

*2018 Edition*

# The Current Landscape of Genetic Testing

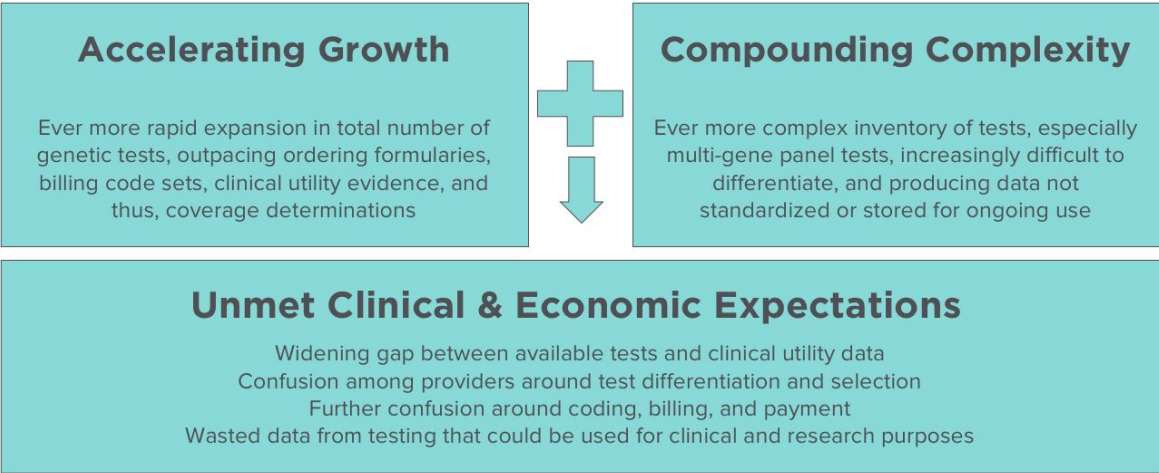
Market growth, reimbursement trends,  
challenges and opportunities

April 2018

# EXECUTIVE SUMMARY

Concert Genetics is a software and managed services company that advances precision medicine by providing the digital infrastructure for reliable and efficient management of genetic testing. Now in its third edition, Concert publishes this report to present the current landscape of clinical genetic testing in the United States. Key points addressed in this document include:

**1. Continued testing growth, especially in multi-gene panel tests, is compounding complexity across the system.**



**2. Healthcare organizations face operational, clinical, and analytic challenges.**

<b>Challenges for Healthcare Providers</b>	<ul style="list-style-type: none"> <li>Complex test order workflow increasingly draws upon staff time</li> <li>Reimbursement issues compound (e.g. denials, dissatisfied patients)</li> <li>Failure to track testing/results in clinical record compromises quality and continuity of care</li> </ul>
<b>Challenges for Health Plans</b>	<ul style="list-style-type: none"> <li>Inability to adapt policies for growing number and variety of tests</li> <li>Compounding confusion in payment integrity</li> <li>Compromised relationships with providers and members from unmet expectations</li> </ul>
<b>Both</b>	<ul style="list-style-type: none"> <li>Paying more than necessary for genetic testing</li> <li>Losing pace with the market</li> <li>Missing opportunities to harness the value of precision medicine</li> <li>Suboptimal quality/continuity of care with downstream ramifications</li> </ul>

## EXECUTIVE SUMMARY (cont'd)

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3. Improved data and digital infrastructure with the right collaboration models make it possible to realize the benefits of precision medicine.

<b>Right Test</b>	<b>Right Patient</b>	<b>Right Price</b>
Accurate identification of tests to reduce errors and inefficiencies in communication	Evidence-based implementation of genetic tests to provide the right information for patients at the right time	Consistency and transparency in pricing so that all stakeholders may have more clarity and trust

# TABLE OF CONTENTS

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## **1. Market Size**

How many tests are currently on the market?

## **2. Market Growth**

How quickly is the U.S. genetic testing market growing?

## **3. Multi-Gene Panels**

What's the status of the market for multi-gene panels?

## **4. Exome Sequencing**

What's the status of the clinical integration of whole exome sequencing?

## **5. Billing & Reimbursement**

What are the trends and challenges in reimbursement?

## **6. Solutions & Other Resources**

What solutions does Concert Genetics propose?

# 1. MARKET SIZE

## A. Total Testing Products on the Market

Concert Genetics tracks detailed information on the clinical genetic tests offered by U.S.-based laboratories, including changes to those tests over time. Concert uses the term **Genetic Testing Unit (GTU)** for any orderable combination of analytes (e.g., genes or other targets) and techniques at a specific point in time, sold by a laboratory as a single item in the laboratory's catalog. A GTU is often equivalent to a checkbox on a test requisition form.

As of March 1, 2018, the total number actively marketed by CLIA-certified laboratories in the U.S. was **74,448 GTUs**. While the majority of these tests are single-gene tests, an increasingly large number are multi-gene panel tests, whole exome sequencing tests, and other complex testing products. Later sections will discuss these trends.

