

GENE SEQUENCING

Nature readers flirt with personal genomics

Survey reveals eagerness to use latest DNA technologies.

BY BRENDAN MAHER

onald Worthington lives an examined life. The genome analyst, who works at Southern Illinois University School of Pharmacy in Edwardsville, got funding from his university to have his entire genome sequenced by Complete Genomics in Mountain View, California. He also ordered the sequence of the coding regions of his genome, known as the exome, from Otogenetics in Tucker, Georgia, and he will soon be giving skin and blood samples that will be used to immortalize his cells and DNA. He aims to contribute it all to the Personal Genome Project, an ambitious effort to sequence 100,000 individuals and post their data and medical histories online for anyone to access.

"This approach is the way to jump-start this whole process of integrating human genomic data into clinical medicine," he says.

Worthington is one of 1,588 people who responded last month to a *Nature* poll on readers' attitudes to personal genomics (see 'Genomes by the numbers'). Participants were recruited by e-mail, and through *Nature*'s online Facebook and Twitter accounts, the Nature News Blog and Genomes Unzipped, an independent blog that chronicles developments in personal genomics. It seems that, overall, *Nature* readers are eager to adopt these new technologies. About 18% report having had their genomes analysed in some way, ranging from whole-genome sequencing (about 10 respondents, after correcting for reporting errors) to direct-to-consumer tests. Of the remainder, 66% say they would have their genome sequenced or analysed if the opportunity arose.

Although scientists dominated our sample, only some 20% of those whose genomes had been analysed reported that their research goals or those of others were a major factor in the decision. About 30% did some or all of the

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analysis themselves. Worthington, for example, isolated his DNA for sequencing and is annotating his exome sequence. But 50% used the services of 23andMe of Moun-

tain View, California, a DNA-testing company that surveys each customer's genome for 1 million of the DNA markers known as SNPs to trace ancestry and to predict disease risks.

Worthington, too, bought a 23andMe kit and says that the results allayed his anxieties about having his full genome sequenced after they revealed no susceptibility to serious illnesses. He also notes the naivety of his rationale, saying: "It is easily possible that next week a genome-wide association study will report that, based upon my 23andMe genotypes alone, I am at substantial risk for some as yet underappreciated terrible disease."

But for other respondents, particularly those in medicine or public health, probing disease risk was a primary motivation. Kelly Leight, who coordinates the group Preserving the Future of Newborn Screening, based in Short Hills, New Jersey, became an advocate of personal genomics after her daughter was diagnosed with late-onset congenital adrenal hyperplasia, a genetic disorder that can be serious if untreated. Leight, her husband and her daughter were all sequenced for the causal gene to confirm the diagnosis. Later, she learned about 23andMe. "When I found out about direct-to-consumer genetic testing, I thought, "This is totally for me. I've got to do this.""

Like many poll respondents, she says her genome is now more of a hobby than anything else. Health-risk information from a genome scan did persuade her to lose 60 pounds in weight, putting her among the 27% of those in the poll who changed their behaviour because of information in their genome. She's spread the word to her family, buying genotyping kits for family members. She says that her geneticist and genetic-counsellor friends are shocked that she would push such a personal decision on her family. But Leight sees no harm. Some family members embraced their genomes, others ignored them. But "I didn't put any pressure on them", Leight says. "Well, maybe I did."