



GENETICS: COUNSELING, TESTING, SCREENING*

Effective Date: October 19, 2011

Review Dates: 8/07, 10/07, 8/08, 8/09, 10/09, 2/10,
8/10, 8/11, 10/11

Date Of Origin: August 8, 2007

Status: Current

**This policy includes the following previously separate policies: 91450 Genetic Counseling, Testing and Screening; 91513 Gene Expression Analysis; 91449 Genetic Testing Pre-implantation*

Summary of Changes

Clarifications:

-

Deletions:

-

Additions:

- Pg 8, Section I, K, 1, A, 4, additional criteria for coverage added for Oncotype DX™ Breast (i.e. individuals with 1-3 positive lymph nodes).
- Pg 9, Section I, K, 4, added criteria for the coverage of AlloMap™ Molecular Expression Testing for Heart Transplantation Rejection when specific criteria are met.
- Pg 9, Section I, K, 5, added non-coverage criteria for gene expression testing to predict coronary artery disease, e.g. Corus Dx, as it is considered experimental and investigational for all applications.

I. POLICY/CRITERIA

- A. Coverage for genetic counseling, testing and/or screening is provided to Priority Health members when **all** of the following apply:
1. Appropriate genetic counseling occurs before and after testing*.
 2. Documented informed consent occurs before testing**.
 3. There is documented reasonable expectation based on family history, pedigree analysis, risk factors, and/or symptomatology that a genetically inherited or acquired condition exists and the member displays clinical features, or is at direct risk of inheriting the mutation in question (pre-symptomatic) or comes from the appropriate disease-specific population. A three generation pedigree should accompany the request for testing.
 4. Knowledge of the presence or absence of condition would directly affect medical care of the member.
 - a. The disease is treatable and/or preventable and
 - b. The test results will lead to a marked change in the intensity of surveillance frequency and /or intensity of treatment for that disease.
 5. The testing is FDA / CLIA approved
 6. Testing is ordered by the appropriate provider, see section C below:

*Members must have genetic counseling by a genetic counselor **before** testing can occur for certain conditions (see appendix A below). Counseling can occur at the



same visit as the sample(s) for testing are collected with appropriate documentation as defined above.

** Health care practitioners in the State of Michigan must follow state law regarding informed consent for predictive genetic testing. (Michigan State Law. 333.17020 Genetic test; informed consent.

[http://www.legislature.mi.gov/\(S\(bcot2wnj3puzmg550rnzukyf\)\)/mileg.aspx?page=getoject&objectname=mcl-333-17020](http://www.legislature.mi.gov/(S(bcot2wnj3puzmg550rnzukyf))/mileg.aspx?page=getoject&objectname=mcl-333-17020)

- B. Family Planning and Infertility services may be limited by the member's specific plan and are subject to applicable copays and or coinsurance. See plan documents for details. Family planning and infertility services of any kind are NOT covered for Medicaid members.

C. Limits/Indications

Eight medically appropriate genetic testing categories appear in Sections E through L; these sections identify providers authorized to order the tests and give examples of indications for testing (prior authorization is only *required* for tests in Section I, J, K and L. Obtaining specimens for tests in sections D#2, F, I, J and L and when indicated in sections G and H *must* be coordinated by a Genetic Counselor's office – see appendix A below for a list of specific conditions for which genetic counseling is required and/or recommended).

D. Prenatal Testing

1. Prenatal screening via maternal serum analysis (first and/or second trimester) with nuchal translucency* measurement may be covered for all pregnant women within the prescribed time frame for each of the maternal serum screening options.
2. Prenatal diagnostic genetic testing (via amniocentesis or chorionic villus sampling) will be covered for pregnant women when the member has received genetic counseling by a genetic counselor, the reason for testing is documented, and the testing is ordered by a Family Practice physician providing OB services or by an Obstetrician. Common indications for prenatal diagnosis include but are not limited to:
 - a. Abnormal fetal ultrasound findings
 - b. Abnormal maternal serum first trimester screening, second trimester triple or quad screen, integrated* or alpha-fetoprotein, elevated MSAFP
 - c. Increased risk based on documented family history or carrier status
3. Prenatal diagnostic genetic testing is **not** a covered benefit for:
 - a. Sex determination unless medically indicated
 - b. Prenatal determination of paternity



- c. Preconceptual testing of sperm, ova, embryos for use in assisted reproduction (e.g. artificial insemination, IVF)

*Nuchal translucency (NT) measurement will only be covered when combined with first trimester serum screening (“combined testing”) at centers which have appropriate certification to do so by either the Fetal Medicine Foundation (FMF) or the Nuchal Translucency Quality Review Program (NTQR). Nuchal translucency measurement alone is not a covered benefit.

- E. **Genetic Carrier Screening:** Indications for genetic carrier testing for at-risk individuals include but are not limited to:
1. African American, Caribbean, West-Indian, West African, Hispanic Caribbean, Mediterranean, Asian, Middle Eastern and other individuals who may be at risk for hemoglobinopathies including sickle cell anemia, alpha and/or beta thalassemia based on ethnicity.
 2. Ashkenazi disease screen, for individuals of Jewish descent (e.g., Tay Sachs, Canavan’s Disease, etc.)
 3. Carrier testing* for cystic fibrosis (CF) is medically necessary for members in *any* of the following groups:
 - a. Couples seeking prenatal care; *or*
 - b. Couples who are planning a pregnancy; *or*
 - c. Reproductive partners of persons who have CF or are carriers of CF; *or*
 - d. Males with a diagnosis of congenital bilateral absence of the vas deferens (CBAVD); *or*
 - e. For persons with a family history of CF or a first degree relative identified as a CF carrier see section F below for coverage rules.

Genetic carrier screening for CF is considered experimental and investigational for all other indications.

*covered testing is for a core panel of 25 mutations that are recommended by the American College of Medical Genetics (ACMG) medically necessary for cystic fibrosis genetic testing. The standard mutation panel is available at: <http://www.ama-assn.org/ama/no-index/about-ama/3021.shtm>. Testing for additional CF mutations through an expanded panel will be covered for certain indications when recommended by a genetic counselor.

- F. **Genetic Carrier Testing:** Testing for carrier status of parents with a known genetic risk or suspected carrier status based on their reproductive and/or family history. This testing should only be ordered following genetic counseling by a genetic counselor.
1. Individuals with a known family history of a recessive genetic condition (i.e. Spinal Muscular Atrophy, Sickle Cell, Cystic Fibrosis).



2. Family members of infants identified by newborn screening as affected or carriers of a genetic condition.
3. Parental chromosome analysis following the diagnosis of a child with a chromosome abnormality for which a parent may be carrying a chromosome rearrangement or abnormality.
4. Chromosome analysis for couples with 2 or more miscarriages.

G. Suspected Genetic Conditions/ Diagnostic testing – Pediatrics(<18 years old):

Careful consideration must be given to genetic testing and screening of children to ensure that use of this technology promotes the best interest of the child.

Identification of the genetic condition must provide a clear benefit to the child.

Testing must be recommended by a Genetic Counselor except as noted below. Tests to confirm or rule out suspected genetic conditions in symptomatic individuals in which confirming a diagnosis would alter the medical management for the individual. This includes but is not limited to the following examples:

1. Chromosomal analysis in a newborn with features of Down Syndrome.
2. Fragile X in a child with mental retardation and /or autism
3. Spinal Muscular Atrophy in a child with neuromuscular problems
4. Sickle Cell Disease
5. Priority Health will cover chromosomal analysis or molecular testing (chromosomal microarray (CMA) or comparative genomic hybridization (CGH) to confirm suspected genetic conditions only when ordered by Family Practice, Internal Medicine, Neurology, Obstetric/Gynecology, or Pediatric physicians and in the presence of any of the following:
 - a. Congenital malformation(s)
 - b. Conditions with a known or suspected chromosomal, single gene, mitochondrial, or multifactorial etiology
 - c. Unexplained failure to thrive
 - d. Unexplained developmental delay or loss of developmental milestones
 - e. Unusual growth pattern
 - f. In-utero death of the fetus in the second or third trimester

H. Prognostic Testing: Prognostic testing may be done when the clinical presentation is suggestive of a specific condition and the results will help to determine additional testing or treatment modalities related to existing nonhereditary conditions including, but not limited to the following:

1. Chromosomal analysis for leukemia
2. Flow cytometry for lymphoma
3. Her2Neu for breast cancer
4. HLA Haplotype Analysis for transplant procedures
5. P53 Tumor Marker
6. HLA genotyping for Celiac associated DQ alleles when serologic testing and or intestinal biopsy results are inconsistent with one another or clinical presentation.