Since its founding in 2010, Concert has indexed more than **200,000 GTUs**. Many of these are now inactive, either because the lab stopped offering them, or because the test changed so substantially that it was assigned a new GTU.

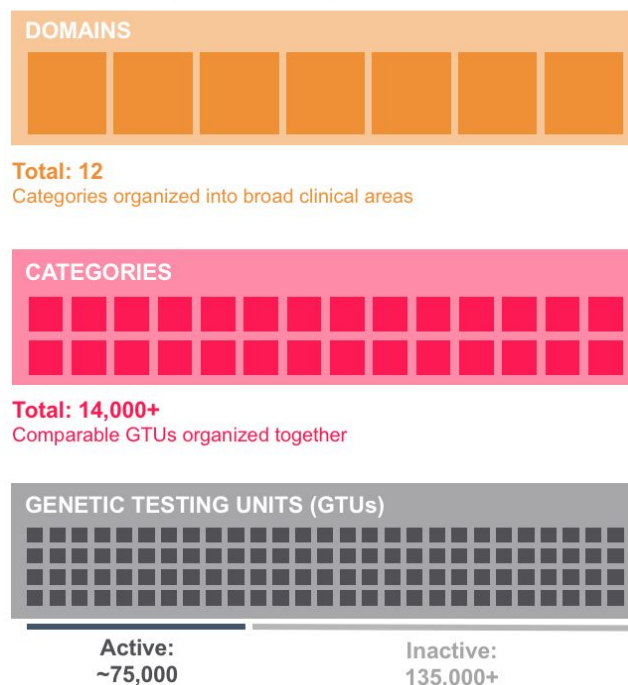
## B. Catalog Structure

Concert's data structure includes a multi-level hierarchy to enable more sophisticated analyses and software solutions. GTUs are the most granular level. At the higher levels are:

**Domains:** These are broad clinical areas, such as Prenatal, Hereditary Cancer, and Neurology.

**Categories:** These group GTUs with the same clinical indication or those designed to answer the same clinical question. Examples of categories: CFTR Targeted Mutation Tests, Hereditary Breast and Ovarian Cancer Panel Tests, and Whole Exome Sequencing Tests.

### Test Catalog Taxonomy



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Key Takeaway:

There are almost 75,000 active genetic testing units (GTUs)  
on the market.

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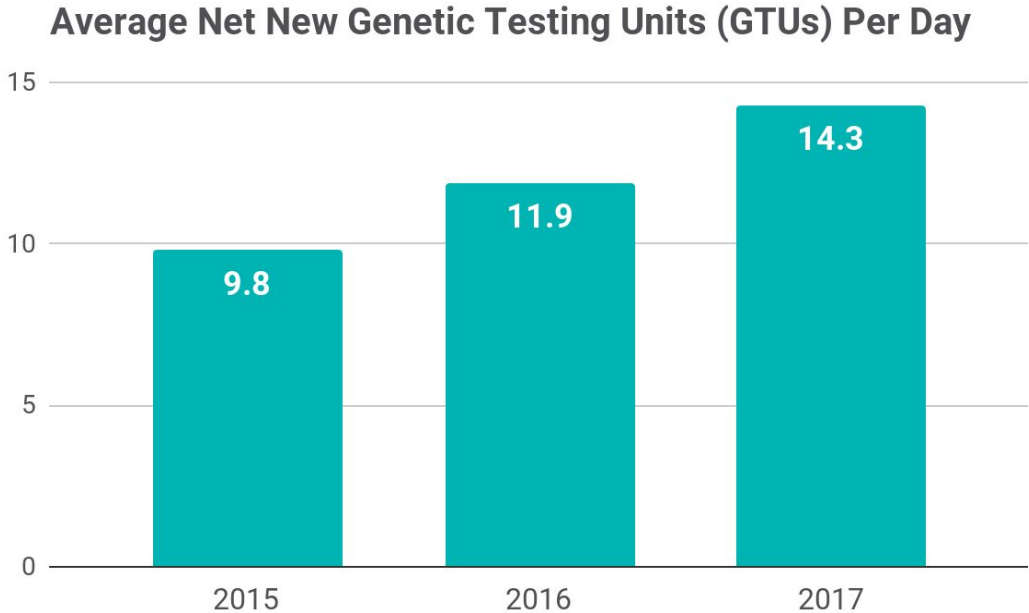
# 2. MARKET GROWTH

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## A. Total New Products on the Market

Concert tracks net new GTUs entering the market on a monthly basis. “Net new” means the total number of GTUs that entered the market minus the total number retired, removed, or otherwise made unavailable from lab catalogs. These net new numbers continue to show remarkable growth. During 2017, a total of **5,210 net new GTUs** entered the market.

Not only is the number of GTUs in the market growing, but the rate at which new testing products enter the market is accelerating. On average, more than **14 GTUs per day** are entering the market – faster than in previous years, as shown here.



