

# GENETIC TESTING

FRAUD, WASTE, & ABUSE



## WHITE PAPER

JULY, 2020



**HEALTHCARE FRAUD  
PREVENTION PARTNERSHIP**

# The Healthcare Fraud Prevention Partnership would like to thank participating Partners for their contributions.

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# EXECUTIVE SUMMARY

Genetic testing is increasingly being used in clinical and public health practices to assist disease diagnosis, predict disease risk, and guide patient care. The completion of the Human Genome Project (HGP) in 2003 led to the development of thousands of tests that can now be used to detect a chromosomal abnormality and the possibility of inherited illness or disease. In the past decade, the number and complexity of genetic tests has rapidly grown, with these tests, collectively, costing the healthcare industry billions of dollars per year.

Today, the increased public awareness and eased modality of genetic testing make it vulnerable to potential fraud, waste, and abuse, which increases healthcare spending and negatively affects the integrity and quality of the healthcare system. Individuals may experience anxiety and frustration or receive medical interventions or services that are incorrect or inappropriate as a result of the potential fraud, waste, and abuse in genetic testing. In this context, several aspects of the healthcare system are particularly vulnerable, such as payer oversight, sufficient evidence-based support, and professional training.

Over the past few years, fraud, waste, and abuse in genetic testing has emerged across the country. As a response, this paper from the Healthcare Fraud Prevention Partnership (HFPP), a public-private partnership of healthcare payers and allied organizations, aims to clarify the clinical genetic testing field and provide a review of current guidance on the appropriate use of genetic testing services. It also identifies several systematic challenges that HFPP Partners believe make genetic testing vulnerable to potential fraud, waste, and abuse cases, including:

- The need for stronger or updated controls and standards for a rapidly expanding clinical domain
- The complexity and continuing evolution of genetic testing
- The increased marketing of tests and individual demand for genetic testing

Within this context, this paper describes schemes seen in this field by HFPP Partners, presents current strategies for stopping genetic testing fraud, waste, and abuse, including pre-payment controls, post-payment audits, and provider engagement, and outlines actions that may be valuable to consider and apply for future interventions.

# INTRODUCTIONS & OBJECTIVES

The Healthcare Fraud Prevention Partnership (HFPP) is a voluntary, public-private partnership between the Federal Government, state and local government agencies, law enforcement, private health insurance plans, employer organizations, and anti-fraud organizations that seeks to identify and reduce fraud, waste, and abuse across the healthcare sector.<sup>1</sup> To advance this effort, HFPP Partners regularly collaborate, share information and data, and conduct studies using a unique cross-payer dataset. Additionally, the HFPP's broad membership provides a platform to discuss emerging healthcare issues.

## Objectives

This paper provides an overview of clinical genetic testing services and examines recent fraud, waste, and abuse schemes in the field of genetic testing, an area of growing concern that negatively affects health organizations' financial stability and patient outcomes. Genetic tests sold directly to consumers are not the focus of this paper. The following sections set out to answer these questions:

- What is the current state of the clinical genetic testing industry?
- What is the current guidance from professional societies, experts, and accrediting bodies regarding the appropriate use of clinical genetic testing?
- What are the susceptibilities that make clinical genetic tests targets for fraud, waste, and abuse?
- What are some examples of major fraud, waste, and abuse schemes in clinical genetic testing?
- What are good practices for preventing and detecting fraud, waste, and abuse in clinical genetic testing?

## What is Genetic Testing?

Genetic testing is the use of laboratory procedures to analyze genes, chromosomes, or gene products (proteins and metabolites) that provide specific information about inherited variations in genes or chromosomes of an individual or their progeny.<sup>2</sup> There are three categories of genetic testing—molecular, cytogenetic, and biochemical—used to assess abnormalities in DNA sequences, chromosome structure, and protein function, respectively. Genetic tests are performed using samples such as those collected from blood, hair, skin, amniotic fluid, or tissue from the inside of the cheek. These tests can assist in identifying changes in genes, chromosomes, or proteins of interest.

The test results provide valuable insights for disease prevention, diagnosis, and treatment options. Several major types of genetic testing performed in clinical settings are listed below.<sup>2-4</sup>

### Diagnostic Testing



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This type of testing can be used to identify or rule out a specific genetic or chromosomal abnormality. If the symptoms of a disease are possibly caused by genetic alterations, diagnostic testing can be used to confirm certain genetic disorders such as cystic fibrosis or Huntington's disease.

### Pre-symptomatic and Predictive Testing



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When there exists a family history of a genetic condition, receiving genetic testing before developing symptoms may indicate whether an individual has an increased risk for developing a particular disease. Tests of this type are often used in cancer genetic testing (CGx) to assess an asymptomatic person's risk for developing hereditary cancer such as breast or colorectal cancer.

### Newborn Screening



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This is the most common type of genetic testing. All states in the U.S. require that newborns be tested for certain genetic and metabolic disorders that cause specific diseases such as sickle cell disease, or phenylketonuria (PKU). Immediate care or treatment can be given when such a disorder is detected.

### Prenatal Testing



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Genetic testing during pregnancy can detect some types of abnormalities in the fetal genes. Genetic disorders, such as Down syndrome and other chromosomal abnormalities, are often screened using noninvasive procedures.

### Carrier Screening Testing



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This test is often performed for people with a family history of a genetic disorder or at an increased risk for a specific disorder who are pregnant or planning a pregnancy. The results provide information about the chances of having a child with a genetic disorder such as sickle cell disease or Tay-Sachs disease.

### Preimplantation Testing



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This is a screening test that identifies the presence of aneuploidy (abnormal number of chromosomes) in a developing embryo. This test is considered when one or both parents have a known genetic abnormality to determine if an embryo carries a genetic defect such as Marfan syndrome or cystic fibrosis.

### Pharmacogenomic Testing



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A pharmacogenomic test (PGx) identifies variations in an individual's genetic makeup that affect how a person may respond to certain medications. It assists in determining the most appropriate choice of medication and/or the dosage of the drug therapy. PGx may be performed in specific clinical circumstances for a particular category of drug being prescribed for a particular person.