Priority Health will cover prognostic genetic testing only when recommended by Genetic Counselors **or** ordered by specialty physicians with expertise in the specific clinical area for which the testing is being done.

- I. **Predictive Genetic Testing:** Predictive testing is offered to asymptomatic individuals with a family history of a genetic disorder. Predictive testing is of two types:

- **presymptomatic** (eventual development of symptoms is certain when the gene mutation is present, e.g., Huntington disease) and
- **predispositional** (eventual development of symptoms is likely but not certain when the gene mutation is present, e.g., breast cancer).

Predictive testing is **MEDICALLY INDICATED** only if early diagnosis allows interventions which reduce morbidity or mortality. Predictive testing for the presence of presymptomatic or predisposition genetic changes in an at-risk individual may be done for conditions such as:

1. BRCA1/BRCA2
2. Hereditary nonpolyposis colorectal cancer (HNPCC)
3. Huntington's Chorea
4. Multiple Endocrine Neoplasia
5. Myotonic Dystrophy
6. Family history of genetic disorders (for example, a previous child with Duchenne's Muscular Dystrophy)
7. CADASIL Genetic Testing: DNA testing for CADASIL is considered to be medically necessary for *either* of the following indications:
 - a. Symptomatic individuals who have a family history consistent with an autosomal dominant pattern of inheritance of this condition (clinical signs and symptoms of CADASIL include stroke, cognitive defects and/or dementia, migraine, and psychiatric disturbances); *or*
 - b. Pre-symptomatic individuals where there is a family history consistent with an autosomal dominant pattern of inheritance and there is a known mutation in an affected member of the family.CADASIL genetic testing is considered experimental and investigational for all other indications.
8. Priority Health will cover hereditary predisposition/pre-symptomatic genetic testing only when recommended by a Genetic Counselor.
9. **All** genetic testing in this Section **requires** prior authorization by Priority Health, and must include documentation:
 - a. Of medical necessity
 - b. That genetic counseling has been accomplished
 - c. That informed consent has been obtained
 - d. Results of familial mutation / test results are available upon request

- J. **Preimplantation Genetic Diagnosis (PGD):** Preimplantation genetic diagnosis (PGD), including the oocyte polar body or cleavage stage embryo biopsy procedure, associated genetic testing, and pre- and post-test genetic counseling associated with



PGD, is considered medically necessary when the results of the genetic test will impact clinical decision making and/or clinical outcome. PGD in the following situations is intended to be used as an alternative to prenatal genetic diagnosis using amniocentesis or chorionic villus sampling (CVS) and only when a reliable genetic test exists for the disorder and is well established:

1. For the detection of single gene disorders* for the following situations:
 - a. Both partners are known carriers of a single autosomal recessive gene
 - b. One partner is a known carrier of a single gene autosomal dominant disorder
 - c. One partner is a known carrier of a single X-linked disorder

*The disorders resulting from the above genetic disorders (a-c) are potentially lethal or disabling and have limited treatment options and include but are not limited to the following: Tay Sachs, spinal muscular atrophy, myotonic dystrophy, Huntington's disease, and X-linked disorders including Duchenne's muscular dystrophy and Fragile-X syndrome. All disorders for which testing is requested should have significant probability for recurrence, i.e. 25% or more likelihood for recurrence.

2. Couples in whom one or more partners has a known chromosomal abnormality such as a balanced translocation.
3. PGD is NOT covered for any of the following indications or those not specified specifically above because they are considered experimental, investigational or unproven:
 - a. Screening of common aneuploidy in women of advanced maternal age (i.e., age 35 or older), with repeat IVF failures or recurrent spontaneous abortions, or for the purpose of improving IVF implantation success
 - b. Detection of chromosomal translocations in women of advanced maternal age, with repeat IVF failures or recurrent spontaneous abortions, or for the purpose of improving IVF implantation success
 - c. For the purpose of human leukocyte antigen (HLA) typing of an embryo to identify a future suitable stem cell, tissue or organ transplantation donor
 - d. For the purposes of carrier testing to determine carrier status of the embryo (determination of carrier status is performed on individuals contemplating reproduction)
 - e. Using blastocyst stage biopsy
 - f. For adult-onset/late-onset disorders (e.g., Alzheimer's disease; cancer predisposition)
 - g. Testing of embryos for non-medical gender selection or non-medical traits
4. All other services associated with PGD are subject to the member's specific policy and associated certificate of coverage (COC). This includes, but is not limited to, all services and supplies relating to artificial insemination, in-vitro fertilization, embryo or ovum transfer procedures, any other assisted



reproduction procedure, prescription drugs designed to achieve pregnancy, and ultrasounds for egg harvest. Please refer to the appropriate COC for further information.

5. For Medicaid and MICHild – IVF, artificial insemination and other forms of infertility treatment are not a covered benefit, therefore genetic counseling/testing/screening done in conjunction with these services would not be covered.

K. Gene Expression Analysis: see specific areas of application below. Prior authorization is required for any covered applications. The only exception is Breast Cancer Treatment Assessment with Oncotype DX™.

1. Breast Cancer Treatment Assessment

Several panels of gene expression markers have been developed for the purpose of predicting the likelihood of breast cancer recurrence in various populations of women with node-negative disease. These panels may be useful for identifying women who are unlikely to experience recurrence and, thus, unlikely to benefit from adjuvant chemotherapy. Such panels are designed to identify women who can safely avoid adjuvant chemotherapy, without negatively affecting disease-free and overall survival outcomes.

Currently there are several panels available for use in determining treatment strategy. However, based upon published reports of reproducibility outside of the initial study, only Oncotype DX™ (21-gene panel; Genomic Health) is available as a covered benefit at this time. Oncotype DX™ measures the levels of expression of 21 genes (whether they are transcribed into messenger RNA) in breast tumors. This assessment is intended to help guide a person's risk of recurrence more precisely than standard characteristics, such as tumor size and grade alone. Based on the Oncotype DX™ gene expression analysis, a recurrence score from 0 to 100 is generated; the higher the score, the greater a woman's chance of having a recurrence if treated with hormonal therapy alone. Based on their recurrence score, women are assigned to three different treatment groups:

- a. Women with a recurrence score higher than 25 generally receive chemotherapy plus hormonal therapy (the standard of care)
- b. Women with a recurrence score lower than 11 generally will receive hormonal therapy alone
- c. Women with a recurrence score of 11 to 25 are encouraged to consider participating in the TAILORx trial (see Note below). Locations for enrollment and participation are available through <http://clinicaltrials.gov>

A. Oncotype DX™ Breast (21-gene panel; Genomic Health) is considered medically appropriate to assess the need for adjuvant chemotherapy in



women with recently diagnosed breast cancer when **all** of the following criteria 1-6 are met. Prior authorization is not required.

1. Breast tumor is stage 1 or stage 2.
2. Breast tumor is estrogen-receptor positive.
3. Breast tumor is HER2-receptor negative, or breast tumor is HER2 receptor positive and less than 1 cm in diameter.
4. There is no evidence of metastatic breast cancer, and the patient is axillary-node negative (nodes with micrometastases less than 2 mm in size are considered node negative) or has 1-3 positive nodes.
5. The patient is a candidate for possible adjuvant chemotherapy (i.e., chemotherapy is not precluded due to other factors).
6. Member and physician (prior to testing) have discussed the potential results of the test and agree to use the results to guide therapy (i.e., member will forgo adjuvant chemotherapy if Oncotype Dx score is low).

- B. For patients choosing chemotherapy for an intermediate risk score, the clinical rationale must be provided for treatment since chemotherapy is still of unproven benefit.

Note:

Unfortunately treatment algorithms do not exist for women with a recurrence score of 11 to 25. The TAILORx trial is designed primarily to evaluate the effect of chemotherapy on those with a recurrence score of 11 to 25. Women participating in this trial who are in this group will be randomly assigned to receive adjuvant hormonal therapy, with or without chemotherapy. The TAILORx seeks to determine if the *Oncotype DX*TM test will be helpful in future treatment planning for this group.

- C. *Oncotype DX*TM is not covered for any other clinical evaluation including *Oncotype DX*TM Colon Cancer Assay.
- D. Other assays of genetic expression in tumor tissue (e.g., MammaPrint®, Rotterdam Signature 76-Panel) are not covered because they are considered experimental, investigational or unproven.

