Transforming the management of genetic testing

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E-book introduction

New technologies don’t wait to emerge until the policies for handling them are in place. That’s certainly the case with genetic testing. It’s been coming over the horizon for years, but healthcare has been caught unprepared by the dramatic growth in the adoption of testing and the variety of tests. Providers and health plans are scrambling to determine the validity and utility of tests, while dealing with an inadequate system for coding and management.

If genetic testing is to fulfill its potential to improve healthcare, it must be conducted through a scientific, evidence-based system that recommends test use in diagnosing, treating, managing, and monitoring disease while ensuring the right test for the right member at the right time determines the right care to deliver. This system also gives providers, labs, and health plans greater transparency into ordering and reporting while controlling costs and preventing waste, fraud, and abuse.

Through three chapters, this e-book will explore the inadequacies of the current genetic test management system and how a new, tailored genetic testing approach can unlock this exciting technology's full potential to fulfill its promise. While each chapter is a solid standalone read, when assembled, they form a thorough perspective. In the first chapter, we will explore the promise and problems of genetic testing.

Chapter 1
The promise and problems of genetic testing

Introduction

Genetic testing is one of the most promising developments in the history of medicine. The ability to identify the genetic causes of conditions and diseases, diagnose those conditions, and design treatments for them is transforming healthcare. Used correctly and managed adequately, genetic testing will lead to a healthier population served by a better-informed and more efficient healthcare industry.

And the enthusiasm for genetic testing is not limited to researchers and clinicians. Patients have embraced testing through their physicians and with at-home kits like 23andMe. Incorporating all the tests and resulting data into a comprehensive and effective healthcare program is a challenge for patients, providers, and insurance plans, but one that must be successfully met if we are to realize the full benefits of genetic testing.

Growth of testing

Since the human genome was mapped 20 years ago, genetic testing volume has dramatically accelerated and shows no signs of slowing.

More than 175,000[1] genetic tests are on the clinical market today, and 10[2] new tests are introduced daily. This is partly driven by the declining cost of gene sequencing, which fell from $1 million in 2007 to $600 in 2023.[3]

However, while the cost of individual genetic tests has decreased, overall spend on genetic testing has exploded. After analyzing the utilization and spend of genetic tests among Avalon clients across all books of business, we found that genetic tests comprised 10% of all lab tests in 2022, with an average spend of $779 per healthcare plan member per year. That was a 15% increase in utilization and an 11% increase in spending YOY.

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The genetic testing market costs $5.2 billion to $14.8 billion in the United States, depending on the source.[4],[5],[6] Data from the Centers for Medicare and Medicaid Services (CMS) show a significant increase in its compounded annual growth rate for genetic testing from 2015 to 2021, with it accounting for 20.4% of CMS' total lab spending in 2021.[7],[8]

Medicare fee-for-service data

Year	Total Lab Spending	Genetic Test Spending	% Genetic Test Spend	CAGR Genetic
2015	$6.69 B	$ 289 MM	**4.2%**	
2016	$6.77 B	$ 393 MM	**5.8%**	36.0%
2017	$7.13 B	$ 473 MM	**6.6%**	27.9%
2018	$7.59 B	$ 969 MM	**12.8%**	49.7%
2019	$7.68 B	$1.58 B	**17.7%**	47.3%
2020	$8.00 B	$1.20 B	**15.0%**	-24.0%
2021	$9.30 B	$1.90 B	**20.4%**	58.3%

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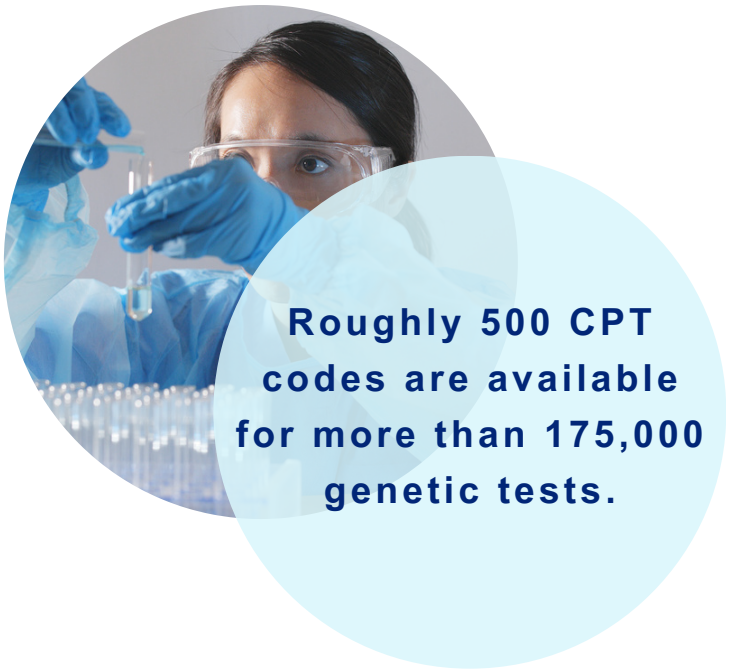
Insufficient CPT coding

Genetic tests are being created faster than new codes can be assigned to identify them. As a result, a wide range of genetic testing is lumped under a few nondescript Current Procedural Terminology (CPT) codes. While some genetic tests can be ordered using particular codes that apply only to a single gene or analyte, many other tests use a non-specific code that moves through prior authorization for approval. For example, there are more than 40,000 different tests with CPT 81479. In all, only about 500 CPT codes are available for more than 175,000 tests.



