack of a coagulation factor, most often factor VIII. Alcaptonuria is caused by genetic mutations in the HGD gene, which encodes homogentisate oxidase, an enzyme that helps break down the amino acids phenylalanine and tyrosine.