2. Fecal DNA Screening for Colorectal Cancer

Fecal DNA testing is performed on stool samples that are submitted to a laboratory after being collected by patients at home. The test is designed as a screening option for patients who are unwilling or unable to undergo a colonoscopy. The test detects colorectal cancer based on the presence of specific, cancer-associated mutations in DNA that is extracted from the stool sample. These specific DNA mutations in the stool arise from tumors within the colon. This DNA is continuously shed from the tumor into the stool. Patients with a positive fecal DNA test result must then undergo a definitive test for colon



cancer, such as a colonoscopy. Fecal DNA screening is considered to be experimental and investigational and is not a covered benefit.

3. Cytochromes P450 (CYP450)

The AmpliChip CYP450 test was developed as a clinical test to evaluate an individual's metabolic capacity for certain drugs by identifying polymorphisms of 2 CYP450 enzymes (ie, CYP2D6 and CYP2D19). Pharmacogenetic factors operate at pharmacokinetic as well as pharmacodynamic levels- the two components of the dose-response curve of a drug. Polymorphisms in drug metabolizing enzymes, transporters and/or pharmacological targets of drugs may profoundly influence the dose-response relationship between individuals. For some drugs, although retrospective data from case studies suggests that these polymorphisms are frequently associated with adverse drug reactions or failure of efficacy, the clinical utility of such data remains unproven. CYP450 is considered to be experimental and investigational and is not a covered benefit.

4. The AlloMap™ Molecular Expression Test for Heart Transplantation Rejection is medically necessary and, therefore, covered when the individual is at least 12 months post--heart transplant and:

- a. Is at least 15 years of age
- b. Is considered to be stable (ie, if within the first 12-month transplant window there has been no evidence of Grade 2R or Grade 3R graft rejection detected by endomyocardial biopsies in the first six months post-transplant and the individual has not deteriorated since the prior clinical assessment).
- c. Is considered to be a "low risk" for cardiac events (ie, absence of a history of severe allograft vasculopathy, absence of antibody-mediated rejection, and absence of any signs or symptoms of heart failure).

5. Gene expression testing to predict coronary artery disease, e.g. Corus Dx, is considered experimental and investigational for all applications

L. Testing of member and non-member relatives:

1. Genetic testing is *not* a covered benefit if the test results do not provide direct medical benefit to the member unless it provides direct medical benefit to other relatives that are also Priority Health members and this benefit is documented.
2. Genetic testing of a non-member relative of a member *may* be a covered benefit if *all* of the criteria in a through e are met:
 - a. The test results are for the direct medical benefit of the member and testing the nonplan relative is the most cost effective method to obtain the medically necessary information for the member.
 - b. The nonplan relative's insurance company has been billed and payment has been denied.



- c. Coverage is limited to the testing of *five* nonplan relatives as a lifetime benefit for a member.
 - d. Testing of the non-member relative has been recommended by a genetics counselor *and* approved by Priority Health.
 - e. All genetic testing must be processed through a Priority Health provider phlebotomist and laboratory, unless otherwise specified by the Genetics Counselor.
3. In the absence of specific information regarding advances in the knowledge of mutation characteristics for a particular disorder, the current literature indicates that genetic tests for inherited disease need only be conducted once per lifetime of the member.

M. Whole Genome Sequencing

Whole genome sequencing (WGS), also known as full genome sequencing (FGS), complete genome sequencing, or entire genome sequencing is a laboratory procedure which seeks to determine an individual's entire DNA sequence, specifying the order of every base pair within the genome at a single time. The relationship between mutations in the genomic material of asymptomatic individuals and the development of specific diseases is still being analyzed and the role of whole-genome sequencing in the clinical setting has yet to be established. Because there is currently no consensus on genotype/phenotype relationship or how to incorporate this information into direct medical management, the use of WGS is considered to be experimental and investigational.

N. Exclusions

1. The following are examples of services that are not covered:
 - a. Routine, ongoing, or long term genetic counseling.
 - b. Genetic testing to determine the paternity of a child.
 - c. Genetic testing to determine the sex of the child.
 - d. General population screening for genetic disorders (e.g., cystic fibrosis).
 - e. ApoE for hyperlipidemia and/or Alzheimer's Disease

Special Notes: Informed *consent* is *required* for all genetic tests in accordance with Michigan law PUBLIC HEALTH CODE (EXCERPT) Act 368 of 1978: 333.17020 Genetic test; informed consent Sec. 17020

Informed consent indicates the ordering clinician has discussed:

The potential benefits, harms and limitations of the test to be ordered and the implications of positive, negative or ambiguous results.

Members should have access to genetic counseling before genetic testing.



A referral to genetics counseling should be made when a positive, abnormal or equivocal genetics test result is obtained.

Priority Health Medical Policy developed in cooperation with Spectrum Health Department of Genetics: Helga Toriello, PhD, FACMG; Cindy R. Bos, MD, PhD; Debra Duquette, MS; Karen Lewis, MS.

II. MEDICAL NECESSITY REVIEW

- ☒ Required as defined in above sections. All tests performed at non-participating laboratories will require prior authorization.

This policy reflects the recommendations of the Technology Assessment Committee reviews completed March 3, 2006, June 9, 2006 and September 9, 2011.

III. APPLICATION TO PRODUCTS:

Coverage is subject to member's specific benefits. Group specific policy will supersede this policy when applicable.

- ❖ **HMO/EPO:** *This policy applies to insured HMO/EPO plans.*
- ❖ **POS:** *This policy applies to insured POS plans.*
- ❖ **PPO:** *This policy applies to insured PPO plans. Consult individual plan documents as state mandated benefits may apply. If there is a conflict between this policy and a plan document, the provisions of the plan document will govern.*
- ❖ **ASO:** *For self-funded plans, consult individual plan documents. If there is a conflict between this policy and a self-funded plan document, the provisions of the plan document will govern.*
- ❖ **INDIVIDUAL:** *For individual policies, consult the individual insurance policy. If there is a conflict between this medical policy and the individual insurance policy document, the provisions of the individual insurance policy will govern.*
- ❖ **MEDICARE:** *Coverage is determined by the Centers for Medicare and Medicaid Services (CMS); if a coverage determination has not been adopted by CMS, this policy applies.*
- ❖ **MEDICAID:** *If there is a discrepancy between this policy and the Michigan Medicaid Provider Manual and the Michigan Medicaid Fee Schedule, the Michigan Medicaid Provider Manual and the Michigan Medicaid Fee Schedule at: http://www.michigan.gov/mdch/0,1607,7-132-2945_42542_42543_42546_42551-159815--,00.html will govern.*
- ❖ **MICHILD:** *For MICHILD members, this policy will apply unless MICHILD certificate of coverage limits or extends coverage.*

IV. DESCRIPTION

- A. Genetic Counseling*** Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:
 1. Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.



2. Education about inheritance, testing, management, prevention, resources and research.
3. Counseling to promote informed choices and adaptations to the risk or condition.
4. Discussion of the ethical and legal aspects of autonomy, privacy, equity and confidentiality as applied to each individual seeking genetic testing.
5. Psychosocial aspects should be addressed during the pre-test and post-test counseling session surrounding any genetic testing.

**Journal of Genetic Counseling, Vol. 15, April 2006*

Genetic counselors are defined by the plan as American Board of Medical Genetics or American Board of Genetic Counseling certified physicians or masters or doctorate level-trained genetic counseling professionals who have received formal training in genetics and genetic counseling from an accredited institution. It is the genetic counselor's or physician specialist's role to provide information to the individual or family regarding the genetic disorder that will allow them to make an informed decision.

B. Genetic Testing. A genetic test is the analysis of human DNA, RNA, chromosomes, proteins, or certain metabolites in order to detect alterations related to a heritable or acquired disorder. This can be accomplished by directly examining the DNA or RNA that makes up a gene (direct testing), looking at markers co-inherited with a disease-causing gene (linkage testing), assaying certain metabolites (biochemical testing), or examining the chromosomes (cytogenetic testing). Clinical genetic tests are those in which specimens are examined and results reported to the provider or patient for the purpose of diagnosis, prevention or treatment in the care of individual patients.

C. Genetic Screening refers to examining the genes and/or gene products of phenotypically normal or otherwise people to see if they are carriers of an abnormal gene. Carriers usually do not themselves have symptoms related to the gene mutation. Carrier testing is offered to individuals who have family members with a genetic condition, family members of an identified carrier, and individuals in ethnic or racial groups known to have a higher carrier rate for a particular condition.

V. CODING INFORMATION

See also related policies:

91583 *Markers for Digestive Disorders*

91562 *Tumor Markers*

91570 *Pharmacogenomic Testing*

ICD-9 Codes that may support medical necessity

See criteria

CPT/HCPCS Codes

All claims should be submitted using 2012 Molecular Pathology codes when applicable.