Genetic testing has evolved significantly during the past several decades, especially after the completion in 2003 of the Human Genome Project (HGP), which led to significant growth in the number of available genetic tests. Genetic tests vary by the genes being analyzed such as single gene testing, multiple-gene testing, or whole genome/exome sequencing (WGS/WES) testing. In recent years, tests have been expanded to include not only molecular biomarkers but also patient demographics and clinical characteristics to generate information regarding disease diagnosis, prognosis, or prediction such as Multianalyte Assays with Algorithmic Analyses (MAAA) testing for breast cancer. Genetic tests also vary by the techniques used, for example, sequencing, polymerase chain reaction, or microarrays.

Since 1977, Sanger sequencing<sup>5</sup> has been the gold standard for clinical diagnostic and predictive testing, however, its high cost and inefficiency for larger-scale efforts (e.g., whole genome testing) have called for a paradigm shift. In 2012, Next Generation Sequencing (NGS) technology, simultaneously screening for thousands of genes to detect multiple variant types across targeted areas of the genome, began to be applied in clinical diagnosis, offering a cost and time-effective approach for capturing genetic makeup.<sup>6</sup>

Genetic testing increasingly influences healthcare delivery and, when used appropriately, may lead to substantial improvement in individual and population health outcomes. The appropriate use of genetic testing offers great opportunities to facilitate precision medicine and personalized care. However, genetic testing that is not medically necessary may negatively affect and harm people. Individuals receiving covered services by fraudulent means may be denied future coverage, as many genetic tests for inherited diseases are once-in-a-lifetime payments. Concerns also exist regarding incorrect test results or result interpretations leading to inappropriate decisions for treatment(s).

# GENETIC TESTING INDUSTRY IN THE CONTEXT OF FRAUD

## Clinical Labs Performing Genetic Tests

According to the findings of a recent federal investigation on genetic testing fraud, over 840 clinical laboratories provide genetic testing in 45 states of the United States, with California having the largest number of laboratories (n=99), followed by Texas (n=92), Tennessee (n=77), Florida (n=53), and New York (n=44).<sup>7</sup> To understand the complexity of genetic testing as experienced by members of the HFPP, datasets from the HFPP data warehouse were used. The HFPP dataset, at the time of analysis, contains claims from Partners representing more than 30% of covered lives in the United States, including claims from the Centers for Medicare & Medicaid Services (CMS), state Medicaid agencies, and private payers. A total of 221 distinct procedure codes for genetic testing were identified for the year 2018. The number of services was the highest for single gene procedure codes accounting for 62.0% of all genetic testing services, followed by MAAA, (18.4%), noninvasive prenatal testing (NIPT, 2.6%), multiple-gene testing (2.0%), and whole-genome sequencing (WGS, 1.8%).

## Cost Estimate of Genetic Testing

The actual costs of genetic testing can range from under \$100 to several thousand dollars depending on the type and the complexity of the test, as well as payer policy. Federal investigators reported that there were approximately ten genetic test-related procedure codes in 2016, which accounted for \$1.3 billion dollars in Medicare spending. By 2019, approximately 250 procedure codes existed, for a total cost estimate of \$7 billion dollars<sup>7</sup> – a 438% increase in genetic test spending in only three years. The comparatively high cost of genetic testing can also be seen in context. A recent study from a commercial payer claims database reported genetic testing expenditure by clinical domain: prenatal genetic tests (30 - 40%), hereditary cancer tests (30%), and oncology diagnostics and treatment (10%).<sup>8</sup> Using the HFPP data warehouse, numbers for the spending on genetic testing services in 2018 show that procedure codes for single gene tests made up 50.2% of the overall spending, followed by MAAA at 36.4%, multiple-gene testing at 4.7%, NIPT at 3.9%, and WGS accounting for less than 1%.

## Regulation of Clinical Laboratories for Genetic Testing Services

CMS regulates laboratories (except research laboratories) that perform testing on specimens obtained from humans, including genetic testing, through the Clinical Laboratory Improvement Amendments (CLIA) program. The CLIA regulation categorizes laboratory tests by their complexity: waived for tests of simple complexity and non-waived for tests of moderate and high complexity. Molecular genetic testing for hereditary diseases and conditions is high complexity testing, requiring that laboratories performing these tests must meet the CLIA standards for quality, accuracy, and reliability of testing, as well as personnel requirements for high-complexity testing.<sup>9</sup> The states of New York and Washington have adopted laws related to laboratory requirements that are equal to or more stringent than CLIA requirements. Therefore, laboratories in these two states have an either full or partial exemption from CLIA's requirement.

The U.S. Food & Drug Administration (FDA) has the authority to regulate genetic tests that are intended for use in the diagnosis of disease or other conditions, including a determination of the state of health, in order to cure, mitigate, treat, or prevent disease or its sequelae.<sup>10</sup> A test may come to market as a commercial “testing kit” and be sold to laboratories who perform the test. These test kit manufacturers are expected to comply with applicable provisions of the Federal Food, Drug, and Cosmetic Act (FDCA),



including clearance or approval from the FDA before selling many of their products, depending on their risk. More commonly, genetic tests are available as laboratory-developed tests (LDTs), where the tests are developed and performed within a single certified laboratory. These tests are offered under FDA's policy of enforcement discretion for LDTs, meaning that the FDA does not evaluate the analytical and clinical validity of many LDTs being used.<sup>11</sup>

In addition, all laboratories, including those that perform genetic testing, need to pass state requirements to be licensed and perform health-related testing. The accuracy and usefulness of genetic and genomic tests can be evaluated and regulated by the following three factors: analytic validity, clinical validity, and clinical utility.