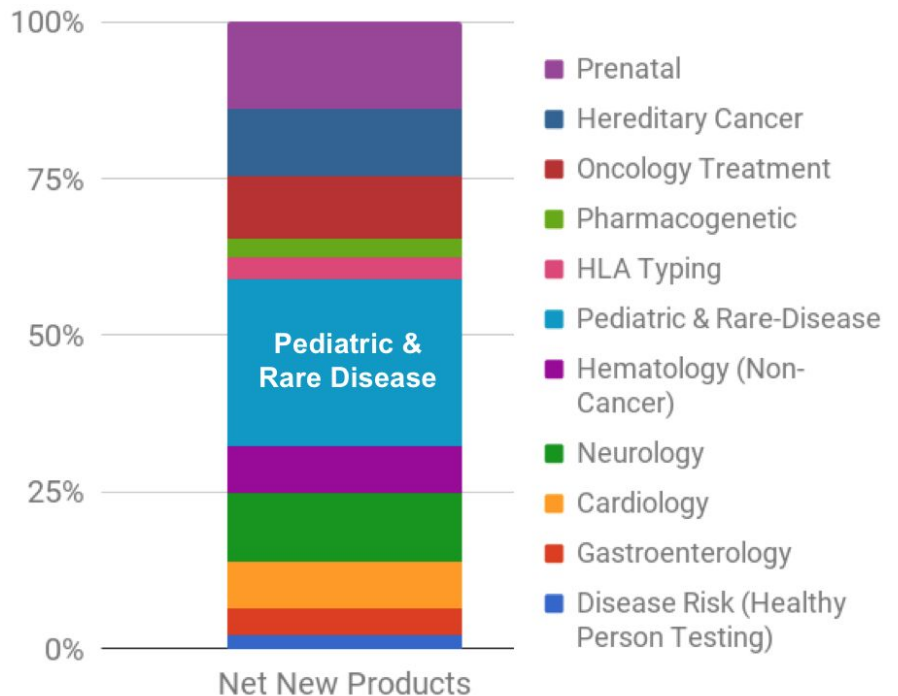
Not all tests are novel. Many are directly comparable and/or competitive to tests that already existed on the market. Others are major modifications to previously released tests. Still, the pace of new test introduction is accelerating.

## B. Clinical Domain of New Products

New GTUs introduced in 2017 are distributed across many clinical domains. One of the notable aspects of clinical genetic testing is its breadth of application across medical specialties and settings.

However, new tests are not evenly distributed across domains. The number of Pediatric and Rare Disease tests grew faster than any other domain. Prenatal, Cancer, Hematology and Neurology also saw a substantial proportion of new tests.

As shown later, the distribution of newly launched tests differs significantly from patterns in test reimbursement, suggesting a significant lag between test introduction and insurance coverage.



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### Key Takeaway:

The genetic testing market is growing rapidly, with growth across many specialties and clinical settings. This rapid pace of growth presents challenges to stakeholders across the healthcare system.

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## 3. MULTI-GENE PANELS

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### A. Market Growth in Panel Testing

The market for multi-gene panels has continued to grow. During the 12 months ending March 1, 2018, a net total of **801 new panels** entered the market. These testing products are entering the market at a rate of 15+ per week, and this year's new entrants represent more than 8% of the total number of panels (9,488) on the market.

Support and guidance for the use of panels has grown, too. In March 2017, the National Society of Genetic Counselors (NSGC) issued an endorsement for the use of multi-gene panel tests when clinically warranted and appropriately applied.<sup>1</sup> The same month, the American College of Obstetricians and Gynecologists (ACOG) issued an opinion that expanded carrier screening panels (often quite large) are acceptable for pre-pregnancy and prenatal carrier screening.<sup>2</sup> Meanwhile, the the American Medical Association (AMA) launched education programs on the use of precision medicine tools, notably including expanded carrier panels and somatic cancer panels.<sup>3</sup> Finally, just last month, the Centers for Medicare and Medicaid Services (CMS) finalized a National Coverage Determination that covers tests using next-generation sequencing (NGS) for patients with advanced cancer.<sup>4</sup> Their statement indicated a determination that, even beyond the narrow case of companion diagnostics, results from NGS can help determine a patient's candidacy for cancer clinical trials.