Claims for Medicare and Medicaid members should also use the existing codes in the Cytogenetics, Cytopathology, and Molecular Diagnostic categories. Claims for Priority Medicare and Medicaid members will be paid according to the current stacked codes and the new codes will be captured for reporting purposes only.

Molecular Pathology Tier 1

- 81200 ASPA (aspartoacylase) (eg, Canavan disease) gene analysis, common variants (eg, E285A, Y231X)
- 81205 BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (eg, Maple syrup urine disease) gene analysis, common variants (eg, R183P, G278S, E422X)
- 81206 BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; major breakpoint, qualitative or quantitative
- 81207 BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; minor breakpoint, qualitative or quantitative
- 81208 BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; other breakpoint, qualitative or quantitative
- 81209 BLM (Bloom syndrome, RecQ helicase-like) (eg, Bloom syndrome) gene analysis, 2281del6ins7 variant
- 81210 BRAF (v-raf murine sarcoma viral oncogene homolog B1) (eg, colon cancer), gene
- 81211 BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; full sequence analysis and common duplication/deletion variants in BRCA1 (ie, exon 13 del 3.835kb, exon 13 dup 6kb, exon 14-20 del 26kb, exon 22 del 510bp, exon 8-9 del 7.1kb)
- 81212 BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; 185delAG, 5385insC, 6174delT variants
- 81213 BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; uncommon duplication/deletion variants
- 81214 BRCA1 (breast cancer 1) (eg, hereditary breast and ovarian cancer) gene analysis; full sequence analysis and common duplication/deletion variants (ie, exon 13 del 3.835kb, exon 13 dup 6kb, exon 14-20 del 26kb, exon 22 del 510bp, exon 8-9 del 7.1kb)
- 81215 BRCA1 (breast cancer 1) (eg, hereditary breast and ovarian cancer) gene analysis; known familial variant
- 81216 BRCA2 (breast cancer 2) (eg, hereditary breast and ovarian cancer) gene analysis; full sequence analysis
- 81217 BRCA2 (breast cancer 2) (eg, hereditary breast and ovarian cancer) gene analysis; known familial variant
- 81220 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)
- 81221 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; known familial variants
- 81222 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; duplication/deletion variants
- 81223 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; full gene sequence
- 81224 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; intron 8 poly-T analysis (eg, male infertility)
- 81228 Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (eg, Bacterial Artificial Chromosome



- [BAC] or oligo-based comparative genomic hybridization [CGH] microarray analysis)
- 81229 Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants for chromosomal abnormalities 81241 F5 (coagulation Factor V) (eg, hereditary hypercoagulability) gene analysis, Leiden variant
- 81242 FANCC (Fanconi anemia, complementation group C) (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A>T)
- 81243 FMR1 (Fragile X mental retardation 1) (eg, fragile X mental retardation) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
- 81244 FMR1 (Fragile X mental retardation 1) (eg, fragile X mental retardation) gene analysis; characterization of alleles (eg, expanded size and methylation status)
- 81245 FLT3 (fms-related tyrosine kinase 3) (eg, acute myeloid leukemia), gene analysis, internal tandem duplication (ITD) variants (ie, exons 14, 15)
- 81250 G6PC (glucose-6-phosphatase, catalytic subunit) (eg, Glycogen storage disease, Type 1a, von Gierke disease) gene analysis, common variants (eg, R83C, Q347X)
- 81251 GBA (glucosidase, beta, acid) (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+1G>A)
- 81255 HEXA (hexosaminidase A [alpha polypeptide]) (eg, Tay-Sachs disease) gene analysis, common variants (eg, 1278insTATC, 1421+1G>C, G269S)
- 81256 HFE (hemochromatosis) (eg, hereditary hemochromatosis) gene analysis, common variants (eg, C282Y, H63D)
- 81257 HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis, for common deletions or variant (eg, Southeast Asian, Thai, Filipino, Mediterranean, alpha3.7, alpha4.2, alpha20.5, and Constant Spring)
- 81260 IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (eg, familial dysautonomia) gene analysis, common variants (eg, 2507+6T>C, R696P)
- 81261 IGH@ (Immunoglobulin heavy chain locus) (eg, leukemias and lymphomas, B-cell), gene rearrangement analysis to detect abnormal clonal population(s); amplified methodology (eg, polymerase chain reaction)
- 81262 IGH@ (Immunoglobulin heavy chain locus) (eg, leukemias and lymphomas, B-cell), gene rearrangement analysis to detect abnormal clonal population(s); direct probe methodology (eg, Southern blot)
- 81263 IGH@ (Immunoglobulin heavy chain locus) (eg, leukemia and lymphoma, B-cell), variable region somatic mutation analysis
- 81264 IGK@ (Immunoglobulin kappa light chain locus) (eg, leukemia and lymphoma, B-cell), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)
- 81265 Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing, or maternal cell contamination of fetal cells)
- 81266 Comparative analysis using Short Tandem Repeat (STR) markers; each additional specimen (eg, additional cord blood donor, additional fetal samples from different cultures, or additional zygosity in multiple birth pregnancies) (List separately in addition to code for primary procedure)



- 81267 Chimerism (engraftment) analysis, post transplantation specimen (eg, hematopoietic stem cell), includes comparison to previously performed baseline analyses; without cell selection
- 81268 Chimerism (engraftment) analysis, post transplantation specimen (eg, hematopoietic stem cell), includes comparison to previously performed baseline analyses; with cell selection (eg, CD3, CD33), each cell type
- 81270 JAK2 (Janus kinase 2) (eg, myeloproliferative disorder) gene analysis, p.Val617Phe (V617F) variant
- 81275 KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene) (eg, carcinoma) gene analysis, variants in codons 12 and 13
- 81280 Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); full sequence analysis
- 81281 Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); known familial sequence variant
- 81282 Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); duplication/deletion variants
- 81290 MCOLN1 (mucolipin 1) (eg, Mucopolipidosis, type IV) gene analysis, common variants (eg, IVS3-2A>G, del6.4kb)
- 81291 MTHFR (5,10-methylenetetrahydrofolate reductase) (eg, hereditary hypercoagulability) gene analysis, common variants (eg, 677T, 1298C)
- 81292 MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
- 81293 MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants
- 81294 MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants
- 81295 MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
- 81296 MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants
- 81297 MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants
- 81298 MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
- 81299 MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants
- 81300 MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants
- 81301 Microsatellite instability analysis (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) of markers for mismatch repair deficiency (eg, BAT25, BAT26), includes comparison of neoplastic and normal tissue, if performed
- 81302 MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; full sequence analysis



- 81303 MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; known familial variant
- 81304 MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; duplication/deletion variants
- 81310 NPM1 (nucleophosmin) (eg, acute myeloid leukemia) gene analysis, exon 12 variants
- 81315 PML/RARalpha, (t(15;17)), (promyelocytic leukemia/retinoic acid receptor alpha) (eg, promyelocytic leukemia) translocation analysis; common breakpoints (eg, intron 3 and intron 6), qualitative or quantitative
- 81316 PML/RARalpha, (t(15;17)), (promyelocytic leukemia/retinoic acid receptor alpha) (eg, promyelocytic leukemia) translocation analysis; single breakpoint (eg, intron 3, intron 6 or exon 6), qualitative or quantitative
- 81317 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
- 81318 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants
- 81319 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants
- 81330 SMPD1(sphingomyelin phosphodiesterase 1, acid lysosomal) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330)
- 81331 SNRPN/UBE3A (small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A) (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis
- 81332 SERPINA1 (serpin peptidase inhibitor, clade A, alpha-1 antiproteinase, antitrypsin, member 1) (eg, alpha-1-antitrypsin deficiency), gene analysis, common variants (eg, *S and *Z)
- 81340 TRB@ (T cell antigen receptor, beta) (eg, leukemia and lymphoma), gene rearrangement analysis to detect abnormal clonal population(s); using amplification methodology (eg, polymerase chain reaction)
- 81341 TRB@ (T cell antigen receptor, beta) (eg, leukemia and lymphoma), gene rearrangement analysis to detect abnormal clonal population(s); using direct probe methodology (eg, Southern blot)
- 81342 TRG@ (T cell antigen receptor, gamma) (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)
- 81350 UGT1A1 (UDP glucuronosyltransferase 1 family, polypeptide A1) (eg, irinotecan metabolism), gene analysis, common variants (eg, *28, *36, *37)
- 81355 VKORC1 (vitamin K epoxide reductase complex, subunit 1) (eg, warfarin metabolism), gene analysis, common variants (eg, -1639/3673)
- 81370 HLA Class I and II typing, low resolution (eg, antigen equivalents); HLA-A, -B, -C, -DRB1/3/4/5, and -DQB1
- 81371 HLA Class I and II typing, low resolution (eg, antigen equivalents); HLA-A, -B, and -DRB1/3/4/5 (eg, verification typing)
- 81372 HLA Class I typing, low resolution (eg, antigen equivalents); complete (ie, HLA-A, -B, and -C)
- 81373 HLA Class I typing, low resolution (eg, antigen equivalents); 1 locus (eg, HLA-A, -B, or -C), each
- 81374 HLA Class I typing, low resolution (eg, antigen equivalents); 1 antigen equivalent (eg, B*27), each