Several state agencies (e.g., California, New York) have adopted more stringent criteria in addition to the CLIA requirements. In California, all laboratories need to be licensed by the state and meet federal CLIA requirements, and tests cannot be offered to individuals without a physician's order. Also, California state law addresses genetic testing to ensure that test results are accurate and valid and offered only with sufficient medical oversight to avoid unnecessary harm.<sup>12</sup>

In addition to government agencies, many other groups assist in oversight, including payers, professional societies and industry organizations, private-sector accreditation bodies, and individual advocacy groups. This oversight has been achieved by developing evidence-based clinical and laboratory practice guidelines, establishing standards, and accrediting clinical laboratories. Multiple clinical guidelines have recommended an evidence-based practice on when, what, and whom to test. For example, professional associations such as the National Comprehensive Cancer Network (NCCN) and American Society of Clinical Oncology (ASCO) have defined the role of genetic testing in the diagnosis and management of individuals diagnosed with or at risk for cancer, issuing evidence-based recommendations that are revised on a regular basis.

## Coverage Determination Process

HFPP Partners have noted that payers have been deliberate in establishing their own program policies of coverage determination for genetic testing. Insurance organizations develop their own clinical policy on genetic testing for use if there is no existing coverage available for the requested genetic test. The clinical policies are determined based on a review of the following: government agency/program approval status; clinical practice guidelines of leading health professional organizations; generally accepted standards of medical practice; peer-reviewed medical literature; positions of leading national health professional organizations; views of physicians practicing in relevant clinical areas affected by this clinical policy; and other available clinical information. The coverage for specific tests is usually evaluated for medical necessity on a case-by-case basis.

## Analytic Validity

Assesses the accuracy of a genetic test in detecting the presence or absence of a particular gene or genetic variation, such as a DNA sequence variant, chromosomal deletion, or biochemical indicator.

## Clinical Validity

Refers to the extent to which an analytically valid test result can predict the presence, absence, or risk of a specific disease, drug response, or other outcomes. It is commonly quantified by sensitivity, specificity, positive predictive value, and negative predictive value of a specific test. The FDA requires evidence of clinical validity for authorization of genetic tests.

## Clinical Utility

Measures whether the test can provide information about the diagnosis, treatment, management, or prevention of a disease that will lead to an improved health outcome. Consumers, health providers, and health insurance companies are often the ones who determine the clinical utility of a genetic test. This is the most challenging criterion to demonstrate, often requiring long-term follow-up and/or clinical trials.

# EXISTING GUIDANCE REGARDING THE APPROPRIATE USE OF GENETIC TESTING

## Appropriate Use of Genetic Testing

Defining and implementing genetic testing protocols that have a high likelihood of providing benefit is an important and challenging health-policy task. Figure 1 below, at a high-level, illustrates the appropriate process for conducting clinical genetic testing.

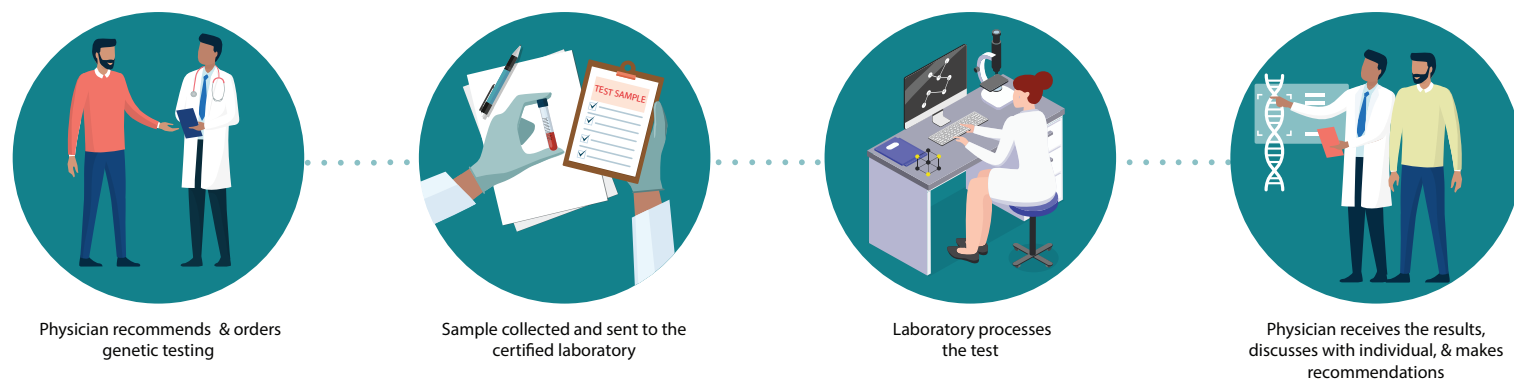


Figure 1: Guidance for Conducting Genetic Testing

There are several important considerations from scientific organizations, HFPP Partner interviews, and accrediting bodies during this process to ensure the appropriate use of genetic testing:

- Provider Qualification for Ordering Genetic Testing:** Once a person agrees to proceed with genetic testing, a qualified healthcare professional, such as medical geneticists, primary care physicians, oncologists, obstetrician-gynecologists, pediatricians, or nurse practitioners, can order the test. Typically, the provider who ordered a genetic test(s) or referred the patient for one should have specific knowledge of the patient and the test, and an established therapeutic relationship with the individual before ordering the test.
- Medical Necessity Determination for Genetic Testing:** Medical necessity criteria vary for specific genetic tests. The individual's medical record must contain documentation that justifies the medical necessity for a particular genetic testing service: medical history, physical examination, and results of pertinent diagnostic tests or procedures. In general, genetic testing is considered medically necessary when the following criteria are met:
  - The individual displays clinical features, or is at direct risk of inheriting the mutation in question due to its identification in a family member (pre-symptomatic)
  - The results of genetic testing are being used to inform clinical interventions, detect diseases when treatments are available, manage symptoms, and/or slow the progression of an established disease
  - A definitive diagnosis remains uncertain after completion of traditional diagnostic studies, physical examination, pedigree analysis, and genetic counseling
  - Disease-specific criteria are met