### B. Example: Panels with *BRCA1/2*

Hereditary breast and ovarian cancer (HBOC) testing continues to grow and increasingly consists of panel tests. *BRCA1/2* testing emerged in the 1990s as one of the first commercially available clinical genetic tests, and the first covered by many health plans. For many years, the predominant clinical application of this was as single-gene assays for *BRCA1* and *BRCA2*, later with the addition (often sequentially) of *TP53*. However, in the years since the U.S. Supreme Court overturned patents on *BRCA1/2*, several things have unfolded:

- Many labs started offering *BRCA1/2* testing
- Labs have assembled panels that include *BRCA1/2* and many other gene combinations
- Clinical applications of *BRCA1/2* have expanded beyond HBOC

(1) <https://www.nsgc.org/p/bl/et/blogaid=870>

(2) <https://www.acog.org/-/media/Committee-Opinions/Committee-on-Genetics/co690.pdf>

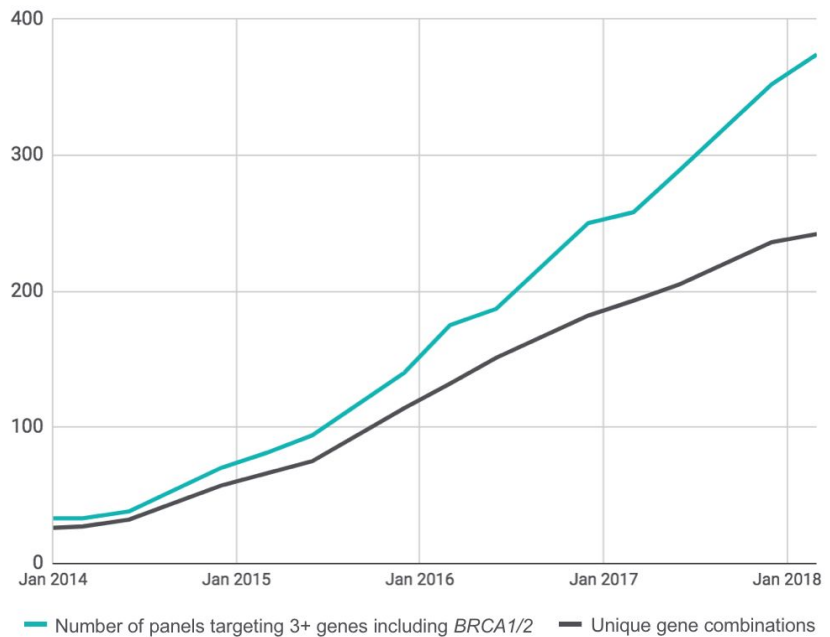
(3) <https://www.ama-assn.org/ama-announces-new-modules-help-physicians-use-cancer-panel-testing>

(4) <https://www.cms.gov/Newsroom/MediaReleaseDatabase/Press-releases/2018-Press-releases-items/2018-03-16.html>

As a result of these factors, the volume and variety of panels including *BRCA1/2* has grown rapidly. Between January 2014 and March 2018, the total number of panels with 3+ genes including *BRCA1/2* grew by more than 10x. Furthermore, 242 (65%) of the 374 GTUs that meet these criteria offer unique gene combinations.

This convergence of volume and complexity introduces a number of challenges that inhibit the integration of genetic testing into the healthcare system. For example, an increasing number of unique panels makes head-to-head clinical utility and value comparison difficult. Conducting a prospective study for each new panel is impractical. However, without sufficient evidence, clinicians may struggle to identify the best test for a clinical scenario, and health plans may not have sufficient information to update medical policies and issue favorable coverage determinations.

### Number and Variety of Multi-Gene Panels Including *BRCA1/2*



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### Key Takeaway:

Multi-gene panels are growing by 15+ per week, while new guidelines and endorsements from clinical organizations have emerged. However, the growing variety of testing options also introduces challenges for both clinical test selection and insurance coverage.