- 81375 HLA Class II typing, low resolution (eg, antigen equivalents); HLA-DRB1/3/4/5 and -DQB1
- 81376 HLA Class II typing, low resolution (eg, antigen equivalents); 1 locus (eg, HLA-DRB1/3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1), each
- 81377 HLA Class II typing, low resolution (eg, antigen equivalents); 1 antigen equivalent, each
- 81378 HLA Class I and II typing, high resolution (ie, alleles or allele groups), HLA-A, -B, -C, and -DRB1
- 81379 HLA Class I typing, high resolution (ie, alleles or allele groups); complete (ie, HLA-A, -B, and -C)
- 81380 HLA Class I typing, high resolution (ie, alleles or allele groups); 1 locus (eg, HLA-A, -B, or -C), each
- 81381 HLA Class I typing, high resolution (ie, alleles or allele groups); 1 allele or allele group (eg, B*57:01P), each
- 81382 HLA Class II typing, high resolution (ie, alleles or allele groups); 1 locus (eg, HLA-DRB1, -DRB3, -DRB4, -DRB5, -DQB1, -DQA1, -DPB1, or -DPA1), each
- 81383 HLA Class II typing, high resolution (ie, alleles or allele groups); 1 allele or allele group (eg, HLA-DQB1*06:02P), each

Molecular Pathology Tier 2

- 81400 Molecular pathology procedure, Level 1 (eg, identification of single germline variant [eg, SNP] by techniques such as restriction enzyme digestion or melt curve analysis) ACADM (acyl-CoA dehydrogenase, C-4 to C-12 straight chain, MCAD) (eg, medium chain acyl dehydrogenase deficiency), K304E variant ACE (angiotensin converting enzyme) (eg, hereditary blood pressure regulation), insertion/deletion variant AGTR1 (angiotensin II receptor, type 1) (eg, essential hypertension), 1166A>C variant CCR5 (chemokine C-C motif receptor 5) (eg, HIV resistance), 32-bp deletion mutation/794 825del32 deletion DPYD (dihydropyrimidine dehydrogenase) (eg, 5-fluorouracil/5-FU and capecitabine drug metabolism), IVS14+1G>A variant F2 (coagulation factor 2) (eg, hereditary hypercoagulability), 1199G>A variant F5 (coagulation factor V) (eg, hereditary hypercoagulability), HR2 variant F7 (coagulation factor VII [serum prothrombin conversion accelerator]) (eg, hereditary hypercoagulability), R353Q variant F13B (coagulation factor XIII, B polypeptide) (eg, hereditary hypercoagulability), V34L variant FGB (fibrinogen beta chain) (eg, hereditary ischemic heart disease), -455G>A variant Human Platelet Antigen 1 genotyping (HPA-1), ITGB3 (integrin, beta 3 [platelet glycoprotein IIIa], antigen CD61 [GPIIIa]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-1a/b (L33P) Human Platelet Antigen 2 genotyping (HPA-2), GP1BA (glycoprotein Ib [platelet], alpha polypeptide [GPIba]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-2a/b (T145M) Human Platelet Antigen 3 genotyping (HPA-3), ITGA2B (integrin, alpha 2b [platelet glycoprotein IIb of IIb/IIIa complex], antigen CD41 [GPIIb]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-3a/b (I843S) Human Platelet Antigen 4 genotyping (HPA-4), ITGB3 (integrin, beta 3 [platelet glycoprotein IIIa], antigen CD61 [GPIIIa]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-4a/b (R143Q) Human Platelet Antigen 5 genotyping (HPA-5), ITGA2 (integrin, alpha 2 [CD49B, alpha 2 subunit of VLA-2 receptor] [GPIa]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-5a/b (K505E) Human Platelet Antigen 6 genotyping (HPA-6w), ITGB3 (integrin,



- beta 3 [platelet glycoprotein IIIa, antigen CD61] [GPIIIa] (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-6a/b (R489Q) Human Platelet Antigen 9 genotyping (HPA-9w), ITGA2B (integrin, alpha 2b [platelet glycoprotein IIb of IIb/IIIa complex, antigen CD41] [GPIIb]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-9a/b (V837M) Human Platelet Antigen 15 genotyping (HPA-15), CD109 (CD109 molecule) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-15a/b(S682Y) SERPINE1 (serpine peptidase inhibitor clade E, member 1, plasminogen activator inhibitor -1, PAI-1) (eg, thrombophilia), 4G variant
- 81401 Molecular pathology procedure, Level 2 (eg, 2-10 SNPs, 1 methylated variant, or 1 somatic variant [typically using nonsequencing target variant analysis], or detection of a dynamic mutation disorder/triplet repeat) ABL (c-abl oncogene 1, receptor tyrosine kinase) (eg, acquired imatinib resistance), T315I variant ACADM (acyl-CoA dehydrogenase, C-4 to C-12 straight chain, MCAD) (eg, medium chain acyl dehydrogenase deficiency), common variants (eg, K304E, Y42H) ADRB2 (adrenergic beta-2 receptor surface) (eg, drug metabolism), common variants (eg, G16R, Q27E) APOE (apolipoprotein E) (eg, hyperlipoproteinemia type III, cardiovascular disease, Alzheimer disease), common variants (eg, *2, *3, *4) CFBF/MYH11 (inv(16)) (eg, acute myeloid leukemia), qualitative, and quantitative, if performed CCND1/IGH (BCL1/IgH, t(11;14)) (eg, mantle cell lymphoma) translocation analysis, major breakpoint, qualitative, and quantitative, if performed CFH/ARMS2 (complement factor H/age-related maculopathy susceptibility 2) (eg, macular degeneration), common variants (eg, Y402H [CFH], A69S [ARMS2]) CYP3A4 (cytochrome P450, family 3, subfamily A, polypeptide 4) (eg, drug metabolism), common variants (eg, *2, *3, *4, *5, *6) CYP3A5 (cytochrome P450, family 3, subfamily A, polypeptide 5) (eg, drug metabolism), common variants (eg, *2, *3, *4, *5, *6) DMPK (dystrophia myotonica-protein kinase) (eg, myotonic dystrophy, type 1), evaluation to detect abnormal (eg, expanded) alleles F11 (coagulation factor XI) (eg, coagulation disorder), common variants (eg, E117X [Type II], F283L [Type III], IVS14del14, and IVS14+1G>A [Type I]) FGFR3 (fibroblast growth factor receptor 3) (eg, achondroplasia), common variants (eg, 1138G>A, 1138G>C) FIP1L1/PDGFR4 (del[4q12]) (eg, imatinib-sensitive chronic eosinophilic leukemia), qualitative, and quantitative, if performed GALT (galactose-1-phosphate uridylyltransferase) (eg, galactosemia), common variants (eg, Q188R, S135L, K285N, T138M, L195P, Y209C, IVS2-2A>G, P171S, del5kb, N314D, L218L/N314D) HBB (hemoglobin, beta) (eg, sickle cell anemia, hemoglobin C, hemoglobin E), common variants (eg, HbS, HbC, HbE) HTT (huntingtin) (eg, Huntington disease), evaluation to detect abnormal (eg, expanded) alleles RUNX1/RUNX1T1 (t(8;21)) (eg, acute myeloid leukemia) translocation analysis, qualitative, and quantitative, if performed SEPT9 (Septin 9) (eg, colon cancer), methylation analysis TPMT (thiopurine S-methyltransferase) (eg, drug metabolism), common variants (eg, *2, *3) VWF (von Willebrand factor) (eg, von Willebrand disease type 2N), common variants (eg, T791M, R816W, R854Q)
- 81402 Molecular pathology procedure, Level 3 (eg, > 10 SNPs, 2-10 methylated variants, or 2-10 somatic variants [typically using non-sequencing target variant analysis], immunoglobulin and T-cell receptor gene rearrangements, duplication/deletion variants 1 exon) CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide 2) (eg, congenital adrenal hyperplasia, 21-hydroxylase deficiency), common variants (eg, IVS2-13G, P30L, I172N, exon 6 mutation cluster [I235N, V236E,