- **The Test is Performed in a Certified Laboratory:** Laboratories performing molecular genetic testing for heritable diseases and conditions must meet the CLIA requirements. Certified laboratories are required to develop and follow the policies and procedures for patient preparation, specimen collection, handling, specimen referrals, and test requests. In addition, laboratories that perform non-waived testing must have a qualified clinical consultant to assist laboratory clients with the appropriate ordering of tests to meet clinical expectations.
- **Pre- and Post-Genetic Counseling:** National evidence-based guidelines, expert opinions, and accrediting bodies have recommended providing pre- and post-test genetic counseling services to assist individuals in complex clinical decision-making.<sup>13-16</sup> Before receiving a genetic test, individuals need to understand the testing procedure, the benefits and limitations of the test, and the possible consequences of the test results. For example, the NCCN guidelines suggest that genetic counseling is a critical component of the disease risk assessment process. The pre-test counseling should discuss a wide range of topics, including why the test is being offered and how test results may impact disease management, benefits and limitations of the test, explanation of possible test results for the individual and family, technical aspects and accuracy of the test, and cost considerations.<sup>14</sup> Also, results from genetic testing can be a source of anxiety and concern without proper post-test genetic counseling or explanation from healthcare professionals in the context of the individual's personal and/or family history. Per the NCCN guidelines, post-test counseling should include results disclosure, discussion of the significant results, a discussion of the effect on psychosocial aspects and on the disease management of the individual, and additional resources for relevant information and follow-up care.<sup>14</sup>

### How Frequently Should Genetic Testing Be Performed?

DNA-based genetic testing of inherited gene variants does not change over time. This would normally mean that an individual would not need to receive a specific genetic test more than once. However, HFPP Partners have described the following situations in which individuals may receive the same genetic test multiple times:

- **Changes in the Testing Panel:** Technological advancement has made it possible to measure multiple genes and perform WGS/WES tests, which may offer new and valuable information for patient care. Also, advances in technology can warrant re-testing of previously under-read genes or genomic regions.
- **Advancement of Knowledge:** Advances in the knowledge of mutation characteristics for a particular disorder, or new information to substantiate the clinical validity of previously untested genetic variants may recommend repeat testing.
- **Evolution of Results Interpretation:** New data interpretation that warrants repetition and/or revised reporting of a test.

## CONCERN ABOUT THE CLINICAL VALIDITY AND UTILITY OF SOME TESTS

The FDA has raised concerns about the clinical validity of certain PGx marketed to predict the best medications to treat certain conditions, such as depression and acid reflux, based on an individual's genetics. While the clinical validity of many PGx are clear based on FDA's approved drug labels and guidance and peer-reviewed expert guidelines, some tests on the market but not reviewed by the FDA may claim to predict a person's responses to medications without scientific or clinical evidence.<sup>17-19</sup>

Clinical validity can be limited by two factors: genetic heterogeneity and incomplete penetrance.<sup>20</sup> Genetic heterogeneity describes situations in which mutations at two or more locations of the same gene or different genes are associated with the same phenotype. The current technology often cannot identify all instances of disease-related variants, thus reducing a test's clinical validity. Penetrance refers to the likelihood that a clinical condition will develop when a particular genotype is present. When penetrance is incomplete, a test's clinical validity is reduced. For example, testing for the C282Y mutation in the Hemochromatosis gene (HFE) is of limited clinical validity since a minority of individuals with the C282Y homozygous genotype will develop hemochromatosis.<sup>21</sup>

In addition, the clinical utility of a genetic test depends on the available management options. For instance, testing for conditions such as Huntington's disease that have limited or no treatment options may have little clinical value compared to tests for other conditions, even though the test itself may have high analytic and clinical validity.

## GENETIC TESTING VULNERABILITIES IN THE CONTEXT OF FRAUD, WASTE, AND ABUSE

As the use of genetic testing grows, understanding its vulnerabilities can help address the increasing number of fraudulent, wasteful, and abusive instances observed. There are substantial challenges described below, as identified by the HFPP Partners, that make clinical genetic testing vulnerable to fraud, waste, and abuse. Several of the vulnerabilities identified are not specific to genetic testing but are associated with laboratory testing more broadly, and are similar to the systemic challenges discussed in the HFPP's previous white paper on fraud and abuse with clinical laboratory services.<sup>22</sup> The vulnerabilities presented by genetic testing include:

- **The need for stronger or updated controls and standards for a rapidly expanding clinical domain.** HFPP Partners indicated that scientific and technological advances in genetic testing present certain challenges to existing frameworks of oversight. Existing regulations, billing standards, and claims edits may not apply to genetic testing or to telemedicine, which is commonly the method used by those perpetrating fraud or abuse schemes in genetic testing. For example, establishing billing standards can be a challenge with rapidly increasing test availability and complexity, such as with multi-gene panels and WGS/WES. HFPP Partners also believe that a few hundred codes are not sufficient to capture the tens of thousands of available tests. Little agreement exists on how these tests should be billed and what code should be used. Different laboratories may use different codes for a single genetic test. The lack of standardized billing guidance leads to variations in the amount of payment for the same genetic test.

- **The complexity and continuing evolution of genetic testing.** The increasing complexity of genetic test options require tremendous effort to evaluate their clinical validity and utility. Many HFPP Partners claimed that the new era of genomic medicine poses a challenge to the practice of medicine because of the insufficient education about genetic testing. The rapid development of genetic tests can make it difficult for providers and patients to stay abreast of the role of genetics in health and disease. Furthermore, most individuals do not adequately appreciate the complexity of interpreting probabilistic genetic information about an individual's risk, relative to that of the general population, for common disorders like breast cancer, heart disease, and diabetes.
- **The increased marketing of tests and individual demand for genetic testing.** Several HFPP Partners noted that the marketing of direct-to-consumer genetic tests increased patient demand for potentially unnecessary tests. There are misperceptions about the role of genetic testing within the general public, and people overestimate the role of genes and underestimate the role of nongenetic, environmental factors. In some instances, clinicians may feel obliged to accept an individual's request and order unnecessary or unwarranted genetic tests. In addition, the technology being employed is becoming more readily available, is less costly, and provides faster results.

These challenges are often combined to create opportunities for these fraudulent actors to exploit loopholes. The following section discusses specific fraud and abuse schemes that have been identified by HFPP Partners.

