ISSUE DIGEST

# Consumer-directed genetic testing

What do you need to know about the new wave of genetic testing?

**PREPARED FOR**

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In this digest:

# What do you need to know about the new wave of genetic testing?

- The Advisory Board's take
- Recommendations for your team
- Highlights from recent publications

**Our Issue Digests cover top strategic priorities for the health plan Chief Medical Officer.**

Each digest focuses on a question, opportunity, or challenge facing the health plan CMO. We summarize key insights—which may include thoughts on relevance, urgency, and value—on a particular issue, offer recommendations for your team, and provide an overview of recent publications on the topic.

*A sampling of other topics covered:*

- **The cost curve is bending—now what?** *How plan CMOs should respond to changing trends in health spend*
- **Why are providers reluctant to engage in downside risk?** *A comparison of findings from the top five industry surveys*
- **What role should plans serve in addressing social determinants of health to improve patient outcomes?** *Identifying effective programs to address patients' non-clinical needs*
- **What are the potential costs of cost-sharing?** *An examination of the health care affordability crisis*
- **What strategies can payers deploy to reduce pharmacy spend?** *Trends in specialty pharmacy utilization*

# What do you need to know about the new wave of genetic testing?

## An examination of the consumer-directed genetic testing industry

### OUR QUICK TAKE

- **The direct to consumer (DTC) genetic testing industry is projected to continue growing in size as it reduces cost to consumers.** DTC genetic testing's market value is projected to be $6.36 billion by 2028. More individuals will access testing as advancements in techniques lower costs. A procedure that costs $100 in 2017 could drop to $1 in 2026.
- **Network physicians will need technological support and clinical training as DTC products change the physician-patient relationship.** These tests will equip members with previously inaccessible data. PCPs aren't geneticists, yet are at the front lines of the delivery system and members expect clinical advice on what to do if they test positive for certain genetically-liked disease. As an example, Kaiser's Smartphrase EHR platform equips PCPs with the tools to interpret patients' direct-to-consumer genetic test results. When a PCP is presented with a DTC test Smartphrase will provide evidence based guidelines for further steps for the patient (no action, or refer to a geneticist and genetic counselor for further testing).
- **Some health plans have started to embrace and reimburse DTC.** Aetna, Humana, and United Healthcare have approved Invitae, a direct to consumer gene vendor, for diagnostic tests for BRCA1, BRCA2 (the breast cancer-susceptibility gene) and ovarian cancer. Members who receive this test may be able to begin preventive action early and avoid more expensive treatments down the road.

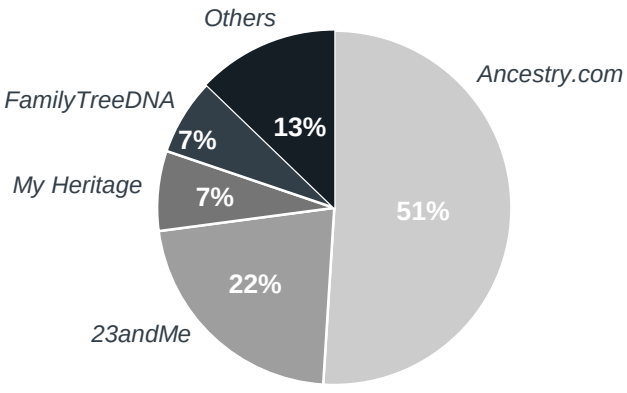
### DATA SNAPSHOT

Rapidly growing direct to consumer industry is dominated by a few players

#### Key DTC genetic testing industry players

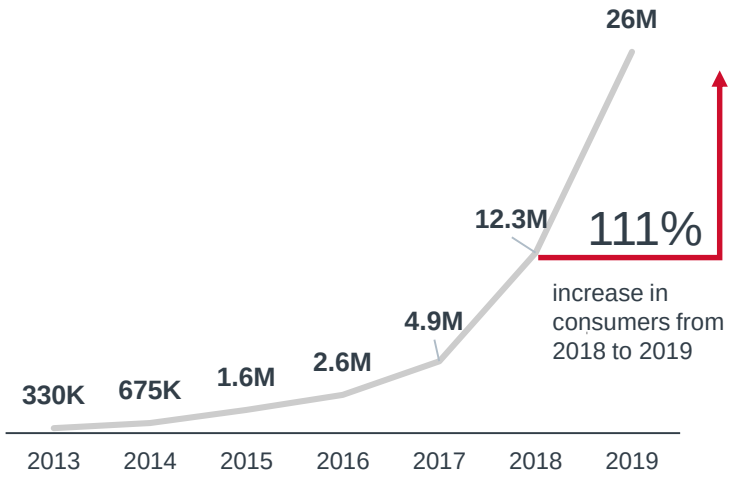
*Market share by volume, global, 2017*

n=~100 products




$821M Global market revenue, 2017

#### Consumers who have completed DTC Genetic testing




Sources: Small, Leslie, "What the rapid growth of personal genetic testing could mean for health insurers" *Fierce Healthcare*, November 2017. https://www.fiercehealthcare.com/technology/what-rapid-growth-genetic-testing-will-mean-for-payers. "Global direct-to-consumer genetic testing market to reach $6.36 billion by 2028" *prnewswire*, May 2019 https://www.prnewswire.com/news-releases/global-direct-to-consumer-genetic-testing-market-to-reach-6-36-billion-by-2028--300853946.html. DTC genetic testing market 2019|top key players analysis, trends, global size forecast to 2025" Reuters. July 2019. https://www.reuters.com/brandfeatures/venture-capital/article?id=137928. Chief Medical Officer Roundtable research and analysis.

