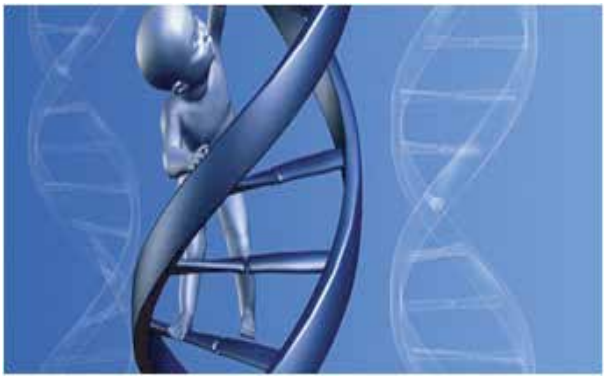
Medical News

Internet of DNA: A global network of millions of genomes could be medicine's next great advance



Noah is a six-year-old suffering from a disorder without a name. This year, his physicians will begin sending his genetic information across the internet to see if there's anyone, anywhere, in the world like him.

A match could make a difference. Noah is developmentally delayed, uses a walker, speaks only a few words. And he's getting sicker. MRIs show that his cerebellum is shrinking. His DNA was analyzed by medical geneticists at the Children's Hospital of Eastern Ontario. Somewhere in the millions of As, Gs, Cs, and Ts is a mis-spelling, and maybe the clue to a treatment. But unless they find a second child with the same symptoms, and a similar DNA error, his doctors can't be sure which mistake in Noah's genes is the crucial one.

In January, programmers in Toronto began testing a system for trading genetic information with other hospitals. These facilities, in locations including Miami, Baltimore, and Cambridge, UK, also treat children with Mendelian disorders, which are caused by mutation in a single gene. The system, called 'Match Maker Exchange', represents something new; a way to automate the comparison of DNA from sick people around the world.

One of the people behind this project is David Haussler, a bioinformatics expert based at the University of California, Santa Cruz. The problem Haussler is grappling with now is that genome sequencing is largely detached from our greatest tool for sharing information: the Internet. That's unfortunate because more than 200,000 people have already had their genomes sequenced, a number certain to rise into the millions in years ahead. The next era of medicine depends on large-scale comparisons of these genomes, a task for which he thinks scientists are poorly prepared. "It's all incomplete and locked down." Haussler is a founder and one of the technical leaders of the Global Alliance for Genomics and Health, a nonprofit organization formed in 2013 that compares itself to the W3C, the standards organization devoted to making sure the Web functions correctly. Also known by its unwieldy acronym, GA4GH, it's gained a large membership, including major technology companies like Google. Its products so far include protocols, application programming interfaces, and improved file formats for moving DNA around the Web. But the real problems it is solving are mostly not technical. Instead, they are sociological: scientists are reluctant to share genetic data, and because of privacy rules, it's considered legally risky to put people's genomes on the Internet.

But pressure is building to use technology to study many, many genomes at once and begin to compare that genetic information with medical records. That is because scientists think they'll need to sort through a million genomes or more to solve cases - like Noah's - that could involve a single rogue DNA letter, or to make discoveries about the genetics of common diseases that involve a complex combination of genes.

If people's DNA data is made more widely accessible, Haussler hopes, medicine may benefit from the same kind of "network effect" that's propelled so many commercial aspects of the Web. The argument for quick action is that the amount of genome data is exploding. The largest labs can now sequence human genomes to a high polish at the pace of two per hour. (The first genome took about 13 years.) Calculations suggest that fast machines for DNA sequencing will be capable of producing 85 petabytes of data this year worldwide, twice that much in 2019, and so on. For comparison, all the master copies of movies held by Netflix take up 2.6 petabytes of storage.

Today scientists are broadly engaged in what is, in effect, a project to document every variation in every human gene and determine what the consequences of those differences are. Individual human beings differ at about three million DNA positions, or one in every 1,000 genetic letters. Most of these differences don't matter, but the rest explain many things and might be responsible for or a higher than average chance of developing serious disorders.

So imagine that in the near future, you had the bad luck

to develop cancer. A doctor might order DNA tests on your tumor, knowing that every cancer is propelled by specific mutations. If it were feasible to look up the experience of everyone else who shared your tumor's particular mutations, as well as what drugs those people took and how long they lived, that doctor might have a good idea of how to treat you. The unfolding calamity in genomics is that a great deal of this life-saving information, though already collected, is inaccessible. "The limiting factor is not the technology," says David Shaywitz, chief medical officer of DNA nexus, a bioinformatics company that hosts several large collections of gene data. "It's whether people are willing."

Last summer Haussler's alliance launched a basic search engine for DNA, which it calls Beacon. Currently, Beacon searches through about 20 databases of human genomes that were previously made public and have implemented the alliance's protocols. Beacon offers only yes-or-no answers to a single type of question. You can ask, for instance, "Do any of your genomes have a T at position 1,520,301 on chromosome 1?"

Privacy, of course, is an obstacle to sharing. People's DNA data is protected because it can identify them, like a fingerprint—and their medical records are private too. Some countries don't permit personal information to be exported for research. But Haussler thinks a peer-to-peer network can sidestep some of these worries, since the data won't move and access to it can be gated. More than half of Europeans and Americans say they're comfortable with the idea of sharing their genomes, and some researchers believe patient consent forms should be dynamic, a bit like Facebook's privacy controls, letting individuals decide what they'll share and with whom—and then change their minds.

"Our members want to be the ones to decide, but they aren't that worried about privacy. They're sick," says Sharon Terry, head of the Genetic Alliance, a large patient advocacy organization.

The risk of not getting data sharing right is that the genome revolution could sputter. Some researchers say they are seeing signs that it's happening already. Kym Boycott, head of the research team that sequenced Noah's genome, says that when the group adopted sequencing as a research tool in 2010, it met with immediate success. Over two years, between 2011 and 2013, a network of Canadian geneticists uncovered the precise molecular causes of 146 conditions, solving 55 percent of their undiagnosed cases.

But the success rate appears to be tailing off, says Boycott. Now it's the tougher cases like Noah's that are left, and they are getting solved only half as often as the others. "We don't have two patients with the same thing anymore. That's why we need the exchange," she says. "We need more patients and systematic sharing to get the success rate back up."

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Medical Joke

Cardiac surgeon vs motor mechanic

A mechanic was removing a cylinder head from a motorbike, when he spotted a famous heart surgeon in his garage. The heart surgeon was waiting for the service manager to come and take a look at his bike.

The mechanic shouted across the garage, 'Hey Doc can I ask you a question?' The famous surgeon, a bit surprised, walked over to him. The mechanic straightened up, wiped his hands on a rag and asked, 'So Doc, look at this engine. I also can open hearts, take valves out, fix them, put in new parts and when I finish this will work just like a new one. So, how come you get the really big money, when you and I are doing basically the same work?'

The surgeon paused, smiled and leaned over and whispered in mechanic's ear, 'Try doing it with the engine running.'

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