# FRAUD, WASTE, AND ABUSE IN GENETIC TESTING

Genetic testing is susceptible to the same fraudulent schemes found in other areas of laboratory services, thus many schemes presented in the previous HFPP white paper on clinical laboratory services<sup>22</sup> apply here. Furthermore, as noted by many HFPP Partners, the move from requiring a blood draw, performed by a trained professional at a laboratory or in a doctor's office, to the use of a simple cheek swab, which can easily be performed by an individual at home, is a significant factor in the susceptibility of genetic testing to fraud, waste, and abuse. The following section describes fraud, waste, and abuse schemes identified by HFPP Partners. The next section reviews preventive actions being taken, along with suggestions for additional actions. These examples can guide future discussions and interventions to help address the fraud, waste, and abuse in genetic testing services.

## Fraud, Waste, and Abuse Schemes

Major fraud, waste, and abuse schemes related to genetic testing seen by the HFPP Partners include kickbacks, manipulation of billing codes, and unnecessary genetic testing. Fraudulent schemes that investigators have reported so far show that older people are often the target population of potential fraudulent, wasteful, and abusive testing services. There are signs and concerns of suspicious activities starting to rise around the testing of pregnant women or parents of young children, although no clear fraudulent activity has been demonstrated to date.

### Kickback Telemedicine/Telemarketing Fraud Scheme

In September 2019, multiple law enforcement agencies, including the U.S. Department of Justice (DOJ), U.S. Department of Health and Human Services Office of the Inspector General (HHS OIG), and the Federal Bureau of Investigation (FBI), led and coordinated the investigation and prosecution of genetic testing-related fraud and abuse. One of the largest healthcare fraud schemes ever, involving, to date, charges against 35 individuals across the country, including physicians, marketers, and others responsible for \$2.1 billion in losses. The scheme involved illegal kickbacks that were paid to medical professionals working with fraudulent telemedicine companies in exchange for referrals. The tests were allegedly not provided, medically unnecessary, or ordered with little supervision or no established patient-physician relationship.<sup>23</sup>

According to HFPP Partners, those perpetrating fraud use marketers, telemarketers, or sales representatives, to solicit insured individuals for lab samples. At a high level, *Figure 2: Kickback Telemedicine/Telemarketing Fraud Scheme* illustrates the general attributes involved in the scheme experienced by HFPP Partners.

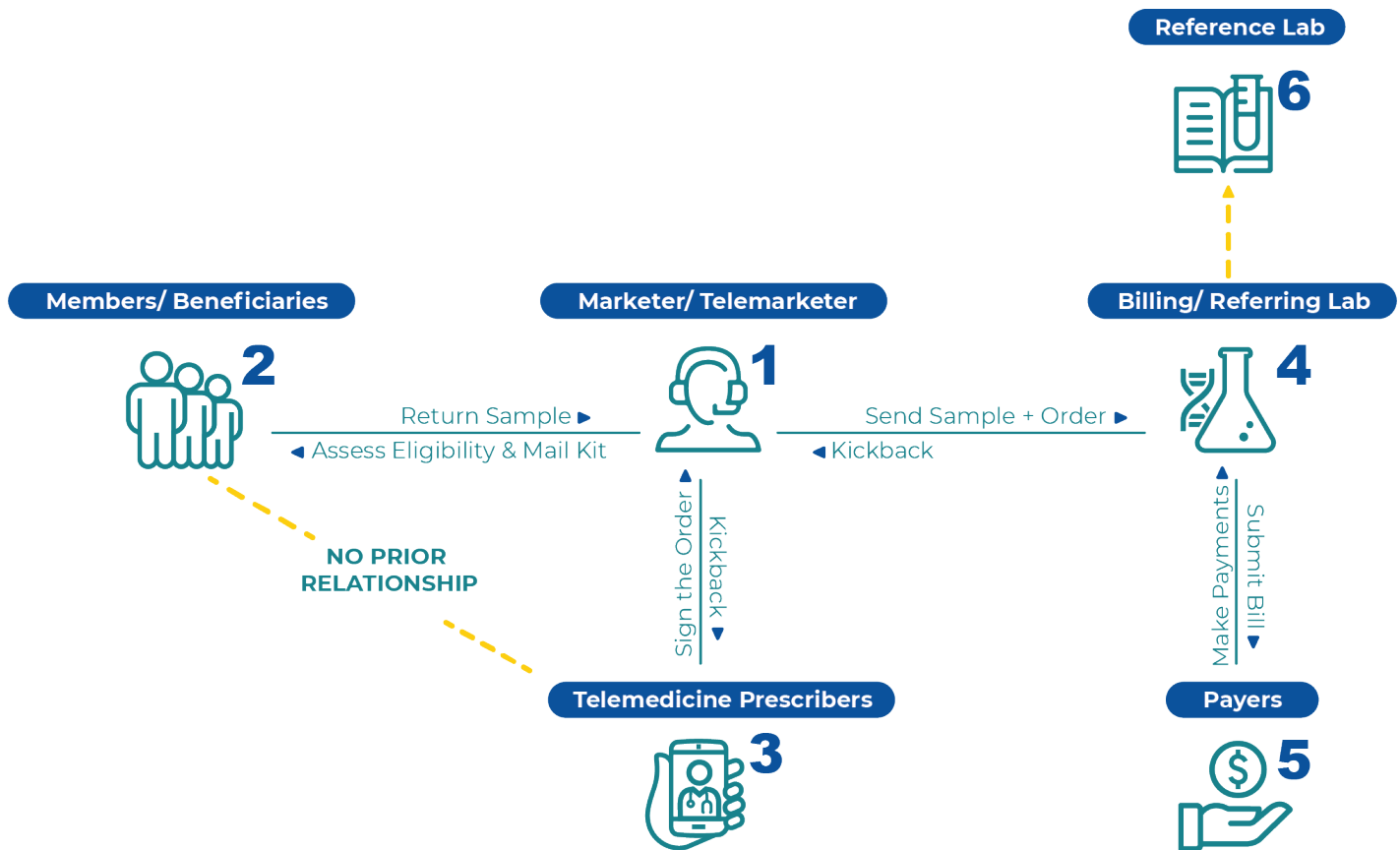


Figure 2: Kickback Telemedicine/Telemarketing Fraud Scheme

Individuals, most commonly seniors, are recruited through multiple avenues such as telemarketing calls, door-to-door visits, and social media advertisements. They are also approached at health fairs, church events, homeless shelters, senior centers, and home health agencies by those perpetrating the scams. The individuals are offered gift cards, free food, or other items as an enticement to participate. Testing is alleged to be free or offered at no charge. Some payers do not require a copayment for laboratory testing, while in other instances, bad actors waive or cap the costs for individuals, regardless of payer reimbursement.

