of prophylactic oophorectomy in carriers of BRCA mutations provided a strong rationale for genetic testing in women with a strong family history of breast cancer.\textsuperscript{12c}

These two NEJM studies, one of which was a prospective study of 170 BRCA mutation carriers, and the other a retrospective study of 551 women carrying BRCA mutations, helped validate the importance of predictive diagnostic testing in identifying individuals at high risk of disease, and in taking steps to prevent the disease. Cancer can be prevented in many cases and early detection provides the greatest opportunity for long-term survival among those diagnosed with cancer. Many experts feel that knowledge of an individual’s family history is not sufficient for recommending the risk-reducing oophorectomy surgery. In families at high risk of hereditary breast or ovarian cancer, detection of a mutation in \textit{BRCA1} or \textit{BRCA2} is required in order to know which individuals in the family have inherited mutations and should consider preventive strategies.

DTC Genetic Testing Rapidly Expands Following Myriad’s DTC Marketing Success

Myriad conducted market research with 300 at-risk women, based on criteria for family history of either cancer. Results indicated that 85% would contact their physician about the genetic test for breast and ovarian cancer, and 62% would switch to a provider offering the test. In September 2002, following an intensive regional physician education program, Myriad launched the first ever direct-to-consumer advertising campaign for HBOC in Denver and Atlanta, using television advertising (on programs such as \textit{ER}, \textit{The Practice}, \textit{CSI Miami}, \textit{Providence}, \textit{Oprah}, \textit{Regis and Kelly}, and the \textit{TODAY} show), radio, and print media (including \textit{Better Homes & Gardens}, \textit{Ladies’ Home Journal}, and \textit{Women’s Health Monitor}).

Myriad gave birth to DTC genetic testing that year, following a marketing model similar to one which pharmaceutical companies had been using for their drugs (\textit{i.e.}, “Ask your doctor about….”). Then, from September 2007 through spring 2008, the New England advertising and media campaign was implemented in Boston, Hartford, Providence, as well as in New York, which the company claimed represented about 12% of their market for these tests. A similar campaign was launched in Texas and Florida (about 18% of their market) at the end of September 2008, which was well timed with Ovarian Cancer Awareness Month (September) and Breast Cancer Awareness Month (October). These consumer programs were preceded by medical education campaigns in the same geographic area.
California’s Recent Citations to DTC Companies Were Accompanied by Print Media and Online Fanfare, Especially in the Gene-Blogosphere Community

A more recent effort by California to curb direct-to-consumer genetic testing services overlapped with that of New York and has received considerably more media coverage. It is conceivable that the extensive media attention may have caught state regulators by surprise, since New York cited more than twice as many firms over the course of three years, and no one appeared to notice. Both states are often at the forefront of emerging regulatory policy, so the outcome of the states’ regulatory action had the potential to set a precedent for what other states and the federal government might do in setting policy and regulations that could significantly influence business models and industry growth. On their surface, the regulatory challenges looked more perfunctory than revolutionary, but the positive outcomes for the US industry standard-bearers, Navigenics and 23andMe, strongly suggest that a small but important hurdle has been overcome. The sequence of events and the issues raised are detailed below.

The California Department of Public Health (CDPH) sent identical “Cease-and-Desist” letters on June 9, 2008 to 13 genetic testing companies, listed in Table 2.2. A generic copy of the letter is shown in Appendix B.

Table 2.2. California Issues Cease-and-Desist Orders to 13 DTC Genetic Testing Companies in June 2008

<table>
<thead>
<tr>
<th>Company</th>
<th>Type of Testing</th>
</tr>
</thead>
<tbody>
<tr>
<td>23andMe, Mountain View, CA</td>
<td>Personal genome scans</td>
</tr>
<tr>
<td>* CGC Genetics, Oporto &amp; Lisbon, Portugal</td>
<td>Individual disease gene tests</td>
</tr>
<tr>
<td>deCODE genetics, Reykjavik, Iceland (with several US offices and labs)</td>
<td>Broad genome scans</td>
</tr>
<tr>
<td>DNATraits, Houston, TX</td>
<td>Broad genome scans</td>
</tr>
<tr>
<td>Gene Essence (BioMarker Pharmaceuticals), San Jose, CA</td>
<td>Genome-wide arrays</td>
</tr>
<tr>
<td>** Hair Dx, Irvine, CA</td>
<td>Hair loss assessment</td>
</tr>
</tbody>
</table>

Continued
3.4. Direct-To-Consumer Genetics and Genomics: Personal Genomics Companies Launched in November 2007

The remainder of this chapter will discuss the newly emerging DTC personal genomics and genetics companies, emphasizing those who are well funded and shaping the sector for personal genomic scans, with much fanfare and not without controversy. The analysis will be limited to those firms that market and sell genetic information for health-related decision-making directly to consumers, usually online, with or without physician involvement, rather than companies specializing in genealogy and ancestry, paternity and other family relationship determinations, forensic genetics, and gene-based testing products sold at retail pharmacies.

The recent US entrants receiving the most media attention to their launch are discussed first. Each will likely attract a different type of consumer:

- **23andMe**, who communicates the entertainment value of knowing about one’s genetic traits (apparently deemphasizing genetic disease), and is making “knowing your genes” fun and fashionable.