- M238K], V281L, L307FfsX6, Q318X, R356W, P453S, G110VfsX21, 30-kb deletion variant) ESR1/PGR (receptor 1/progesterone receptor) ratio (eg, breast cancer) KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (eg, mastocytosis), common variants (eg, D816V, D816Y, D816F) MEFV (Mediterranean fever) (eg, familial Mediterranean fever), common variants (eg, E148Q, P369S, F479L, M680I, I692del, M694V, M694I, K695R, V726A, A744S, R761H) MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (eg, myeloproliferative disorder), common variants (eg, W515A, W515K, W515L, W515R) TRD@ (T cell antigen receptor, delta) (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population
- 81403 Molecular pathology procedure, Level 4 (eg, analysis of single exon by DNA sequence analysis, analysis of > 10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons) ABL1 (c-abl oncogene 1, receptor tyrosine kinase) (eg, acquired imatinib tyrosine kinase inhibitor resistance), variants in the kinase domain DAZ/SRY (deleted in azoospermia and sex determining region Y) (eg, male infertility), common deletions (eg, AZFa, AZFb, AZFc, AZFd) GJB1 (gap junction protein, beta 1) (eg, Charcot-Marie-Tooth X-linked), full gene sequence JAK2 (Janus kinase 2) (eg, myeloproliferative disorder), exon 12 sequence and exon 13 sequence, if performed KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene) (eg, carcinoma), gene analysis, variant(s) in exon 2 MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (eg, myeloproliferative disorder), exon 10 sequence VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), deletion/duplication analysis VWF (von Willebrand factor) (eg, von Willebrand disease types 2A, 2B, 2M), targeted sequence analysis (eg, exon 28)
- 81404 Molecular pathology procedure, Level 5 (eg, analysis of 2-5 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 6-10 exons, or characterization of a dynamic mutation disorder/triplet repeat by Southern blot analysis) BTBD9 (biotinidase) (eg, biotinidase deficiency), full gene sequence CYP11B1 (cytochrome P450, family 1, subfamily B, polypeptide 1) (eg, primary congenital glaucoma), full gene sequence DMPK (dystrophia myotonica-protein kinase) (eg, myotonic dystrophy type 1), characterization of abnormal (eg, expanded) alleles EGR2 (early growth response 2) (eg, Charcot-Marie-Tooth), full gene sequence FKBP1 (Fukutin related protein) (eg, congenital muscular dystrophy type 1C [MDC1C], limb-girdle muscular dystrophy [LGMD] type 2I), full gene sequence FOXG1 (forkhead box G1) (eg, Rett syndrome), full gene sequence FSHMD1A (facioscapulohumeral muscular dystrophy 1A) (eg, facioscapulohumeral muscular dystrophy), evaluation to detect abnormal (eg, deleted) alleles FSHMD1A (facioscapulohumeral muscular dystrophy 1A) (eg, facioscapulohumeral muscular dystrophy), characterization of haplotype(s) (ie, chromosome 4A and 4B haplotypes) HBB (hemoglobin, beta, Beta-Globin) (eg, thalassemia), full gene sequence KIT (C-kit) (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (eg, GIST, acute myeloid leukemia, melanoma), targeted gene analysis (eg, exons 8, 11, 13, 17, 18) LITAF (lipopolysaccharide-induced TNF factor) (eg, Charcot-Marie-Tooth), full gene sequence MEFV (Mediterranean fever) (eg, familial Mediterranean fever), full gene sequence NRAS (neuroblastoma RAS viral oncogene homolog) (eg, colorectal carcinoma), exon 1 and exon 2 sequences PDGFRA (platelet-derived growth factor receptor alpha polypeptide) (eg,



- gastrointestinal stromal tumor), targeted sequence analysis (eg, exons 12, 18) RET (ret proto-oncogene) (eg, multiple endocrine neoplasia, type 2B and familial medullary thyroid carcinoma), common variants (eg, M918T, 2647_2648delinsTT, A883F) SDHD (succinate dehydrogenase complex, subunit D, integral membrane protein) (eg, hereditary paraganglioma), full gene sequence VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), full gene sequence VWF (von Willebrand factor) (eg, von Willebrand disease type 1C), targeted sequence analysis (eg, exons 26, 27, 37)
- 81405 Molecular pathology procedure, Level 6 (eg, analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons) CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide2) (eg, steroid 21-hydroxylase isoform, congenital adrenal hyperplasia), full gene sequence FKTN (fukutin) (eg, limb-girdle muscular dystrophy [LGMD] type 2M or 2L), full gene sequence MPZ (myelin protein zero) (eg, Charcot-Marie-Tooth), full gene sequence NEFL (neurofilament, light polypeptide) (eg, Charcot-Marie-Tooth), full gene sequence RET (ret proto-oncogene) (eg, multiple endocrine neoplasia, type 2A and familial medullary thyroid carcinoma), targeted sequence analysis (eg, exons 10, 11, 13-16) SDHB (succinate dehydrogenase complex, subunit B, iron sulfur) (eg, hereditary paraganglioma), full gene sequence TGFBR1 (transforming growth factor, beta receptor 1) (eg, Marfan syndrome), full gene sequence TGFBR2 (transforming growth factor, beta receptor 2) (eg, Marfan syndrome), full gene sequence THRB (thyroid hormone receptor, beta) (eg, thyroid hormone resistance, thyroid hormone beta receptor deficiency), full gene sequence or targeted sequence analysis of >5 exons TP53 (tumor protein 53) (eg, Li-Fraumeni syndrome, tumor samples), full gene sequence or targeted sequence analysis of >5 exons VWF (von Willebrand factor) (eg, von Willebrand disease type 2N), targeted sequence analysis (eg, exons 18-20, 23-25)
- 81406 Molecular pathology procedure, Level 7 (eg, analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons, cytogenomic array analysis for neoplasia) CAPN3 (Calpain 3) (eg, limb-girdle muscular dystrophy [LGMD] type 2A, calpainopathy), full gene sequence Cytogenomic microarray analysis, neoplasia (eg, interrogation of copy number, and loss-of-heterozygosity via single nucleotide polymorphism [SNP]-based comparative genomic hybridization [CGH] microarray analysis) GALT (galactose-1-phosphate uridylyltransferase) (eg, galactosemia), full gene sequence HEXA (hexosaminidase A, alpha polypeptide) (eg, Tay-Sachs disease), full gene sequence LMNA (lamin A/C) (eg, Emery-Dreifuss muscular dystrophy [EDMD1, 2 and 3] limb-girdle muscular dystrophy [LGMD] type 1B, dilated cardiomyopathy [CMD1A], familial partial lipodystrophy [FPLD2]), full gene sequence PAH (phenylalanine hydroxylase) (eg, phenylketonuria), full gene sequence POLG (polymerase [DNA directed], gamma) (eg, Alpers-Huttenlocher syndrome, autosomal dominant progressive external ophthalmoplegia), full gene sequence POMGNT1 (protein O-linked mannose beta1,2-N acetylglucosaminyltransferase) (eg, muscle-eye-brain disease, Walker-Warburg syndrome), full gene sequence POMT1 (protein-O-mannosyltransferase 1) (eg, limb-girdle muscular dystrophy [LGMD] type 2K, Walker-Warburg syndrome), full gene sequence POMT2 (protein-O-mannosyltransferase 2) (eg, limb-girdle muscular dystrophy [LGMD] type 2N, Walker-Warburg syndrome), full gene sequence RYR1 (ryanodine receptor 1, skeletal) (eg, malignant hyperthermia), targeted sequence analysis of exons with functionally confirmed mutations VWF (von Willebrand factor) (von Willebrand



- disease type 2A), extended targeted sequence analysis (eg, exons 11-16, 24-26, 51, 52)
- 81407 Molecular pathology procedure, Level 8 (eg, analysis of 26-50 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of > 50 exons, sequence analysis of multiple genes on 1 platform) SCN1A (sodium channel, voltage-gated, type 1, alpha subunit) (eg, generalized epilepsy with febrile seizures), full gene sequence
- 81408 Molecular pathology procedure, Level 9 (eg, analysis of > 50 exons in a single gene by DNA sequence analysis) FBN1 (fibrillin 1) (eg, Marfan syndrome), full gene sequence NF1 (neurofibromin 1) (eg, neurofibromatosis, type 1), full gene sequence RYR1 (ryanodine receptor 1, skeletal) (eg, malignant hyperthermia), full gene sequence VWF (von Willebrand factor) (eg, von Willebrand disease types 1 and 3), full gene sequence