As shown in *Figure 2: Kickback Telemedicine/Telemarketing Fraud Scheme*, for individuals who agree to genetic testing, a marketing/telemarketing company [1] verifies the person's [2] eligibility and health insurance coverage for genetic testing over the phone or through in-person screening. The marketing/telemarketing company then sends a cheek swab testing kit, usually by mail, for the individual to self-administer and return. The marketing/telemarketing company recruits telemedicine doctors [3] to sign orders for genetic tests often without reviewing each patient's medical history. The physician who "orders" these fraudulent tests frequently has no prior relationship with the patient.

Marketers/telemarketers send the sample together with the order to a laboratory [4] for genetic testing. The laboratory bills the individual's insurance [5] and makes illegal kickback payments to marketers and doctors for their roles in collecting the samples and signing the order. The laboratory may bill insurance without bothering to process and analyze the test. In some instances, the billing laboratory contracts with another laboratory [6] to perform the test.

There are potential negative consequences associated with this scheme. Individuals may experience medical identity theft resulting from giving out personal information that can be misused for fraudulent claims in the future. In addition, the investigation of this fraud scheme has revealed that some samples were not correctly obtained by the individual or properly stored during transportation, which may lead to incorrect test results for those tests that are analyzed. It is likely that the treating provider would be unaware whether the individual was previously tested. If that individual were to have hereditary cancer testing ordered by a treating provider in the future, the claim might be denied as most hereditary genetic testing is only allowed once-in-a-lifetime. At a critical time where the test is needed in earnest, the record may show such an analysis was already performed and is no longer an available benefit for that individual.

## Manipulation of Billing Codes

Fraudulent and abusive billing practices identified as particular areas of concern by the HFPP Partners are described below.

- **Billing for Services Not Rendered or Performed:** This occurs when a provider falsifies the bills and/or medical records to charge for services that are not rendered. One example is known as a "gang visit," where an improbable number of tests occurs in a single location on a single day, such as a nursing home, assisted living, health affairs, and other places where there is a concentrated number of insured individuals.
- **Unbundling of Claims:** "Unbundling" occurs when a clinical laboratory bills each test separately, rather than using an appropriate panel of bundled tests, to maximize reimbursement. For example, tests typically analyze multiple genes simultaneously on a single piece of equipment using NGS where laboratories often bill multiple molecular codes rather than a panel code.
- **Blanket Ordering:** Blanket ordering refers to indiscriminate ordering of a number of tests for patients without regard to their individual circumstances. One HFPP Partner mentioned a potentially fraudulent situation in which an obstetrician-gynecologist ordered a large number of genetic tests for pregnant women, when, in fact, the majority of these tests were medically unnecessary and should have been tailored to individual need.

## Medically Unnecessary Testing

Another area of significant concern identified by HFPP Partners is when specific genetic tests are performed on individuals without proper justification for the test, given the individual's circumstances. These tests may not be useful if the results do not provide information to direct care or guide treatment strategies. These unwarranted tests have little value for the individual due to limited impact on a patient's care. The results from these tests only provide information about risks for developing a particular disease, and individuals can take only minimal action on the information, particularly when



the results are presented to patients without proper interpretation or guidance by their treating physician or a healthcare provider.

The federal government alleged that GenomeDx submitted claims to Medicare between September 2015 and June 2017 for the Decipher prostate cancer test that were not medically reasonable and necessary, in violation of the False Claims Act. The Decipher test, a post-operative genetic test for prostate cancer patients, identifies men who may have a higher risk of the disease progression and may benefit from adjuvant radiation treatment after radical surgery for prostate cancer. According to the U.S. Attorney's Office, the patients for whom claims were submitted did not have risk factors that required the test.<sup>24</sup>

Another problem noted by HFPP Partners is the inclusion of extraneous tests in a laboratory developed panel. Some cancer susceptibility testing panels may include genes that have not been associated with hereditary breast or ovarian cancer and, in some cases, are not clinically actionable. However, testing with a targeted panel may be indicated as a cost-effective strategy when the individual's symptoms or family history meets testing criteria for more than one hereditary cancer syndrome.

# STRATEGIES FOR DETECTING AND PREVENTING FRAUD, WASTE, AND ABUSE

Fraud prevention and detection are crucial for reducing inappropriate expenditures attributed to healthcare services. While genetic testing fraud, waste, and abuse can be difficult to identify, there are several strategies for detection that are borne out across the industry. HHS-OIG, FBI, CMS, and the Senior Medicare Patrol may receive complaints from members, providers, and health plans via hotline or email about unusual billings of potential fraud and abuse cases. Further, private payers rely on Special Investigation Units (SIUs) to detect, prevent, and correct fraudulent instances through data analysis, healthcare claims auditing, and identification of suspicious billing activities. Key investigative strategies target the analysis of claims, which are regularly monitored and compared for submission inconsistencies, irregularities, outliers at different levels (e.g., provider, billing code, and laboratories), and other indicators of potential fraud, waste, and abuse. Evidence gathered from a range of anti-fraud activities identified characteristics and scenarios that are strongly associated with fraud.

## Indicators of Potential Fraud, Waste, and Abuse

Prevention and detection strategies consistently point to the behavior and scenarios listed below as red flags. These flags alone are not direct evidence of fraudulent and abusive cases, but they are strong indicators that additional review or investigation may be necessary.