# Recommendations for your team

## DISCUSSION QUESTIONS

### Targeted questions to bring to your next team meeting

- How can plans support network physicians to incorporate a patient's DTC genetic test into a medical discussion?
- How would including DTC genetic testing in our product improve its marketability?
- How can we more effectively partner with DTC companies to educate members about their results?
- How can we assure the DTC tests are meeting quality clinical guidelines for reimbursement?

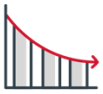
## FACTS TO CONSIDER

### Pros and cons of direct to consumer genetic testing[1]

**Potential benefits of clinical genetic testing**

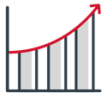
Ability to move beyond diagnosis of existing disease to prediction of future disease

Can improve accuracy of diagnosis and prescribing medication and specialty drugs



With targeted deployment, could lower health care spending by minimizing diagnostic errors and preventing future disease

**Challenges associated with genetic testing today**



Increase in genetic testing could lead to unnecessary health care utilization

Accuracy of DTC tests varies

Physicians unsure how to interpret DTC genetic data

## RELEVANT READING

Recommended for your internal teams

- Blog Post: Time for your annual physical—and genetic screening test?
- Blog Post: Up to 35% of patients would switch PCPs for genetic testing. But is it a good investment for you?
- Daily Briefing: Ancestry will offer health DNA tests, setting its sights on 23andMe
- Daily Briefing: 23andMe already has millions of people's DNA. Now it wants their health data too

External journal articles

- Leighton JW, Valverde K, Bernhardt BA. "The General Public's Understanding and Perception of Direct-to-Consumer Genetic Test Results." Public Health Genomics. 2012;15:11–21.
- Wyckoff AS. "Lack of reliability, privacy plague DTC genetic tests for newborns." Amer Acad Ped. Oct 2018. (accessed 12/28/2018).
- Garg R. "Why do people undergo genetic testing – for personal benefits, for family, or for science and society?" Atlas Sci. Jan 2016.

1. "More Than 26 Million People Have Taken an At-Home Ancestry Test," MIT Technology Review, February 11, 2019; Cathelijne H. van der Wouden et al.,"Consumer Perceptions of Interactions with Primary Care Providers after Direct-to-Consumer Personal Genomic Testing," *Annals of Internal Medicine*, 164:513-522 (2016).

# Highlights from recent publications

### Molecular Genetics & Genomic Medicine (July 2017): Consumers are satisfied with DNA test insights but struggle interpreting results Learn more

- **Summary of major findings:** 67% of participants in the study were highly motivated to explore raw DNA for ancestral information while 62% were interested in individual health implications and 40% interested in both. Approximately 30% of participants shared results with a medical provider and among the 96 consumers (out of 321) who shared with a medical provider, 80% shared with a primary care provider. 47% were motivated by learning about their health shared with their providers versus 9% who were motivated by ancestral knowledge. Participants were highly satisfied with their decision to analyze raw DNA while citing difficulty interpreting results from the data.
- **Methodology:** Study participants were recruited online from social media platforms. A total of 321 survey respondents reported using third-party services for raw DNA interpretation.
- **Limitations:** This study recruited participants via social media through paid advertisements (Facebook/Twitter) and posts on relevant discussion threads (Reddit) which may favor attracting consumers with similar experiences.

**ADVISORY BOARD INSIGHTS**

Health plans will likely see an increase in members submitting claims for DTC testing either directly from the member or through providers.

Plan CMOs should establish and update medical necessity guidelines to account for these DNA tests.

### Associated Press-NORC (July 2018): Consumers remain highly interested in DTC testing despite clear privacy concerns Learn more

- **Summary of major findings**: Among adults age 18 and older who responded to the AP-NORC poll on genetic testing (n=1,109), 17% had been genetically tested and another 52% were interested in having it done. However, 50% were extremely or very concerned that for-profit DNA testing companies would share genetic information without consent while 32% shared those feelings about medical doctors. While about 4 in 10 respondents say a DNA test can be used to identify ethnic heritage or diagnose disease, only a third are convinced about its ability to predict disease or influence proper treatment.
- **Methodology**: Survey was conducted by NORC at the University of Chicago from June 13 to 18 2018, using the AmeriSpeak Omnibus® survey. Participants were contacted by web and telephone interviews in either English or Spanish.
- **Limitations**: The overall margin of sampling error is +/- 4.1%.

While many consumers voice concerns over privacy, a majority are still interested in getting genetic testing so plans should consider partnering with trusted brands of DTC companies on messaging and education, as well as encouraging members to receive consultations from a geneticist once receiving their DNA test results.

### Genetics in Medicine (March 2018): False positives in DTC raw data shows importance of clinical guidance for consumers Learn more

- **Summary of major findings:** If requested, some DTC companies provide raw genotyping data to customers. This data may include variants in genes that are reported as “secondary findings”. This study finds that 40% of variants in a variety of genes reported in the DTC raw data were false positives. In addition, some variants designated with the “increased risk” classification in direct to consumer raw data or by a third-party interpretation service were classified as benign several clinical laboratories.
- **Methodology:** This study collected and identified 49 patient samples for further testing. These samples featured previously identified genetic variants reported in the DTC raw data. Testing at the clinical laboratory was performed by sequencing analysis.
- **Limitations:** The sample size is small and the findings are limited to the genes for which the selected clinical laboratory offered testing.

While uptake is on the rise, the validity of DTC tests needs to be further vetted by health care institutions. Raw data can be easily misinterpreted by consumers and even untrained providers, opening the door for unnecessary care utilization and increased costs.

CMOs should establish guidelines for how providers should engage with members who bring in direct to consumer tests for further interpretation.

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