

## Genetic Testing: An Ever-Evolving Health Field Raises Complex Coverage Issues

Since the 1990s, there has been explosive growth in both the number of genetic tests and the costs associated with them. As part of their constant efforts to manage their health plan costs, trustees of multiemployer health funds need to be aware of these developments so they can periodically revisit their plans' coverage for genetic testing to ensure that it has medical value. This *NewsLetter* presents an overview of the issues and options.

### WHAT IS GENETIC TESTING?

Genetic testing is a type of medical test that identifies changes in chromosomes, genes or proteins and is used to find changes that are associated with inherited disorders. There are many situations where genetic tests can provide useful information. For example, the results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or not developing a genetic disorder.

Genetic testing is not new. For decades, obstetricians have been screening for genetic disorders at birth and during the early months of pregnancy. Newborn screening for Phenylketonuria (PKU), an enzyme deficiency that can affect brain development, started in the 1960s. Screening for Down's syndrome during pregnancy began in the 1970s. Lately, however, payment for genetic tests has raised pressing concerns for plan sponsors mainly

because of the rising number of available genetic tests and the associated costs.

### THE RANGE OF GENETIC TESTING TODAY

Several hundred genetic tests are currently in use. The graph below shows the dramatic growth in the number of diseases for which genetic testing is available. More genetic tests are being developed all the time.<sup>1</sup> For example, between late October and late November of 2009, seven new genetic tests were added to the GeneTests Laboratory Directory maintained by the National Center for Biotechnology Information (NCBI).<sup>2</sup> The NCBI keeps a current

<sup>1</sup> Genetic home reference by U.S. National Library of Medicine: <http://ghr.nlm.nih.gov/handbook/testing/genetic-testing>

<sup>2</sup> The NCBI is a division of the National Library of Medicine at the National Institutes of Health. The GeneTests Laboratory Directory is available on the following page of the NCBI's Web site: <http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/whatsnew/wndz3.shtml>

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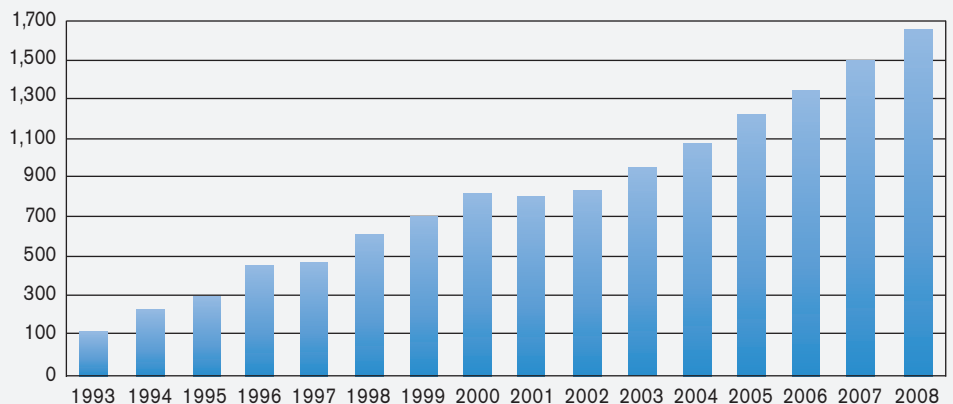
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Number of Diseases for which Genetic Testing Is Available



Source: GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1993-2010. All Rights Reserved. Available at <http://www.genetests.org>. Accessed January 15, 2010. This information in this graph is from the graph on the following Web page: <http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/whatsnew/labdirgrowth.shtml>

“Some genetic tests can improve care.”

list of genetic tests.<sup>3</sup> As the availability of genetic technologies has grown, their adoption by clinicians and consumers has increased dramatically.

#### DIRECT AND INDIRECT COSTS ASSOCIATED WITH GENETIC TESTING

The cost of genetic testing varies widely. Some newer diagnostic tests are very expensive. For example, a full-sequence DNA analysis of two genes associated with breast cancer cost \$2,600 in 2009. Moreover, often there are multiple ways to test for a given genetic syndrome or mutation. These different methods can vary significantly in price.