### Red Flags from Data Analysis

- The ordering/referring provider is not listed as the attending physician for the individual or has no treatment relationship with the individual prior to and after the test is ordered. No claim for an office visit/telemedicine visit from the ordering/referring provider is ever submitted.
- The provider is located in a different state from the patient without a treating provider documented on the physician order or result.
- The specialty of the ordering provider is unusual for the type of genetic tests ordered.
- The same laboratory service was billed by a third-party laboratory and by a different provider with a modifier 90 within a short time frame.
- The rendering/billing provider has excessively billed for modifier 91 or 59 or -X{EPSU}.

## Modifier 90

When laboratory procedures are performed by a third-party other than the treating or reporting physician or other qualified healthcare professional, the procedure may be defined by adding modifier 90 to the usual procedure code.<sup>25</sup>

## Modifier 91

Modifier 91 may be appended to laboratory procedure(s) or service(s) to indicate a repeat test or procedure on the same day when appropriate.<sup>26</sup> In this way, the 91-modifier may be used to allow the automated payment system to pay a claim it would not otherwise pay.<sup>22</sup>

## Modifier 59 or -X{EPSU}

Under certain circumstances, the physician may need to indicate that a procedure or service was distinct or independent from other services performed on the same day.<sup>27</sup>

Claims payment systems may be programmed to override any payment claim edit in the system when modifier 91 or 59 or -X{EPSU} is detected. This enables modifier 91 or 59 or -X{EPSU} to be used to allow the automated payment system to pay a claim it would not otherwise pay.

## Red Flags from Medical Review or In-depth Investigation

- Limited medical record information is provided with the claim, and a generic letter of medical necessity may be submitted.
- Several laboratories use the same letter of medical necessity, requisition, and the content of the family and health history questionnaire appeared to be repetitive.
- The records may document an unusual amount of time between the date of test consent and sample collection and the sample being received by the laboratory.
- The date of the test performed is prior to the signed order.
- The laboratory is not equipped to perform genetic testing.
- The testing may be performed by a third-party laboratory, not the billing laboratory.
- Certain location characteristics:
  - Co-located with clinics that advertise as offering "gene therapy" where walk-ins are "genetically tested" for gene therapy.
  - UPS Store, retail business, home business.

## Current Strategies for Stopping Genetic Testing Fraud, Waste, and Abuse

HFPP Partners take a variety of proactive and reactive actions to detect and prevent genetic testing fraud, waste, and abuse. These approaches can be consolidated into three overarching themes: pre-payment controls (pre-payment review, prior authorization, and fraud prevention technology), post-payment audits, and provider engagement (provider education and disciplinary action).

Specifically, they include:

- **Pre-payment Review:** A provider may be placed on pre-payment review when their billing practice is not in compliance with the insurance carrier's policies, and their documentation is not sufficient to support their billing of claims. Each time the provider submits a claim, the claim is held, and a copy of the medical record is requested and reviewed for medical necessity.
- **Genetic Testing Prior Authorization:** To ensure that the results of genetic testing are beneficial in patient medical treatment, some HFPP Partners require the completion of a prior authorization form submitted before payment along with supporting documentation, including a letter of medical necessity from a genetic counselor indicating how the test results will be utilized in the patient medical treatment.

One Partner's organization has adopted a proactive approach to reducing the risk of fraud, waste, and abuse of genetic testing services. A robust pre test process has been established to ensure the appropriate use of genetic tests. To receive a test, an individual needs to consult with a qualified and appropriately trained provider to understand the process and testing options and potential outcomes. Pre-authorization is required, and the information will be reviewed for medical necessity.

- Fraud Prevention Technology:** New technologies, such as machine learning software and artificial intelligence, have been developed to distinguish legitimate and fraudulent behaviors and reveal previously unseen patterns/fraud tactics. Such approaches can be more accurate and efficient than rule-based approaches to identify and combat fraud considering the growing complexities of fraud. Data consolidation from multiple sources and real-time data analysis should be used for healthcare fraud detection and prevention. Payers have implemented technologies to protect program integrity and prevent illegitimate payments for improper billing.
- Post-Payment Audits:** Regular monitoring for inappropriate payment and claims, as well as an assessment of fraud risks and compliance with the applicable laws, are effective ways to prevent healthcare fraud. The indicators of potential fraud, waste, and abuse listed in the previous section may be helpful in identifying potential claims for audit.
- Provider Education:** Providers are given necessary educational materials and training about the policies for specific genetic testing coverage. This offers providers awareness about appropriate uses of particular genetic tests. When taking administrative actions, payers are often first called upon to demonstrate that a provider has been notified/warned/educated about a billing violation. More stringent administrative penalties, such as terminating that provider's billing privileges, would be imposed if the provider's inappropriate billing behavior continues.
- Disciplinary Action:** Individuals engaging in fraudulent activities, such as described above, are subject to national and institutional disciplinary action such as suspension of marketing, enrollment, or payment. At a state level, loss of professional license may be applicable.

Since 2011, CMS has used the Fraud Prevention System to run predictive algorithms against all Medicare fee-for-service claims prior to payment to identify potential fraud.<sup>28</sup>

## Actions to Consider

Based on feedback from HFPP Partners, the following actions may be valuable to consider and apply for future interventions.

- Payment Review Process Assessment:** Assessment of the existing pre- and post-payment review process and identification of risks in the individual system are important initial steps to define the gaps and check where the above-mentioned strategies can be implemented.
- Public Education:** Several HFPP Partners noted a lack of public education about genetic testing. It may be beneficial to develop public education communications and resources to share with patients about fraud schemes and the appropriate use of genetic testing. Individuals are strongly advised to have discussions with a care provider or consult with a genetic counselor before requesting a genetic test to ensure the appropriate use and correct interpretation of the test result(s).

Tips for protecting an individual from scams, shared by the Office of Inspector General for the U.S. Department of Health and Human Services<sup>29</sup> include:

- If an individual receives a genetic testing kit in the mail, they should not accept it unless it was ordered by their physician
- Individuals should be suspicious of anyone who offers “free” genetic testing and then asks for their insurance information
- A physician whom an individual knows and trusts should assess their condition and approve any requests for genetic testing
- Individuals should be cautious of all unsolicited requests for their medical identity information. If anyone other than their physician’s office requests their insurance information, they should not provide it

- **Internal Review of Recent Evidence:** The number of genetic tests is rapidly increasing, with over ten tests coming to the market every day.<sup>8</sup> It is essential for fraud prevention units to continuously monitor for newly approved tests. Implementation of a regular internal review cycle is essential to be informed of the changes or updates to a specific genetic test.

