The current marketplace for direct-to-consumer (DTC) genetic testing is very dynamic and fluid. At least 42 DTC genetic testing companies have been identified to date, and additional market entrants are likely. Firms marketing and selling genetic tests DTC have numerous factors to consider. This report focuses on health-related decision-making applications of DTC genetic tests and examines various components of this emerging business environment:

- The rapidly evolving science and technology
- The complex framework of regulatory oversight
- Social and ethical issues, including genetic privacy
- Consumers’ attitudes toward genetic testing
- Activities of companies shaping this sector
- Views of industry leaders through exclusive interviews

March 2009
Genetic testing has come a long way since the development of the first genetic test in 1963. According to the National Institutes of Health, almost 1,500 genetic tests are now in use. Under the traditional business model, tests are performed with a physician’s involvement, from ordering the test to communication of its results. But with the confluence of the Internet and completion of the Human Genome Project, the procurement of genetic tests is migrating from health professional-controlled domains to cyberspace, where these tests are available to anyone. Marketing and selling genetic tests directly to consumers not only adds a new business model that can exclude the consumer’s physician but raises scientific, regulatory, and even ethical issues.

Direct-To-Consumer Genetic Testing: Business Prospects in the United States aims to sort out these complex issues surrounding the consumer business of scanning or sequencing genomes for health-related genetic information. The Washington-level federal dialog has started over the medical legitimacy of the genetic tests offered to consumers, how the information is being used, and the appropriate way to make sure that good scientific, medical, and ethical standards are set and not compromised. We provide a detailed look at the current US regulatory framework and how the “patchwork quilt” of today’s regulations is ripe for reform.

The latest market entrants offering genomic scans or gene sequencing for disease markers or other personal traits currently have similar business models but appeal to very different consumer segments. We compare and contrast the approaches, offerings, and messaging of various DTC genetic testing firms operating today. We delineate aspects of this emerging business environment that current or prospective participants may need to consider, given its short history and its uncertain outlook as the technology evolves, the regulatory dialog continues, and the voices of many groups—including consumers, physicians, scientists, entrepreneurs, and public interest groups—are heard.

Importantly, the voice of the consumer cannot be understated. Direct-To-Consumer Genetic Testing: Business Prospects in the United States presents a sample of the more recent and relevant research that has been conducted on consumers’ attitudes toward genetic testing, exploring such issues as their willingness and motivation to have a genetic test, how much they would pay for it, who they would share the results with, privacy concerns, and how important they believe physician involvement is. This information is complemented by in-depth interviews with the industry’s thought leaders and pioneers, who share their views on current business dynamics and directions in which the field may go.

Table 3.4. Representative Next-Generation Sequencing Companies

<table>
<thead>
<tr>
<th>Company</th>
<th>Technology</th>
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<tbody>
<tr>
<td>Pacific Biosciences</td>
<td>The SMRT (single molecule, real time) sequencing uses real-time observation of DNA polymers to call bases in a system that could lead to faster turnaround time and longer reads. DNA polymerase uses the DNA molecule to be decoded right down the center, one “letter” at a time. As the enzyme copies the strand, a sequencing machine is reading each letter as it is added. The company hopes to be shipping machines by Q1’10. One goal is to generate 1 megabase of sequence per second.</td>
</tr>
<tr>
<td>Menlo Park, CA</td>
<td>Helicos BioSciences</td>
</tr>
<tr>
<td>Cambridge, MA</td>
<td>The True Single-Molecule Sequencing platform (SMS) enables the simultaneous sequencing of large numbers of strands of single DNA or RNA molecules by using a proprietary form of sequencing-by-synthesis, in which labeled DNA bases are sequentially added to the nucleic acid templates captured in a flow cell. Optimized formulation ensures high accuracy of each base addition, which are detected by the Helicos Single Molecule Sequencer to elucidate the sequence of bound strands. No pre-amplification step is used. The price tag is $3.35 million.</td>
</tr>
<tr>
<td>Complete Genomics</td>
<td>Complete Genomics expects to have a $5,000 genome” in 2009. The CEO reports “…a $4,000 genome was produced in July 2008, but the results have not yet been published… Sequences take an average of 6.5 hours, and concordance rates are… Illumina’s germ cell based on proprietary methods.</td>
</tr>
<tr>
<td>Mountain View, CA</td>
<td>ILMN Source: Insight Pharma Reports</td>
</tr>
</tbody>
</table>

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