There may also be coverage costs associated with positive genetic tests. For example, biotech (also known as specialty) drugs, genetically engineered compounds designed to target and treat specific diseases, may be prescribed to treat the condition discovered by the genetic test. According to the 2004 *Segal Health Plan Cost Trend Survey*, the per-prescription cost of biotech drugs increased 29 percent from 2001 to 2003 compared to 16 percent for conventional drugs during the same period.<sup>4</sup>

Despite the increased use of genetic testing, it is very difficult for plan sponsors to obtain information on their total expenditures for genetic testing. Factors contributing to this

situation include coding inconsistencies on claims and the lack of codes for certain, newer genetic tests.

#### GENETIC TESTING CAN IMPROVE CARE

The various ways in which some genetic tests can improve care include the following:

- *Clarify a diagnosis.* Genetic information can direct a physician toward appropriate treatments. Cystic fibrosis and myotonic dystrophy (progressive muscular dystrophy) are examples of disorders that may be confirmed or ruled out by diagnostic genetic testing.
- *Identify people at high risk for conditions that may be preventable.* For example, aggressive monitoring (e.g., screening colonoscopy) for and removal of colon growths in those inheriting a gene for colon and rectum cancer has saved many lives.
- *Predict an individual’s response to drug therapy.* This “personalized medicine” approach, known as pharmacogenomics, aims to direct specific drug therapy only to individuals who can respond to it and avoid it in individuals who cannot benefit, avoiding the usual “trial-and-error” approach to drug treatment. For example, about 20 percent of people prescribed anti-depressants will find the drugs ineffective because of their genetic factors, delaying effective treatment and unnecessarily increasing prescription drug expenditures. In some children with leukemia, pharmacogenomics testing can identify patients who are poor metabolizers of common anti-cancer drugs and may develop severe side effects if they receive standard doses of drugs. Another example is HER2 testing, which can identify breast cancer patients who would benefit from a very effective, but expensive drug, like

Herceptin. Yet another example is knowing the genetic makeup of the hepatitis C virus<sup>5</sup> in order to influence the duration of drug therapy for hepatitis. These examples of using genetic testing to target drug therapies highlight the potential for eventual plan cost savings, as spending on ineffective treatments eventually declines.

- *Help women, their partners and their doctors find out early in a pregnancy (or even before conception) about any potential health problems their babies may face.* Knowing about possible conditions before and during pregnancy is now a reality for consumers. Plan sponsors will have to determine how they want to address coverage for such diagnostics.

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#### VALUE OF OTHER GENETIC TESTS LESS CLEAR

The value of some genetic tests has not yet been established. For example, the cost-effectiveness of gene tests for adult-onset disorders, such as Alzheimer’s disease and some cancers, is currently debatable. These tests are targeted to healthy (pre-symptomatic) people who, through genetic testing, may be identified as being at high risk because of a strong family medical history for the disorder. However, these tests give only a probability for developing the disorder, and do not provide any more information than is obtained from a query of family history. Uncertainties surrounding test interpretation, the current lack of available medical options for these diseases, the tests’ potential

<sup>3</sup> That alphabetical list is available on the following page of the NCBI’s Web site: <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene>

<sup>4</sup> The survey report is available on the following page of Segal’s Web site: <http://www.segalco.com/publications/surveysandstudies/summer03preliminarytrendsreportfindings.pdf> (According to the 2010 *Segal Health Plan Cost Trend Survey*, the 2010 trend rate for specialty/biotech drugs is expected to remain at 18 percent, which is nearly double that of the retail trend. See <http://www.segalco.com/publications/surveysandstudies/2010trendsreport.pdf>)

<sup>5</sup> Hepatitis C is the most common cause of chronic liver failure and liver transplant in the U.S.

for provoking anxiety, and risks for discrimination and social stigmatization could outweigh the benefits of some genetic testing.<sup>6</sup> Testing might also lead to unnecessary care and treatments.