# CONCLUSION

As genetic testing is still a relatively new field in the clinical setting, challenges remain regarding the need for an evaluation framework for clinical utility, payer oversight, and timely education on genetic testing. There are already examples of fraud, waste and abuse involving genetic testing and they may become more prevalent as the use of genetic testing grows. They also may become more severe, and while no HFPP Partner has reported any known instance of direct patient harm so far as a result of genetic testing, organizations will need to continuously monitor for potential patient harm as a result of genetic testing fraud, waste, and abuse.

The overview of genetic testing in this paper is intended to provide HFPP Partners and other concerned organizations with a general understanding of genetic testing and the areas that make it susceptible to fraud, waste, and abuse. The strategies for detecting fraud, waste, and abuse in genetic testing and actions to consider outlined in this paper can be used as a foundation for each organization to reference in their own efforts to address this issue. While this is a quickly evolving field, there are things that can be done now to reduce the potential for genetic testing fraud waste and abuse going forward.

# REFERENCES

1. CMS. [Healthcare Fraud Prevention Partnership: About the Partnership](#). 2020 [cited 2020 2/26].
2. American Medical Association. [Precision Medicine: Genetic testing](#). 2020 [cited 2020 1/29].
3. Mayo Clinic. [Genetic Testing: Overview](#). 2020 [cited 2020 1/29].
4. National Library of Medicine. [Your guide to understanding genetic conditions: What are the types of genetic tests?](#) 2020 [cited 2020 2/15].
5. Sanger F, Nicklen S, Coulson AR. *DNA sequencing with chain-terminating inhibitors*. Proc Natl Acad Sci. 1977. 74(12): p. 5463-67.
6. Lee H, Martinez-Agosto JA, Rexach J, Fogel BL. *Next generation sequencing in clinical diagnosis*. Lancet Neurol. 2019. 18(5): p. 426.
7. Minton J. *Data Analytics for Genetic Testing Fraud*. In HFPP Webinar Series Featuring DOJ-FBI. Presented 2019-12-10
8. Phillips KA, Deverka PA, Hooker GW, Douglas MP. *Genetic test availability and spending: Where are we now? Where are we going?* Health Aff. 2018. 37(5):710-16.
9. Chen B, Gagnon M, Shahangian S, Anderson NL, Howerton DA, Boone DJ. [Good Laboratory Practices for Molecular Genetic Testing for Heritable Diseases and Conditions](#). 2009 [cited 2020 2/11].
10. Electronic Code of Federal Regulations. [Title 21: Food and Drugs PART 809 – In Vitro Diagnostic Products for Human Use](#). 2020 [cited 2020 7/1].
11. Food and Drug Administration. [Laboratory Developed Tests](#). 2018. [cited 2020 3/9].
12. California Department of Public Health. [Issues Specific To California Clinical Laboratory Law](#). 2017 [cited 2020 3/9].
13. U.S. Preventive Services Task Force. [Final Recommendation Statement, BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing](#). 2019 [cited 2020 1/15].
14. NCCN Guidelines Version 3. [Genetic/Familial High-Risk Assessment: Breast and Ovarian](#). 2019 [cited 2020 2/12].
15. American College of Surgeons. [National Accreditation Program for Breast Centers Standards Manual](#). 2018 [cited 2020 2/12].

16. American College of Obstetricians and Gynecologists. [Committee Opinion: Counseling About Genetic Testing and Communication of Genetic Test Results](#). 2017 [cited 2020 2/12].
17. Gillis NK, Innocenti F. *Evidence required to demonstrate clinical utility of pharmacogenetic testing: The debate continues*. Clin Pharmacol Ther. 2014. 96(6):655-7.
18. Janssens ACJW, Deverka PA. *Useless until proven effective: The clinical utility of preemptive pharmacogenetic testing*. Clin Pharmacol Ther. 2014. 96(6):652-4.
19. Food and Drug Administration. [The FDA Warns Against the Use of Many Genetic Tests with Unapproved Claims to Predict Patient Response to Specific Medications: FDA Safety Communication](#). 2018 [cited 2020 2/11].
20. National Human Genome Research Institute. [Promoting Safe and Effective Genetic Testing in the United States – Chapter 2](#). 2005 [cited 2020 3/9].
21. Rossi E, Jeffrey GP. *Clinical penetrance of C282Y homozygous HFE haemochromatosis*. The Clinical Biochemist Reviews. 2004. 25(3):183-90.
22. Healthcare Fraud Prevention Partnership. [Examining Clinical Laboratory Services: A Review By the Healthcare Fraud Prevention Partnership](#). 2018 [cited 2020 2/25].
23. Department of Justice. [Federal Law Enforcement Action Involving Fraudulent Genetic Testing Results in Charges Against 35 Individuals Responsible for Over \\$2.1 Billion in Losses in One of the Largest Health Care Fraud Schemes Ever Charged](#). 2019 [cited 2020 2/12].
24. Department of Justice. [Genetic Testing Company Agrees to Pay \\$1.99 Million to Resolve Allegations of False Claims to Medicare for Medically Unnecessary Tests](#). 2019 [cited 2020 3/3].
25. CMS. [Medicare Claims Processing Manual Chapter 16 – Laboratory Services](#). 2020 [cited 2020 5/14].
26. CMS. [Medicare Claims Processing Manual Chapter 23 – Fee Schedule Administration and Coding Requirements](#). 2020 [cited 2020 7/1].
27. CMS. [Modifier 59 Article](#). 2017 [cited 2020 4/17].
28. Department of Health and Human Services. [CMS Report to Congress Fraud Prevention System, Second Implementation Year](#). 2014 [cited 2020 3/3].
29. AARP. Scams & Fraud: [How to Avoid DNA Medicare Scams](#). 2019 [cited 2020 3/9].