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Most consumers don't know personal genetic tests exist

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The general public seems to have little awareness of, and even less appetite for, personal genomic testing, according to the first multistate survey assessing knowledge and use of direct-to-consumer (DTC) genetic tests. Public health officials in Connecticut, Michigan, Oregon, and Utah added questions about DTC testing to the 2009 Behavioral Risk Factor Surveillance System (BRFSS), a random telephone survey conducted to assess health-care access and use. In this issue of *Genetics in Medicine*, they report that, despite the availability of such tests in 48 states, awareness of DTC genetic testing remains low, ranging from 16% in Michigan to 29% in Oregon. Perhaps more importantly, the survey reveals that use of such tests, which private companies keep proprietary, is exceedingly low—well under 1% in all four states. Those who said they were aware of DTC genetic tests tended to be older, white, educated, and affluent. Most said that they heard of the tests on television or read about them in a newspaper or magazine. Encouragingly, of those who had availed themselves of DTC tests, about half had voluntarily shared results with their health-care providers. This differs from the findings of previous surveys that only about one in four shared results with his or her physician. Overall, the survey shows a significant opportunity for public education about DTC genetic tests, as the number and availability of these tests are only expected to increase in coming years. *—Karyn Hede, News Editor*

Long-term follow-up of adults with 22q11.2 deletion syndrome reveals variability in life skills

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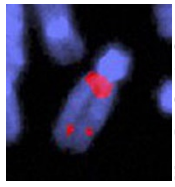

Wessex Reg. Genetics Centre, Wellcome Images

As treatment for inherited disorders improves and more affected individuals survive to adulthood, information about effective interventions to improve quality of life for adults becomes imperative. Anne Bassett and colleagues from the Clinical Genetics Research Program at the University of Toronto describe the findings of long-term follow-up of 100 individuals diagnosed with 22q11.2 deletion syndrome, the most common microdeletion syndrome in humans. Occurring in about 1 in 4,000 live births, the syndrome encompasses cardiac, cognitive, and psychiatric disorders, with a wide variability in severity. Among members of the study group, whose average age was 23, most had difficulty with daily living and communication skills, but a significant subgroup (24 individuals) were functioning relatively normally. They were students or employed and living independently. This group tended to have higher IQ scores, and, significantly, only two had been diagnosed with a psychotic illness. A small group of five with lower IQ—four women and one man—exceeded predicted expectations. All lived in a stable home with relatives or a spouse, and four were being effectively treated for a mood disorder. For health-care providers, these findings may help in providing patients and their families with reasonable expectations as well as in setting long-term goals. The results also point to the importance of support and services for adults with 22q11.2 deletion syndrome to build daily-living skills. *—Karyn Hede, News Editor*

NEWS BRIEFS

Study adds nearly 10% to knowledge of human genetic variation

Western pygmies of Cameroon. Courtesy of Sarah Tishkoff.

A whole-genome analysis of three groups of the world's most genetically diverse people provided an astonishing 3 million new single-nucleotide polymorphisms (SNPs) to the approximately 13 million currently catalogued in publicly accessible databases. The research team, led by Sarah Tishkoff of the University of Pennsylvania, sequenced the genomes of 15 Africans belonging to three distinct groups (Pygmy, Sandawe, and Hadza) that still maintain a hunter–gatherer lifestyle. The sequencing and subsequent statistical analysis provided researchers with a rich expansion in variants, most of which occurred in noncoding sequences. The findings, featured on the cover of the 3 August issue of *Cell*, also suggest that modern humans contain sequences acquired through breeding with a now-extinct early hominid in Africa hundreds of thousands of years ago. As expected, the African genomes contained no traces of Neanderthal DNA. The extent of SNP variation suggests that there remain many more population-specific differences to be catalogued, and that those variations could have important health implications. For instance, although only about 72,000 of the new variants were in coding regions, highly divergent sequences appeared in genes involved in smell and taste as well as immune system–related and growth hormone genes, suggesting that these loci evolved in response to diet, climate and local pathogens. We can expect high-coverage, whole-genome sequencing to reveal more surprises in human evolutionary history, as more ethnically diverse genome data sets are integrated with studies of human health.
—Karyn Hede, News Editor

A significant step forward in the DTC genomics world

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23andMe, arguably the industry leader in the world of direct-to-consumer (DTC) genetic testing, recently announced that it is seeking approval from the US Food and Drug Administration (FDA) for an initial set of seven health-related tests.

I have been a personal critic of the DTC industry in the past. My major concerns have been that interpretation of risk scores for common diseases