- **Navigenics’** Health Compass program has a totally medical, health-management approach to genetic analysis. The program includes genetic counseling and is targeted to helping clients make lifestyle decisions should they face meaningful risk of disease.

- deCODEme, a service offered by deCODE genetics, a medical diagnostics and pharmaceutical company that validates its genetic tests using a genetic database comprised of 60% of the Icelandic population, plus over 100,000 people from the United States, European Union, and Asia. The service builds on its foundation in the genetic sciences.

- For $350,000 **Knome** provides individuals with a digital record of their entire genome sequence—not selected single nucleotide polymorphisms (SNPs) that comprise a small fraction (between 1–2%) of the genome and are markers of common diseases, as other companies do. Likely the first US genome sequencing company to offer its services directly to consumers, others are following suit, thus creating another small stimulus to suppliers developing next-generation sequencing equipment to achieve the “$1,000 Genome” goal: rapid sequencing of all the genes on all 23 chromosomes at a fraction of today’s cost.
Chapter 4

UNDERSTANDING CONSUMERS...
THE BIGGEST VARIABLE IN THE PERSONAL GENOMICS BUSINESS EQUATION

There is a very big consideration for the technologically savvy genetic testing companies who have elected to sell their testing services directly to consumers, and that is the consumers themselves. The startups of 2007 have the best scientists and latest technologies. Yet it is not clear that personal genomics companies have applied the same level of rigor and resources to understanding their consumer marketplace that they have to the technology developments which helped make genetic testing for consumers affordable. Had these startups solicited funds from investors specializing in consumer products businesses, they just might have been ushered out the door empty handed and told to come back when they had appropriately sized their market and could back up an estimate of sales revenues based on sound consumer research. Consumer goods and/or services is a different business altogether than medical diagnostics, pharmaceuticals, or healthcare. The compelling genetic technology advances proudly applied to their testing services would likely take a back seat to well-done consumer research that shows just how many consumers of a certain psychographic profile show a strong intent to purchase. They would ask about the size of the addressable market, the characteristics of the consumers most likely to buy, and the data supporting a quantitative estimate of sales and profitability for 3–5 years.

Certainly, these state-of-the art genetics companies don’t want to be placed in the same bucket as more mass-marketed, “low-tech,” but sometimes high-end consumer products, such as refrigerators and ranges, big screen televisions, Lexus and Porsche cars, headache remedies, furniture, electric toothbrushes, vitamins, pricy wrinkle creams, or athletic footwear. These are not their competitors, after all—other
Institute is an independent, non-profit research organization dedicated to understanding human genetic diseases and providing high-quality genetic science resources. The study will be conducted in conjunction with Cooper University Hospital in Camden and other medical centers in the area (www.coriell.org/index.php/content/view/92/257/, accessed November 15, 2008).

Depending on where laboratory-developed testing reform falls within agency priorities for 2009 and beyond, federal agencies may wait to see what these studies start to suggest regarding consumer behavior as results get published along the way, before deciding whether any regulatory action is needed. But perhaps Coriell’s work could play a different role than a scientific and regulatory one…potentially drawing customers in the Philadelphia/Southern New Jersey region away from personal genomics companies because testing and genetic counseling are free to all volunteers. But for the time being, one has to go in person to the Institute to register, sign consent forms, and provide a saliva sample, minimizing the chance that this program will significantly undermine genome-scan sales over the Internet.

### 5.3. An Uncertain Future with Consumers

In the near term, there will be plenty of curious, early adopters and health-oriented consumers with financial means to generate sales of personal-genome scans as long as these services continue to be featured in the media sources used by a variety of consumer segments, from online news, biotechnology blogs, to magazine subscriptions and even traditional newspapers.

However, it is unlikely that these companies will be able to build long-term businesses based on sales to consumers alone. DNA Direct has a distinctive niche in that they fill a need that the current healthcare system cannot, for lack of specialized resources. They are essentially a genetic counseling company that will contract with the right laboratory for the right test for their clients. Physician referrals are starting to come in, and the company helps clients with insurance reimbursement when coverage is obtainable. It appears that DNA Direct may help clients navigate between all the traditional stakeholders at a time when a positive test result may have a significant emotional impact on a client.

Dietrich Stephan of Navigenics sees consumers armed with their genomic data in hand as one means to help drive physicians to learn more about genetics and utilize the new technologies in their practices.
Chapter 6

EXPERT INTERVIEWS

5.1. Linda Avey

Cofounder, 23andMe, Mountain View, CA

Note: Since this interview, 23andMe has significantly lowered the price of their personal genome scan, from $1,000 to $399 in September 2008.

IPR: First I’d like to ask you about the direct-to-consumer genetic testing industry overall. Is anybody making money? Is there even a “market size” yet or is it just too early?

Ms. Avey: It’s too early to say. I think the passage of GINA is really going to open doors and a lot more people will become interested enough in genetic information to feel comfortable getting tested. They will start feeling like it’s no longer something that could be used against them in a negative way.

IPR: While we’re on the subject of GINA, is there anything that GINA does not accomplish that you had hoped it would?

Ms. Avey: Again, I think it’s too early to say. It takes time for these things to get into place and then to see how they protect people in certain situations. It’s hard to forecast what unintended consequences there might be (from the wording of the new law as passed), but overall, we think very positively about its passage.