

Executive Summary

Direct-To-Consumer Genetic Testing: Business Prospects in the United States covers the business of genetic tests marketed and sold directly to consumers (DTC), with special attention paid to the small, emerging, and definitely attention-getting segment called Personal Genomics. Thanks to the confluence of two transformational technologies that derive from the invention of the Internet and the completion of the Human Genome Project, the availability of genetic tests is migrating from the scientist- and health professional-controlled domains of research (hospital and clinical laboratories) to the wild, wild world of cyberspace, available to anyone, almost anywhere, with the click of a mouse.

This report will address genetic testing services as a consumer business. It will focus on the health-related segment sold over the Internet called Personal Genomics, the DTC business of scanning or sequencing genomes for health-related, and sometimes other, genetic information, which has only recently come into its own in a significant way. How certain will the business outlook be once the “curiosity factor” subsides?

According to the National Institutes of Health (NIH), almost 1,500 genetic tests are already in use, primarily in clinical settings, and some remain in research laboratories. Most of them are for rare, inherited diseases, and only those studying rare diseases know they exist. The average, or even the highly educated, consumer has likely never heard of them, unless they happen to know someone affected.

In a June 2007 Forbes.com article, Piper Jaffray said that doctors and patients worldwide spend \$730 million per year on genetic tests, and the market is growing 20% per year.* If this figure and the 20% growth rate are accurate, that would make the global market approach about \$950 million by early 2009.

*Herper M, Langreth R. Will you get cancer? Forbes.com. June 18, 2007. www.forbes.com/forbes/2007/0618/052.html, accessed February 17, 2009)

Clearly, the genetic tests sold DTC over the Internet, and the newer genome scans or genome sequencing offered by the newest personal genomics companies, comprise only a tiny slice of this larger pie. The vast majority of all firms offering gene analyses and services are privately held, a number are startups, and those that are public have other well-established lines of business to lean on while testing the waters with their first foray selling tests and counseling through cyberspace. The newest entries, prominent for their scientific platforms as well as wide coverage in the media and especially in the genetics blogosphere, are well-funded firms dedicated to DTC genetic testing barely a year out of the gate.

But marketing or selling genetic tests to consumers is not really that new. Though not covered in this report, various parentage and identity tests have been available to consumers since the late 1980s. Some firms that specialized in this space early on now have Web sites with these and additional offerings such as ancestry tests, various pharmacogenetic tests, and nutritional tests based on DNA assessments. Firms offering nutritional (“nutrigenomic”) tests got started in the early 1990s. However, none of these “old timers” have received the attention of the newer genomic scan companies looking at SNPs (single nucleotide polymorphisms) or gene sequencing, for either disease markers or other personal traits, using the latest in gene chip or sequencing technologies.

Historically, genetic testing referred to testing for heritable diseases by examining the metabolic products of single genes, rather than direct DNA analysis. These types of tests are still in active clinical use, such as in the newborn screening programs present in every state, though some may be using more modern chromatographic or spectrophotometric methods for their analyses. By 1995, the NIH redefined genetic testing for a new age, as the “analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes, or karyotypes for clinical purposes which include predicting risk of disease, identifying carriers, and establishing prenatal and clinical diagnosis or prognosis”.*

Myriad Genetics paved the way for DTC personal genomic testing because they took the controversial step of *advertising* their BRCA*Analysis* tests for hereditary breast and ovarian cancers directly to consumers. Initially, these tests could be obtained only through a physician. The consumer response to the advertising campaign was sufficiently strong that it was clear that at least one segment of

*(Acknowledgements and Executive Summary, “Promoting Safe and Effective Genetic Testing in the United States,” www.genome.gov/10002393).

consumers, women concerned about breast cancer (regardless of their family history), wanted genetic information...a clear signal to other firms contemplating the benefits of selling DTC rather than exclusively through physician practices that the time was right for them to launch their businesses. Today, Myriad's tests are available for consumers to purchase on some DTC genetic testing Web sites.

Many doctors and researchers have vigorously criticized the consumer genomics field, saying the nascent industry is trying to ramp up faster than the science can interpret what genomic scans really mean. Some critics have even called for a ban on DTC marketing of such services and for greater oversight from the US Food and Drug Administration (FDA). These concerns, the wave of new gene-chip technologies, and the ease of starting a consumer business offering genetic information has definitely captured the attention of the Department of Health and Human Services (DHHS), which oversees the FDA, and other organizations with regulatory oversight of laboratory tests, as well as the Federal Trade Commission (FTC), an independent agency within the Executive Branch. The federal dialog has started over the medical legitimacy of the genetic tests offered to consumers; their sometimes uncertain clinical validity and utility; how the information is being used; privacy issues; the appropriate way to make sure that good scientific, medical, and ethical standards are set and not compromised and consumers are protected from fraud—all at a time when current regulatory oversight is shared between several agencies. State health departments have already taken action against 13 DTC genetic testing firms. Some companies have faded away as a result, and others passed regulatory muster with flying colors.

The latest market entries that offer genomic scans or gene sequencing for disease markers or other personal traits currently have similar business models but appeal to very different consumer segments. At least one firm is evolving toward a more traditional “medical model,” already partnering with physician groups to help them incorporate genetic information into their practices. For another firm, increasing physician referrals, in addition to consumer orders for specific genetic tests for disorders for which a patient has a family history, are driving the business. Other firms have goals that will lead them in this same direction. But some have put their corporate positioning stake in the ground as being a strictly consumer business, and how they adapt to the realities of a consumer products and services marketplace remains to be seen. Will new consumers keep coming to learn about their predisposition to diseases they have never heard of? Should negative press accumulate about the lack of clinical validation of the tests, and

how the same set of genes can obtain different interpretations from different companies, how will consumers and these companies respond? How will securing repeat business be managed? Will companies adopt any of the well-established consumer research methodologies long relied upon by consumer goods companies to guide their business decisions? When properly executed, consumer research methods have the built-in rigor to forecast with considerable accuracy the market size in units and revenues for a new consumer product or service. The investor community specializing in consumer businesses often demands this kind of information early on, but it appears that biotech investors have not.

For the 40+ DTC companies offering genomic or genetic testing services on the Web—from DNA dating services to scientifically valid, medical genetic tests—it will be more than just technology developments and regulations that shape the future: It will be the consumers themselves. After taking a look at the history, the regulatory environment, and the current business dynamics of DTC genetic testing, and having conversations with executives at some of these dynamic firms, this report will tell you why.