This creates problems and confusion for health plans, which must ensure regulatory compliance while keeping costs under control through prior approval and policies. The wide variation in cost for multiple tests under a single CPT code is also challenging.

A new coding system is required, which goes into greater specificity than CPT codes and allows for the identification and tracking of individual tests to make it easy for all parties involved to know what is ordered and why.

Order and provider confusion

The existing system for managing lab tests has been overwhelmed by the explosion in genetic testing. It's an unmanageable situation for providers and labs trying to put the enormous good of gene testing to work on behalf of patients. Most clinicians do not have the training or expertise to judge the validity and efficacy of genetic tests – and the challenge is only getting more complex. Consider these factors:

- The constantly growing body of knowledge of genetic medicine and the resulting test proliferation add to the complexity
- More complex panel combinations can increase waste
- Genetic lab orders are prone to being misunderstood or misordered
- There is a lot of variation and uncertainty about who provides test counseling to a patient or physician

Waste and errors caused by order and provider confusion can result in improper and missed diagnoses, unnecessary spending, and possible delays in treatment.

The health plan dilemma

The explosion in genetic testing has put health plans in the unenviable position of trying to determine the clinical validity and efficacy of hundreds of thousands of tests. The stakes are high. Needed tests that are performed correctly and whose results are used to inform treatment can save

lives. Unnecessary or poorly performed tests are a waste of money and resources and can potentially harm the patient.

Like clinicians, insurers do not have the resources or expertise to evaluate and validate the flood of tests, though controlling spend and serving members require them to make these difficult decisions. Health plans need a science-oriented partner that can evaluate the evidence and quality of new tests to establish their analytical and clinical validity and utility.

Drawing parallels from pharmaceuticals

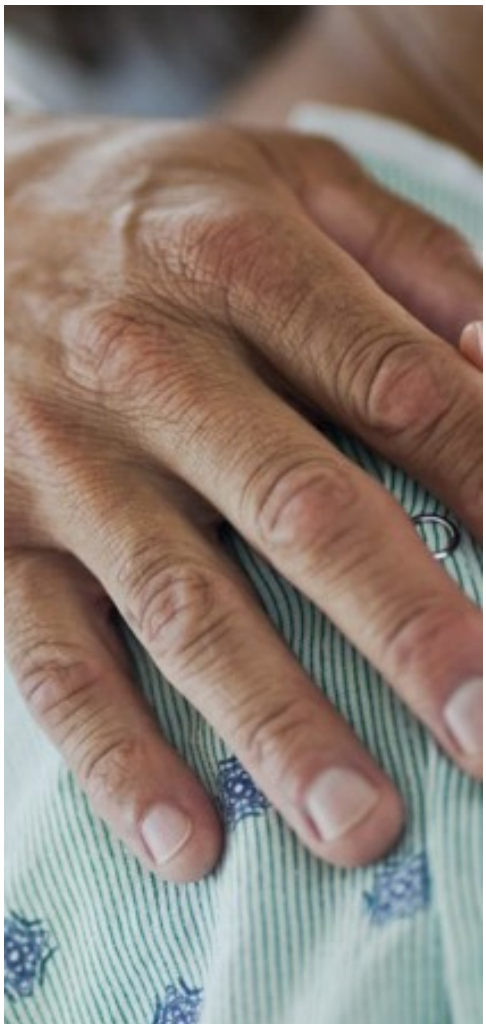
Pharmacy Benefit Management		Lab Benefit Management
Drugs are reviewed for efficacy and safety by the FDA before market approval.	REGULATION	Precision Medicine Tests do not require any review of quality data before being released to the market.
Each molecular entity has a unique identifier (NDC) that classifies the drugs, formulation, and manufacturer.	CODING	CPT codes are used to group tests based on broad indications. There is no national standard.
Clinical and cost benefit data submitted by the manufacturer is evaluated by multiple committees to determine the drug's place on a formulary and relevant utilization management criteria.	COVERAGE	Previews of quality data for Precision Medicine Tests are performed inconsistently or only across a small subset of tests.
Drugs are reviewed for efficacy and safety by the FDA before market approval.	REIMBURSEMENT	Fee schedules exist at a CPT level, not test level, leading to the missed opportunity to negotiate rates using a test formulary.

Vulnerability to fraud, waste, and abuse

Like many fast-growing fields with insufficient controls and oversight, genetic testing has been accompanied by high fraud, waste, and abuse rates. Measures to regulate and oversee the field have been patchwork and need help to keep up with the growth.

While Theranos is an extreme example of outright fraud, misuse also includes overordering. Up to 30% of laboratory testing might be unnecessary.[9,10] Providers might overorder tests for fear of missing something, or by practicing "defensive medicine," they can order an unnecessarily large battery of tests to reduce the threat of malpractice liability. This raises the risk of false-positive and false-negative results and subsequent harm to the patient. Failure to order the proper test affects about 50% of lab orders, which can lead to an incorrect or missed diagnosis.[11]

Independent validation of genetic tests and better management and control systems can reduce these problems.



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Summary

Genetic testing is not slowing down to allow test management systems to catch up – nor should it. Rather, it's incumbent on healthcare to quickly adopt new policies and procedures to ensure that genetic tests can be used for the maximum benefit while minimizing misuse and controlling spend.

The second chapter of this e-book will discuss specific solutions for managing genetic testing that provide greater transparency and control than current methods.

To learn more about how the Precision Genetic Test Management solution from Optum and Avalon can help you better manage your genetic test program, please visit www.avalonhcs.com **or contact us at** avalon-info@avalonhcs.com.

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