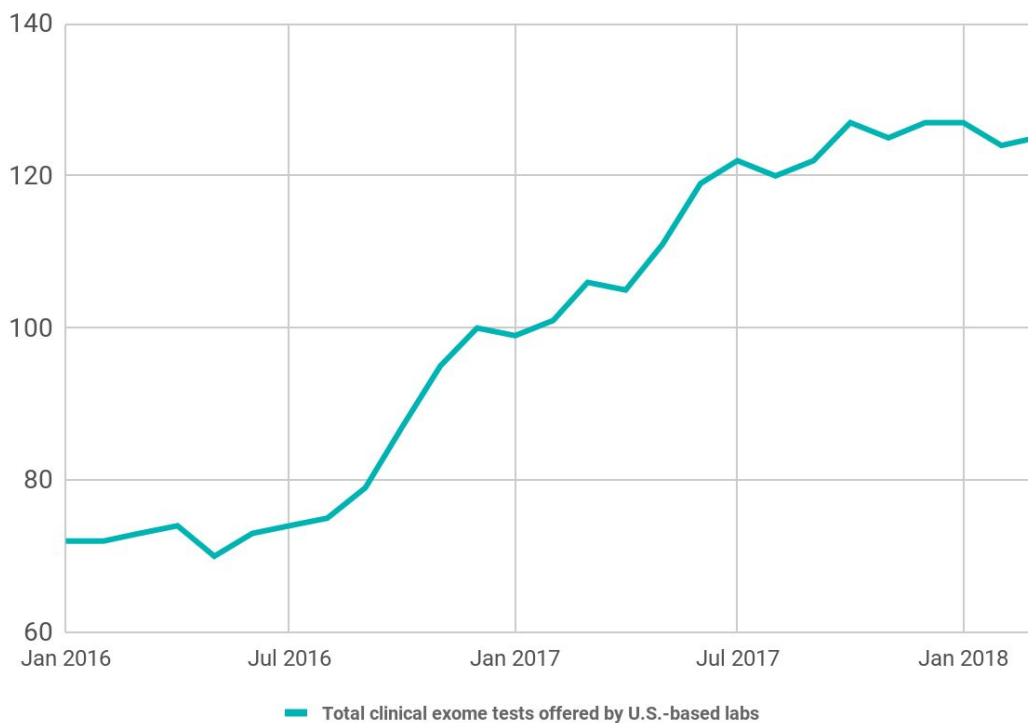
## 4. EXOME SEQUENCING

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Whole exome sequencing is another area seeing growth. Clinicians interested in this testing often see it as a more comprehensive diagnostic approach than single-gene, panel, or some microarray testing – yet offering more focused and medically actionable results than whole genome analysis.

Labs, in response, are increasingly adding exome sequencing tests to their test catalogs. Between January 2016 and March 2018, the number of exome sequencing GTUs in the market grew from 72 to 125 (a 74% increase)

### Number of Available Whole Exome Sequencing Tests



Exome sequencing tests on the market saw particularly rapid growth between July 2016 and July 2017. During this time, more than 40 new exome sequencing GTUs were released.

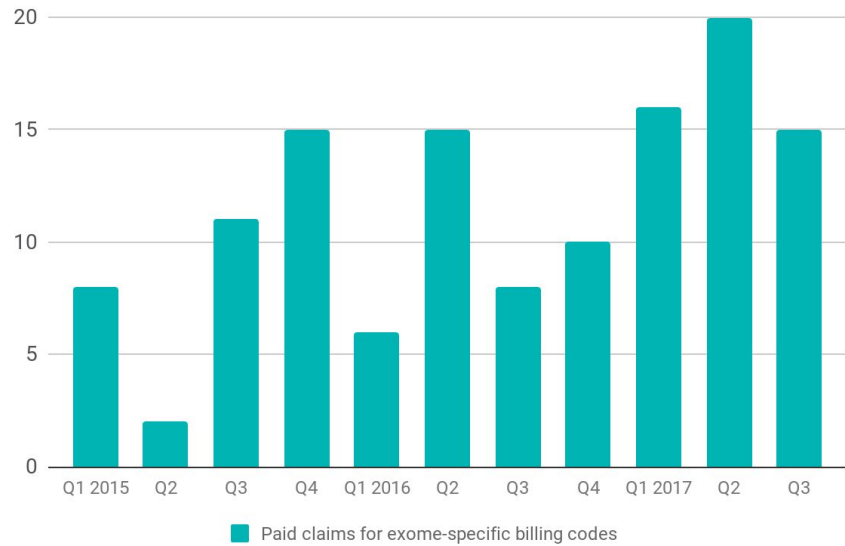
Despite this increase in available exome sequencing options, Concert data suggests that health plans are not frequently paying for this testing.

An analysis of claims from tens of millions of commercially insured health plan members across the U.S. shows that, while the number of paid claims is on the rise, the total volume is marginal.

In this 33-month period, the maximum number of commercial health plan paid claims in any given quarter for the two billing codes specific to exome sequencing testing was just 20 – a fraction of a

percent of the hundreds of thousands of paid claims across all genetic testing for these time periods in this data set. While it is likely that some exome sequencing tests are being billed and paid under miscellaneous molecular CPT® codes, this data suggests that exome sequencing only makes up a tiny portion of overall reimbursements for genetic testing (see Section 7: Billing and Reimbursement).

**The Volume of Exome Sequencing Tests Paid by Commercial Insurance Remains Low**



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### Key Takeaway:

Exome sequencing tests are increasingly introduced to the market, yet health plan payments for these tests are infrequent.

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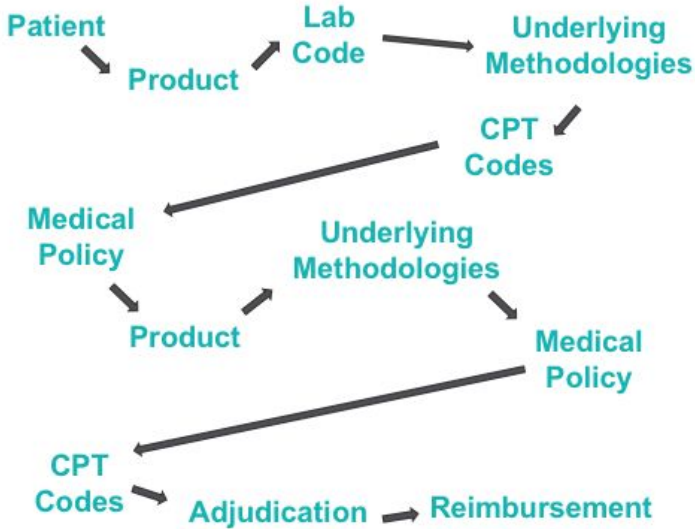
# 5. BILLING & REIMBURSEMENT

The market has seen increased demand for, and utilization of, genetic tests over the past several years. Managing this influx is not simple for health plans, as medical policy, claims adjudication and payment for molecular tests are evolving rapidly. Health plans, institutions, and laboratories struggle to keep up with the pace of change. This is, in part, because detailed data on genetic testing reimbursement is challenging to access.