Another example is testing for the progressive neurodegenerative disease amyotrophic lateral sclerosis (ALS) or Lou Gehrig's disease. Most people with ALS have a form of the condition that is described as sporadic or non-inherited. The cause of sporadic ALS is largely unknown, but probably involves a combination of genetic and environmental factors. The diagnosis of ALS is based on clinical features, electrodiagnostic testing<sup>7</sup> and exclusion of other health conditions with related symptoms. The only genetic test currently available detects the SOD1 mutation.<sup>8</sup> Since only 20 percent of familial ALS patients will test positively for an SOD1 mutation, this test has limited value in genetic counseling. At present, genetic testing in ALS has questionable statistical value in making the diagnosis.

### COVERAGE CRITERIA FOR GENETIC TESTING

Genetic science is complex and its correct use in clinical practice is challenging for most clinicians. A large majority of physicians who do not specialize in genetics (72 percent) rate their knowledge of genetics as fair to poor.<sup>9</sup> In light of the complexity, determining the coverage criteria for

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genetic testing is particularly difficult for insurers and trustees.

In February 2006, the Department of Health and Human Services (HHS) released a report produced by the Secretary's Advisory Committee on Genetics, Health and Society, “Coverage and Reimbursement of Genetic Tests and Services.”<sup>10</sup> The report recommends coverage of genetic testing if at least one of the following criteria is met:

- The patient has current signs and/or symptoms (*i.e.*, the test is being used for diagnostic purposes), and conventional diagnostic procedures are inconclusive.
- The patient has a risk factor or a particular family history that indicates a genetic cause.
- The patient meets defined criteria that place them at high genetic risk for the condition.
- The test is not considered experimental or investigational.
- The test is performed by a laboratory certified by the Clinical Laboratory Improvement Amendments (CLIA).<sup>11</sup>
- The test result will directly influence the disease treatment management of the covered member.
- Testing is accompanied by pre-test and post-test counseling when appropriate under the circumstances.

- Testing is performed by well-trained genetic medicine clinicians. (There are fewer than 2,000 certified genetic counselors in United States.<sup>12</sup>)

The HHS report also recommended when genetic testing should *not* be covered. Those recommendations follow:

- The test result will have no direct influence on the disease treatment management of the covered member.
- The test is for population screening without a personal or family history, with the exception of newborn screening and preconception or prenatal carrier screening for certain conditions, such as cystic fibrosis, Tay-Sachs disease,<sup>13</sup> sickle-cell disease and other hemoglobinopathies.<sup>14</sup>
- The test is for informational purposes only.
- The test is for adult-onset conditions and is being performed on a minor.
- The test is for a relative of a plan participant who is not also a plan participant unless (1) the genetic test results are necessary for the medical care of the plan member, and (2) the relative can provide evidence of coverage denial from his or her health insurance plan.

Plan sponsors should get expert advice when determining their plan's coverage rules for genetic testing. They may want to consider the HHS recommendations as they devise their own policy on coverage of genetic testing.

<sup>6</sup> Human Genome Project Information is available on the following web page: [http://www.ornl.gov/sci/techresources/Human\\_Genome/medicine/genetest.shtml](http://www.ornl.gov/sci/techresources/Human_Genome/medicine/genetest.shtml)

<sup>7</sup> Electrodiagnostic testing measures the electrical activity of nerves and muscles.

<sup>8</sup> SOD1 is an abbreviation for Superoxide dismutase 1, a protein and gene that act as an anti-oxidant.

<sup>9</sup> Menasha, Joshua D., M.D., Clyde Schechter, M.D., and Judith Willner, M.D. “Genetic Testing: A Physician's Perspective,” *The Mount Sinai Journal of Medicine* 2000; 67(2):144-51 ([http://www.mssm.edu/mjsjournal/67/page144\\_151.pdf](http://www.mssm.edu/mjsjournal/67/page144_151.pdf)).

