

Genetic Testing, Straight to the Consumer

DNA Direct's Ryan Phelan on taking genomic advances, in the form of genetic tests, to the public. Still a challenge: getting physician buy-in and clear rules about DNA privacy.

In 2003, Ryan Phelan founded a direct-to-consumer genetic testing company called DNA Direct. *Genome Technology's* Ciara Curtin spoke with Phelan about the obstacles and ethical issues surrounding genetic testing. What follows are excerpts of their conversation, edited for space.

Genome Technology: Your company serves as a liaison between genetic testing labs and patients or physicians. What are genetic testing labs failing to do that makes your service necessary?

Ryan Phelan: Medical laboratories have never reached out directly to consumers before. Even today it's not very easy for a consumer to go and get their cholesterol levels checked, or their estrogen level. It's usually done through a physician's office. Without going to see a medical geneticist, a lot of consumers don't have access to these tests. It's not so much that the lab is failing to do this; it's really a limitation of the integration of genetics and healthcare.

GT: Scientists have long understood that getting genomic advances into a clinical setting would face a major hurdle in getting physicians on board. How are physicians responding to genetic testing so far?

Phelan: "Slowly" is the shortest answer I can give you. I think they're integrating it slowly. What would change that would be really clear medical guidelines. I think if I said two words, I'd say slowly and cautiously, they are proceed-

ing. They are waiting for really clear indications of clinical utility. If we look at something like cystic fibrosis carrier testing, it's clear that when medical guidelines occur for cystic fibrosis, there was a significant increase in the adoption of that technology by physicians. However, that being said, there are still thousands of patients every year that could benefit from carrier testing on cystic fibrosis that are never made aware of the opportunity for that kind of testing.

GT: There's been controversy about developing diagnostic tests for genetic disorders for which there's no treatment available. How much of a factor is that in determining which genetic tests you'll support?

Phelan: It's a significant factor. One of the things that we look at in test selection is actionability. The medical world refers to the term clinical utility, but we actually refer to actionability by the consumer. Those are different. For a consumer knowing that they may be at risk for particular medical conditions, they may want to make different lifestyle choices. They may want to think about an early pregnancy or no pregnancy. There could be lifestyle changes or behavior that they may want to consider, whereas a physician would primarily be looking at a therapeutic intervention. At the end of the day, we take into consideration all of these factors.

GT: As genomic-based testing becomes more prevalent, do you think diagnostic

labs will have to evolve to deal more with a consumer audience?

Phelan: I think they are already. I think they will always need some interpretation services. Traditionally, a molecular diagnostic lab has a level of interpretation that they are giving, which is purely the laboratory analysis and interpretation of the gene. They are not trying to then provide interpretation of what do you do with this information. They are allowing that to be put into the physician's hands.

GT: Like many nations, the US still doesn't have a genetic information nondiscrimination act signed into law yet. What are your own views on making DNA-based data available when the environment is still uncertain?

Phelan: It really needs to be a decision made by the individual consumer of when and how to integrate genetic information into their medical records and their medical care.

GT: How do you see this market going in the future? Where does it all go from here?

Phelan: Where I hope it goes is that genomic information really becomes integrated in healthcare and consumers can utilize this information with their practitioners to make more informed healthcare choices, prevent disease, prevent the early onset of disease, better target therapies. I'm very bullish on the future of the integration of genomics. **GT**