At least three major factors inhibit a market-wide understanding of the billing and reimbursement picture in genetic testing.

- 1. **Aggregate payment data is difficult to obtain**, especially for stakeholders who aren't health plans.
- 2. **Data is irreversibly converted or lost as it crosses the system.** The graphic to the right shows many stages at which data is modified as it traverses the healthcare system.
- 3. **Health plans claims do not translate to test catalog data** because billing codes don't map to tests.

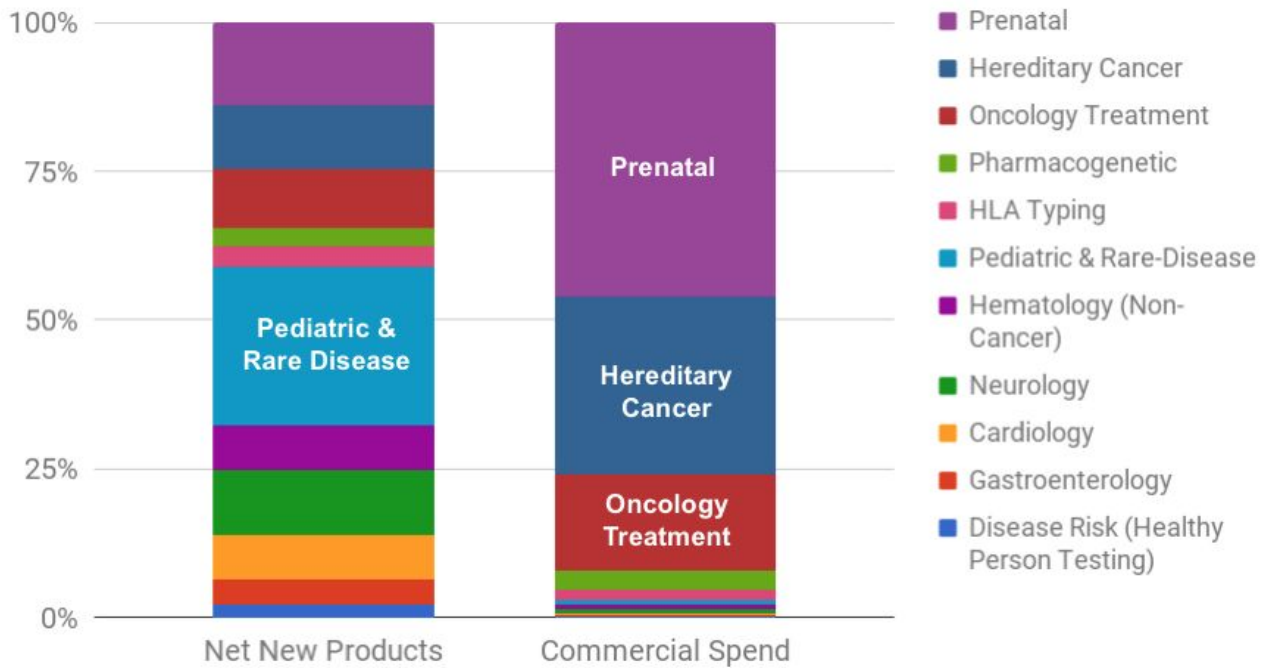
**At every step, some data is unavailable, some is lost, and the rest is transformed**



Concert's data and technology can bring transparency to this market. Leveraging the market-wide catalog of testing options and collaborative relationships with health plans, Concert matches claims (even complex claims for multi-gene panels) to specific tests, categories and domains in its database.

## A. Overview of Genetic Testing Reimbursement

Drawing on a comprehensive catalog of tests in the market and commercial health plan claims data from tens of millions of commercially insured members, the analysis below shows the distribution of commercial payments across high-level clinical domains (shown alongside the distribution of new tests released, from page 8).