<sup>10</sup> The 114-page report, *Coverage and Reimbursement of Genetic Tests and Services*, is available on the following page of the HHS Web site: [http://oba.od.nih.gov/oba/sacghs/reports/CR\\_report.pdf](http://oba.od.nih.gov/oba/sacghs/reports/CR_report.pdf)

<sup>11</sup> Information about the CLIA program, which is managed by the Centers for Medicare & Medicaid Services (CMS), is available on the following page of the CMS Web site: <http://www.cms.hhs.gov/clia/>

<sup>12</sup> Judith Cooksey, M.D., M.P.H., “Genetic Service Providers – Concepts and Findings Emerging from the Genetics Workforce Study.” Testimony to the National Institutes of Health Secretary's Advisory Committee on Genetics, Health, and Society, October 22, 2003: <http://oba.od.nih.gov/oba/sacghs/meetings/October2003/Cooksey.pdf>

<sup>13</sup> Tay-Sachs disease is a fatal genetic condition that affects infants.

<sup>14</sup> Hemoglobinopathies are conditions, such as anemia, related to a deficiency in the genes responsible for producing the hemoglobin protein that red blood cells need to transport oxygen.

**OTHER IMPORTANT CONSIDERATIONS**

Trustees of health funds that decide to cover genetic testing should consider the following issues:

➤ **Compliance Considerations**

The federal government has taken steps to regulate how plans may use genetic information through the Genetic Information Nondiscrimination Act of 2008 (GINA), which is discussed in the text box below.

**About GINA**

GINA prohibits group health plans and health insurance issuers from:

- Adjusting premium or contribution rates for the group on the basis of the genetic information of individuals in the group,
- Requiring or requesting that an individual or family member undergo a genetic test,
- Collecting (i.e., requesting, requiring or purchasing) genetic information for underwriting purposes (broadly defined) or prior to enrollment in the plan, and
- Using or disclosing protected health information (PHI) that is genetic information for underwriting purposes.

GINA defines “genetic information” to include genetic tests and services, as well as family history of a disease or disorder.\* This will affect plans that include questions about family history in health-risk assessments. Proposed GINA regulations issued last fall address this issue.\*\*

GINA takes effect with the plan year that begins after May 21, 2009 (one year after the law was enacted). That means the effective date for calendar-year plans is January 1, 2010. Plan sponsors that fail to comply with GINA may face a civil monetary penalty of \$100 per violation.

\* For more information about GINA, see Segal's June 2008 *Bulletin*, “New Genetic Information Nondiscrimination Act Regulates Group Health Plans”: <http://www.segalco.com/publications/bulletins/june08GINA.pdf>

\*\* For information on that guidance on how GINA affects incentives in wellness and disease management programs, see Segal's October 2009 *Bulletin*, “GINA Regulations Require Redesign of Health Plan Wellness and Disease Management Incentives”: <http://www.segalco.com/publications/bulletins/oct09GINAregs.pdf>

- **The Critical Role of Genetic Counseling** The importance of genetic counseling before a test is done, and again when the test results are delivered, is often overlooked. Pre-test genetic counseling can help determine whether testing is actually appropriate, will help the patient understand the benefits, limitations, and potential risks of genetic testing, and will help psychologically prepare a person who chooses to undergo a test. Post-test counseling is important for helping the person understand the test results and the increased risk/genetic susceptibility they and their at-risk family members have, as well as determine what steps the person should take next for risk reduction and management.

- **Communicating Coverage for Genetic Testing** Communication about a fund's coverage for genetic testing is important because participants have difficulty understanding how genetic technologies can factor into their care. Trustees may wish to take special care in explaining the fund's coverage criteria in summary plan descriptions and other plan documents.

**CONCLUSION**

The ever-evolving health field of genetic testing is complex and is continually evolving. It is a challenge for plan sponsors to stay on top of scientific developments and coverage recommendations in order to make informed decisions about which tests their group health plan will (and will not) cover. In practice, trustees need to rely on expert advice to help with their decision making about coverage of genetic testing, discussions that should take place on a regular basis to keep pace with scientific and medical advances. Trustees should also look to established reimbursement policies set by Medicare, Medicaid and other major payers as

a frame of reference to help establish defensible, written coverage policies for genetic testing.



*Segal can help trustees of multiemployer health funds with decision making about coverage of genetic testing, including implementing the HHS criteria and related plan design issues. For more information, contact your Segal consultant, the nearest Segal office or one of the following experts:*

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