Genetic Testing - Molecular Diagnostics

- 83890 Molecular diagnostics; molecular isolation or extraction
- 83891 Molecular diagnostics; isolation or extraction of highly purified nucleic acid
- 83892 Molecular diagnostics; enzymatic digestion
- 83893 Molecular diagnostics; dot/slot blot production
- 83894 Molecular diagnostics; separation by gel electrophoresis (eg, agarose, polyacrylamide)
- 83896 Molecular diagnostics; nucleic acid probe, each
- 83897 Molecular diagnostics; nucleic acid transfer (eg, Southern, Northern)
- 83898 Molecular diagnostics; amplification of patient nucleic acid, each nucleic acid sequence
- 83900 Molecular diagnostics; amplification of patient nucleic acid, multiplex, first two nucleic acid sequences
- 83901 Molecular diagnostics; amplification of patient nucleic acid, multiplex, each additional nucleic acid sequence (List separately in addition to code for primary procedure)
- 83902 Molecular diagnostics; reverse transcription
- 83903 Molecular diagnostics; mutation scanning, by physical properties (eg, single strand conformational polymorphisms (SSCP), heteroduplex, denaturing gradient gel electrophoresis (DGGE), RNA'ase A), single segment, each
- 83904 Molecular diagnostics; mutation identification by sequencing, single segment, each segment
- 83905 Molecular diagnostics; mutation identification by allele specific transcription, single segment, each segment
- 83906 Molecular diagnostics; mutation identification by allele specific translation, single segment, each segment
- 83907 Molecular diagnostics; lysis of cells prior to nucleic acid extraction (eg, stool specimens, paraffin embedded tissue)
- 83908 Molecular diagnostics; signal amplification of patient nucleic acid, each nucleic acid sequence
- 83909 Molecular diagnostics; separation and identification by high resolution technique (eg, capillary electrophoresis)
- 83912 Molecular diagnostics; interpretation and report
- 83913 Molecular diagnostics; RNA stabilization



- 83914 Mutation identification by enzymatic ligation or primer extension, single segment, each segment (eg, oligonucleotide ligation assay (OLA), single base chain extension (SBCE), or allele-specific primer extension (ASPE))
- 83950 Oncoprotein; HER-2/neu
- 84999 Unlisted chemistry procedure

Cytopathology

- 88184 Flow cytometry, cell surface, cytoplasmic, or nuclear marker, technical component only; first marker
- 88185 Flow cytometry, cell surface, cytoplasmic, or nuclear marker, technical component only; each additional marker (List separately in addition to code for first marker)
- 88187 Flow cytometry, interpretation; 2 to 8 markers
- 88188 Flow cytometry, interpretation; 9 to 15 markers
- 88189 Flow cytometry, interpretation; 16 or more markers

Genetic Testing - Cytogenetics

- 88230 Tissue culture for non-neoplastic disorders; lymphocyte
- 88233 Tissue culture for non-neoplastic disorders; skin or other solid tissue biopsy
- 88235 Tissue culture for non-neoplastic disorders; amniotic fluid or chorionic villus cells
- 88237 Tissue culture for neoplastic disorders; bone marrow, blood cells
- 88239 Tissue culture for neoplastic disorders; solid tumor
- 88240 Cryopreservation, freezing and storage of cells, each cell line
- 88241 Thawing and expansion of frozen cells, each aliquot
- 88245 Chromosome analysis for breakage syndromes; baseline Sister Chromatid Exchange (SCE), 20-25 cells
- 88248 Chromosome analysis for breakage syndromes; baseline breakage, score 50-100 cells, count 20 cells, 2 karyotypes (eg, for ataxia telangiectasia, Fanconi anemia, fragile X)
- 88249 Chromosome analysis for breakage syndromes; score 100 cells, clastogen stress (eg, diepoxybutane, mitomycin C, ionizing radiation, UV radiation)
- 88261 Chromosome analysis; count 5 cells, 1 karyotype, with banding
- 88262 Chromosome analysis; count 15-20 cells, 2 karyotypes, with banding
- 88263 Chromosome analysis; count 45 cells for mosaicism, 2 karyotypes, with banding
- 88264 Chromosome analysis; analyze 20-25 cells
- 88267 Chromosome analysis, amniotic fluid or chorionic villus, count 15 cells, 1 karyotype, with banding
- 88269 Chromosome analysis, in situ for amniotic fluid cells, count cells from 6-12 colonies, 1 karyotype, with banding
- 88271 Molecular cytogenetics; DNA probe, each (eg, FISH)
- 88272 Molecular cytogenetics; chromosomal in situ hybridization, analyze 3-5 cells (eg, for derivatives and markers)
- 88273 Molecular cytogenetics; chromosomal in situ hybridization, analyze 10-30 cells (eg, for microdeletions)
- 88274 Molecular cytogenetics; interphase in situ hybridization, analyze 25-99 cells
- 88275 Molecular cytogenetics; interphase in situ hybridization, analyze 100-300 cells
- 88280 Chromosome analysis; additional karyotypes, each study
- 88283 Chromosome analysis; additional specialized banding technique (eg, NOR, C-banding)
- 88285 Chromosome analysis; additional cells counted, each study



- 88289 Chromosome analysis; additional high resolution study
88291 Cytogenetics and molecular cytogenetics, interpretation and report
88299 Unlisted cytogenetic study

Surgical Pathology

- 88360 Morphometric analysis, tumor immunohistochemistry (eg, Her-2/neu, estrogen receptor/progesterone receptor), quantitative or semiquantitative, each antibody; manual
88361 Morphometric analysis, tumor immunohistochemistry (eg, Her-2/neu, estrogen receptor/progesterone receptor), quantitative or semiquantitative, each antibody; using computer-assisted technology
88384 Array-based evaluation of multiple molecular probes; 11 through 50 probes
88385 Array-based evaluation of multiple molecular probes; 51 through 250 probes
88386 Array-based evaluation of multiple molecular probes; 251 through 500 probes

89290 Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); less than or equal to 5 embryos
89291 Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); greater than 5 embryos

88299 Unlisted Cytogenic Study (*Explanatory notes must accompany claim*)
84999 Unlisted chemistry procedure (*Explanatory notes must accompany claim*)

Nuccal Translucency Measurements

- 76813 Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; single or first gestation
76814 Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; each additional gestation (List separately in addition to code for primary procedure)
84163 Pregnancy-associated plasma protein-A (PAPP-A)
84704 Gonadotropin, chorionic (hCG); free beta chain

Gene Expression Analysis (Oncotype Dx only)

("S" code not payable for Priority Health Medicare or Priority Health Medicaid)

- S3854 Gene expression profiling panel for use in the management of breast cancer treatment
84999 Unlisted chemistry procedure (*Explanatory notes must accompany claim*)

AlloMap™ Molecular Expression Test

- 84999 Unlisted chemistry procedure (*Explanatory notes must accompany claim*)
86849 Unlisted immunology procedure (*Explanatory notes must accompany claim*)
88299 Unlisted cytogenetic study (*Explanatory notes must accompany claim*)

Genetic Counseling

- 96040 Medical genetics and genetic counseling services, each 30 minutes face-to-face with patient/family
S0265 Genetic counseling, under physician supervision, each 15 minutes

Not Covered

S3852	DNA analysis for APOE epsilon 4 allele for susceptibility to Alzheimer's disease
S3855	Genetic testing for detection of mutations in the presenilin, 1 gene
S3890	DNA analysis, fecal, for colorectal cancer screening