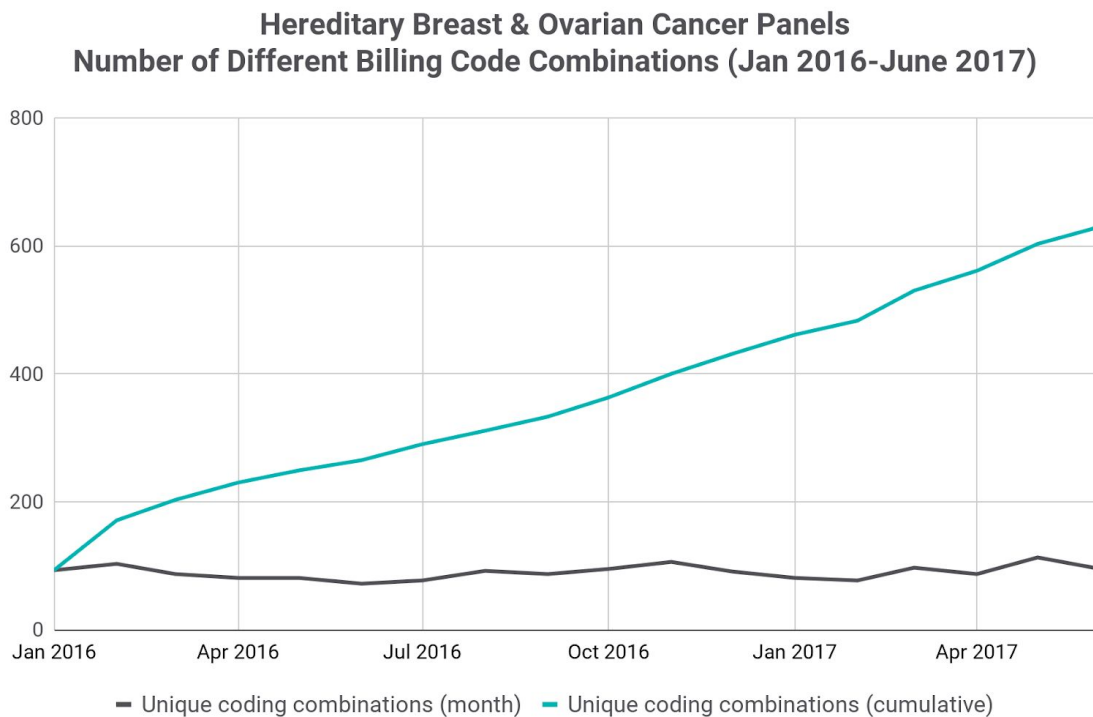
Interestingly, the test types being launched into the market do not correlate with the tests being paid for by commercial health plans. Prenatal, hereditary cancer, and oncology treatment test domains represent roughly 90% of all commercial payments

## B. Examples of Coding Challenges

Coding in genetic testing is challenging, and the complexity is a result of the rapid pace of innovation in the market and the trend towards utilization of multi-gene panels. The CPT® code system for molecular diagnostics does not have a one-to-one relationship between a test and a code. The current system leaves grey area in which there are multiple ways to code multi-gene panels. This introduces complexity on both sides of the billing-reimbursement equation: how to appropriately code a claim as the lab/provider and how to adjudicate and pay a claim as the health plan.

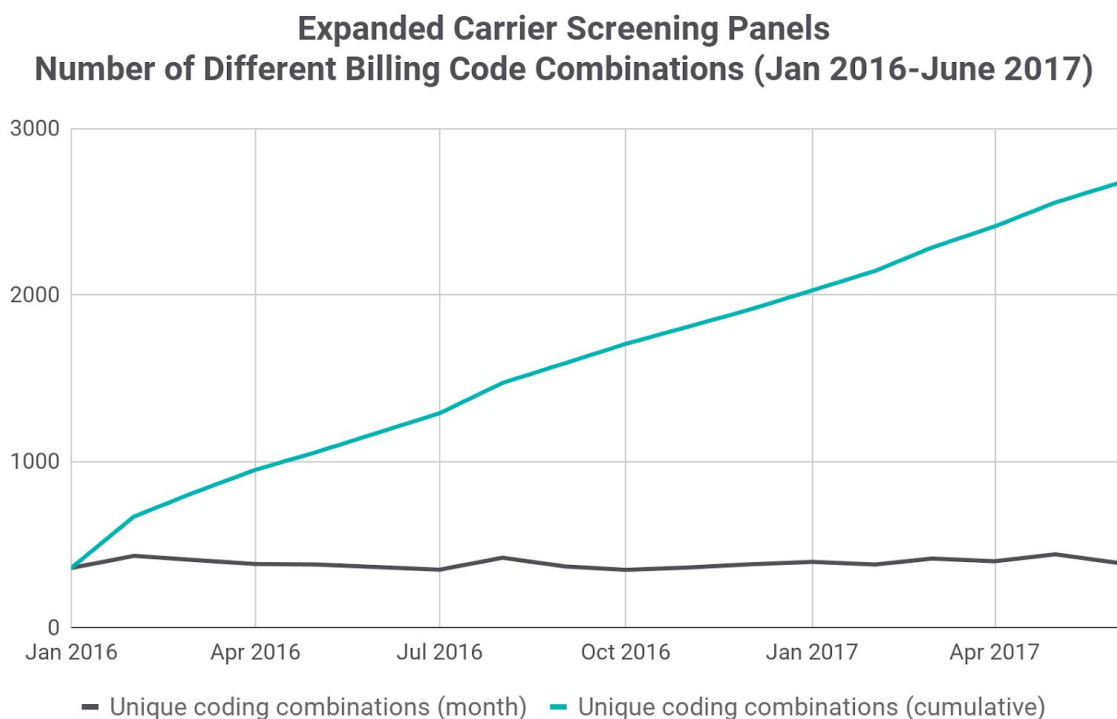
While these challenges are generally recognized by stakeholders across the market, many aren't aware of the extent to which they manifest and compound. The following charts illuminate this.

The first focuses on HBOC panels. A previous chart demonstrated the growing number and variety of panel tests targeting 3+ genes including *BRCA1/2*. Many of those are HBOC panels, and because of their varied gene combinations, they are billed with different code combinations. The chart below demonstrates just how much variation exists.



In any given month (the grey line), 70-100 different coding combinations were used to bill these tests. However, from month to month the combinations changed such that the *cumulative* number of different combinations grew in a linear fashion, ultimately exceeding 600 different combinations during the 18-month period shown.

In a second example, Concert analyzed expanded carrier screening panels. As these tests have gained support among the clinical community, the number of code combinations in claims data has steadily risen.



In any given month, expanded carrier screening panels are billed 350-450 different ways. And during an 18-month period, these panels were billed in more than 2,500 unique combinations.

The growing adoption of panel testing is translating into growing complexity in the coding and billing of tests. Given that there's no universal crosswalk between tests, codes, and payment policies, the influx of these claims is causing significant reimbursement challenges.

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### Key Takeaway:

The majority of reimbursement spend is in prenatal, hereditary cancer and oncology treatment, including many panel tests. Coding of these panels varies widely, challenging health plans' administrative systems.

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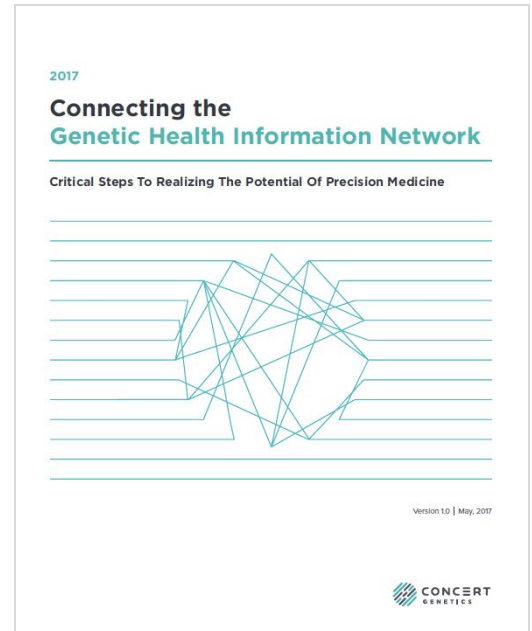


## 6. SOLUTIONS & OTHER RESOURCES

### A. Connecting the Genetic Health Information Network

We believe most, if not all, of the challenges in the genetic testing market stem from limitations in data and digital infrastructure. From the lack of clinical utility evidence on most tests, to the challenges clinicians face in carrying out testing, to reimbursement issues, all can be markedly improved with better connectivity, data and digital tools. We believe the solution to these issues is a more connected Genetic Health Information Network.

Readers who would like to learn more can see the document *Connecting the Genetic Health Information Network* (May 2017, Concert Genetics), which outlines key opportunities, near-term goals, and a long term vision for how to realize the promise of precision medicine. To access the paper, visit [concertgenetics.com/network](http://concertgenetics.com/network) or contact [connect@concertgenetics.com](mailto:connect@concertgenetics.com).



### B. The Concert Platform

For readers who would like to learn more about Concert's solutions, the company website has in-depth information. In particular, the Concert Platform is a proven software tool to achieve many of the goals outlined in this paper.

To learn more about the Concert Platform, visit [www.concertgenetics.com/platform](http://www.concertgenetics.com/platform) or contact [ntazik@concertgenetics.com](mailto:ntazik@concertgenetics.com).

CONCERT GENETICS [www.concertgenetics.com](http://www.concertgenetics.com)

**CONCERT PLATFORM** Power precision medicine with a platform that supports genetic testing from end to end.

The Concert Platform integrates test selection, results retrieval and the insurance workflow in one intuitive system.

**Every Lab, One Platform**

- ✓ Order tests from more than 300 labs in a single user-friendly interface
- ✓ Reduce costs by steering orders to contracted labs
- ✓ Save time by digitally populating requisition forms
- ✓ Track and receive digital results

**Financial Stewardship**

- ✓ Avoid surprises by viewing insurance coverage criteria on a test-specific basis
- ✓ Simplify prior authorization with tracking and support tools
- ✓ Minimize payment denials and unnecessary downstream re-work

Trusted by thousands of genetics experts nationwide, the Concert Platform has a proven track record at leading hospitals and academic medical centers.