VI. REFERENCES

1. *Genetic Testing Medical Policy*, Dean Health Plan, Madison, WI. April 2001
2. *Genetic Counseling/Genetic Testing/ Genetic Screening Medical Policy*, Care Choices, Farmington Hills, MI. December 2000.
3. *Genetic Testing (#0140) and Genetic Counseling (#0189) Medical Policies*, Aetna, July 2001
4. **HAYES technology assessments: First-Trimester Prenatal Screening Using Nuchal translucency Combined with Maternal PAPP-A and Free B-hCG levels.** December 12, 2005
5. First-trimester or second-trimester screening, or both, for Down's syndrome. *N Engl J Med.* 2005 Nov 10;353(19):2001-11.
6. SURUSS in perspective. *BJOG.* 2004 Jun;111(6):521-31
7. Practical strategies in contingent sequential screening for Down syndrome. *Prenat Diagn.* 2005 Aug;25(8):645-52
8. A cost-effectiveness analysis of prenatal screening strategies for Down syndrome. *Obstet Gynecol.* 2005 Sep;106(3):562-8. Erratum in: *Obstet Gynecol.* 2006 Jan;107(1):209
9. Genetics and Public Policy Center. Technical brief: Preimplantation Genetic Diagnosis. Updated June 2005. Accessed January 19, 2006. Available at URL address: <http://www.dnapolicy.org/genetics/pgd.jhtml>
10. Thornhill A, deDie-Smulders C., Geraedts J, Harper J, Harton G, Lavery S, Moutou C, Robinson M, Schmutzler A, Scriven P; Sermon K, Wilton L. ESHRE PGD Consortium 'Best practice guidelines for clinical preimplantation genetic diagnosis (PGD) and preimplantation genetic screening (PGS)'. *Human Reproduction.* 2004 Jan;20(1):35-48
11. Sermon K, Van Steirteghem A, Liebaers I. Preimplantation genetic diagnosis. *Lancet.* 2004 May 15; 363(9421):1633-41.
12. Preimplantation genetics diagnosis international society. Guidelines for good practice in PGD. published in *RBMOnline* 2004; 9(4). Accessed January 19, 2006. Available at URL address: <http://www.pgdis.org/>
13. Cigna healthcare coverage position policy # 0108 revised 7/15/05: accessed January 12, 2006
http://www.cigna.com/health/provider/medical/procedural/coverage_positions/medical/#P
14. Aetna Clinical Policy Bulletins - Prenatal Diagnosis of Genetic Diseases – Policy # 0358 <http://www.aetna.com/cpb/data/CPBA0327.html>
15. The Regence Group – BlueCross/BlueShield- Preimplantation Genetic Diagnosis Policy # 11 <http://www.regence.com/trgmedpol/maternity/mat11.html>
16. A multigene assay to predict recurrence of tamoxifen-treated, node-negative breast cancer. Paik S, Shak S, Tang G, Kim C, Baker J, Cronin M, Baehner FL, Walker



- MG, Watson D, Park T, Hiller W, Fisher ER, Wickerham DL, Bryant J, Wolmark N. N Engl J Med. 2004 Dec 30;351(27):2817-26. Epub 2004 Dec 10
17. Prognostic role of a multigene reverse transcriptase-PCR assay in patients with node-negative breast cancer not receiving adjuvant systemic therapy. Esteva FJ, Sahin AA, Cristofanilli M, Coombes K, Lee SJ, Baker J, Cronin M, Walker M, Watson D, Shak S, Hortobagyi GN. Clin Cancer Res. 2005 May 1;11(9):3315-9.
 18. Gene expression profiling and breast cancer care: What are the potential benefits and policy implications? Oestreicher N, Ramsey SD, Linden HM, McCune JS, Van't Veer LJ, Burke W, Veenstra DL. Genet Med. 2005 Jul-Aug;7(6):380-9
 19. Application of DNA microarray technology in determining breast cancer prognosis and therapeutic response Brennan DJ, O'Brien SL, Fagan A, Culhane AC, Higgins DG, Duffy MJ, Gallagher WM. Expert Opin Biol Ther. 2005 Aug;5(8):1069-83
 20. Prediction of cancer outcome with microarrays: a multiple random validation strategy. Michiels S, Koscielny S, Hill C. Lancet. 2005 Feb 5-11;365(9458):488-92
 21. Detection of stool DNA mutations before and after treatment of colorectal neoplasia. Cancer. 2006 Jan 15;106(2):277-83.
 22. Detection of colorectal cancer by a quantitative fluorescence determination of DNA amplification in stool. Neoplasia. 2004 Sep-Oct;6(5):536-40.
 23. Fecal DNA versus fecal occult blood for colorectal-cancer screening in an average-risk population. N Engl J Med. 2004 Dec 23;351(26):2704-14
 24. Colorectal cancer screening using stool DNA analysis in clinical practice: early clinical experience with respect to patient acceptance and colonoscopic follow-up of abnormal tests. Clin Colorectal Cancer. 2006 Jan;5(5):338-43.
 25. The AmpliChip CYP450 Test: Principles, Challenges, and Future Clinical Utility in Digestive Disease. Clin Gastroenterol Hepatol. 2006 Jul;4(7):822-30. Epub 2006 Jun 22.
 26. Pharmacogenetics in drug regulation: promise, potential and pitfalls. Philos Trans R Soc Lond B Biol Sci. 2005 Aug 29;360(1460):1617-38. Review
 27. Applications of AmpliChip CYP450. Mol Diagn. 2005;9(3):119-27.
 28. GeneTests <http://www.genetests.org> NIH sponsored website accessed 1/10/07.
 29. BlueCross/BlueShield Regence Policy Laboratory Section - Genetic Testing Policy#20 accessed 1/10/07
 30. Aetna Genetic testing policy 0140 accessed 1/10/07
 31. Cigna Genetic Testing policy 0052 accessed 1/10/07
 32. Trepanier A, et al: Genetic cancer risk assessment and counseling: recommendations of the National Society of Genetic Counselors. J Genet Counsel 13:83, 2004.
 33. American Gastroenterological Association medical position statement on the diagnosis and management of celiac disease. Gastroenterology 2006 Dec;131(6):1977-80
 34. Mayo Clinic – Mayo Foundation for medical education and research Celiac Disease Diagnostic testing Algorithm 03/09 <http://www.mayoclinic.com/health/celiac-disease/DS00319/DSECTION=tests%2Dand%2Ddiagnosis> . Accessed 01/2010



35. Up-To-Date: **Genetic counseling and testing.** Last literature review version 19.2: May 2011. Accessed July 24, 2011

AMA CPT Copyright Statement:

All Current Procedure Terminology (CPT) codes, descriptions, and other data are copyrighted by the American Medical Association.

This document is for informational purposes only. It is not an authorization, certification, explanation of benefits, or contract. Receipt of benefits is subject to satisfaction of all terms and conditions of coverage. Eligibility and benefit coverage are determined in accordance with the terms of the member's plan in effect as of the date services are rendered. Priority Health's medical policies are developed with the assistance of medical professionals and are based upon a review of published and unpublished information including, but not limited to, current medical literature, guidelines published by public health and health research agencies, and community medical practices in the treatment and diagnosis of disease. Because medical practice, information, and technology are constantly changing, Priority Health reserves the right to review and update its medical policies at its discretion.

Priority Health's medical policies are intended to serve as a resource to the plan. They are not intended to limit the plan's ability to interpret plan language as deemed appropriate. Physicians and other providers are solely responsible for all aspects of medical care and treatment, including the type, quality, and levels of care and treatment they choose to provide.

The name "Priority Health" and the term "plan" mean Priority Health, Priority Health Managed Benefits, Inc. and Priority Health Government Programs, Inc.



APPENDIX A

Conditions for which Genetic Counseling* is required or recommended prior to genetic testing:

Genetic Counseling Required	Genetic Counseling Recommended
Hereditary Cancer Syndromes: including but not limited to BRCA, HNPCC, FAP, MYH, Cowden Syndrome, Li Fraumeni syndrome, Peutz-Jeghers syndrome, Ataxia Telangiectasia	Prenatal screening: serum screening, sequential screening, etc.
Pre-symptomatic testing: familial conditions including but not limited to Huntington’s Chorea, alzheimer’s disease	Carrier screening: Individuals from ethnic groups recognized to be at increased risk for specific genetic disorders (e.g., African Americans for sickle cell anemia, Ashkenazi Jewish (eastern European) for Tay-Sachs disease)
Pre-implantation diagnosis: Pre-test discussion on conditions to be tested for.	Diagnostic testing: Tests to confirm or rule out suspected genetic conditions in symptomatic individuals in which confirming a diagnosis has the potential to alter the medical management for the individual, i.e., muscular dystrophy, spinal muscular atrophy, microdeletion syndromes.
Prenatal diagnosis: chorionic villus sampling (CVS), amniocentesis	Conditions identified by newborn screening
Carrier testing: testing for carrier status when there is a known or suspected genetic condition in the family (i.e., cystic fibrosis carrier testing, Fragile X pre-mutation carrier testing, etc.)	
Pre and post Testing of at-risk family members for cardiac conditions in which the proband (individual with specific genetic condition has been identified) has an identifiable mutation i.e., Long QT syndrome, hypertrophic cardiomyopathy. Post test counseling for clinically symptomatic individuals.	