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Medicare Advantage Policy Manual **Policy ID:** M-GT64

Genetic and Molecular Diagnostics – Next Generation Sequencing, Genetic Panels, and Biomarker Testing

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IMPORTANT REMINDER

The Medicare Advantage Medical Policy manual is not intended to override the member Evidence of Coverage (EOC), which defines the insured's benefits, nor is it intended to dictate how providers are to practice medicine. Physicians and other health care providers are expected to exercise their medical judgment in providing the most appropriate care for the individual member, including care that may be both medically reasonable and necessary.

The Medicare Advantage medical policies are designed to provide guidance regarding the decision-making process for the coverage or non-coverage of services or procedures in accordance with the member EOC and Centers of Medicare and Medicaid Services (CMS) policies and manuals, along with general CMS rules and regulations. In the event of a conflict, applicable CMS policy or EOC language will take precedence over the Medicare Advantage Medical Policy. In the absence of a specific CMS coverage determination for a requested service, item or procedure, the health plan may apply CMS regulations, as well as their Medical Policy Manual or other applicable utilization management vendor criteria developed with an objective, evidence-based process using scientific evidence, current generally accepted standards of medical practice, and authoritative clinical practice guidelines.

Some services or items may appear to be medically indicated for an individual but they may also be a direct exclusion of Medicare or the member's benefit plan. Medicare and member EOCs exclude from coverage, among other things, services or procedures considered to be investigational (experimental) or cosmetic, as well as services or items considered not medically reasonable and necessary under Title XVIII of the Social Security Act, §1862(a)(1)(A). In some cases, providers may bill members for these non-covered services or procedures. Providers are encouraged to inform members in advance when they may be financially responsible for the cost of non-covered or excluded services. Members, their appointed representative, or a treating provider can request coverage of a service or item by submitting a pre-service organization determination prior to services being rendered.

DESCRIPTION

Genetic testing may be done for several purposes, including but not limited to, diagnosing or predicting susceptibility for inherited conditions, determining carrier status, diagnostic and prognostic testing, or risk screening for common disorders. Panel tests using next generation sequencing (NGS) technology as well as other biomarker tests are currently available for many applications.

While genetic and other biomarker testing has potential benefits for certain conditions, there are also risks associated with this testing, including the risk of incorrect results or treatments, as well as emotional, social, or financial risks. Test results may cause feelings of depression, anger, guilt or anxiety, or they reveal information about other family members who did not provide consent for testing. For some conditions, genetic and biomarker tests can provide only limited information. Such tests may not provide clear answers about whether a person will ever show symptoms of a disorder, how severe the symptoms will be, or whether the disorder will progress over time. Another limitation of genetic and biomarker testing is that there may not be treatments or cures available for conditions related to the testing. Candidates for genetic and

biomarker testing may wish to consult with a genetics professional who can explain in detail the benefits, risks, and limitations of a particular test.

Some genetic and biomarker tests may be eligible for general Medicare coverage, while others may only be covered in select individuals or for certain conditions, and still others may not be eligible for coverage at all, due to the nature of the Medicare program and the applicable requirements for reasonable and necessary services and diagnostic laboratory testing coverage.

NOTE: See the "Policy Guidelines" below for important notes regarding Medicare and diagnostic laboratory and genetic testing services.

MEDICARE ADVANTAGE POLICY CRITERIA

Note: This policy provides information regarding Medicare local carrier jurisdiction, Medicare guidance for coverage of specific genetic tests, as well as information on coverage for certain *types* or *categories* of genetic and biomarker tests.

I. See Table 2 for a list of tests or types of tests with known Medicare coverage or non-coverage guidance. Some tests are never considered medically reasonable or necessary, while others have criteria which must be met for the genetic test to be covered.

   A. Note, the genes and codes included in the tables are provided as a courtesy. Individual laboratories may choose to use different coding, and gene lists are subject to change.

   B. Some small panel tests may be reviewed by gene. If a panel test is not found in this Medicare Advantage Medical Policy, but all of the individual genes are addressed, the coverage decisions from the single gene policy for each individual gene may be applied if the applicable references are appropriate for the performing laboratory's service area.

II. Additional research may be necessary to determine coverage for tests that are not listed in Table 2, or when testing is performed in a geographical area that is not addressed.

Table 1: MolDX Program and Medicare Jurisdictions

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Medicare jurisdictions which have adopted the MolDX Program are indicated below (MolDX site). If the performing laboratory is not located in one of the marked states, MolDX guidelines should not apply. Other Medicare guidance may be available.

STATE	MolDX	STATE	MolDX	STATE	MolDX
Alabama	X	Alaska	X	Arizona	X
Arkansas		California	X	Colorado	
Connecticut		Delaware		Florida	
Georgia	X	Hawaii	X	Idaho	X
Illinois		Indiana	X	Iowa	X
Kansas	X	Kentucky	X	Louisiana	
Maine		Maryland		Massachusetts	

STATE	MolDX	STATE	MolDX	STATE	MolDX
Michigan	X	Minnesota		Mississippi	
Missouri	X	Montana	X	Nebraska	X
Nevada	X	New Hampshire		New Jersey	
New Mexico		New York		North Carolina	X
North Dakota	X	Ohio	X	Oklahoma	
Oregon	X	Pennsylvania		Rhode Island	
South Carolina	X	South Dakota	X	Tennessee	X
Texas		Utah	X	Vermont	
Virginia	X	Washington	X	West Virginia	X
Wisconsin		Wyoming			

Table 2: Genetic and Biomarker Tests with Medicare Coverage References

Note: Tests related to medication selection (pharmacogenomics), infections disease pathogen panels, transplant rejection status, cardiovascular risk, and cancer, including cancer risk, diagnosis, prognosis or treatment are addressed in separate policies (see Cross References). The tests listed in this table have known Medicare guidance available. Some tests are considered "not medically necessary," while others may have criteria which must be met for the genetic test to be covered. Please review the "Medicare Rationale/Reference" source carefully.

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database* Back to Criteria
ApoE Genotype (APOE) *(81401)*	For cardiovascular risk assessment or treatment selection for Alzheimer's disease, see Cross References. ***For risk of Alzheimer's disease:*** *Title XVIII of the Social Security Act, Section 1862(a)(1)(A) states " ...no Medicare payment shall be made for items or services which are not reasonable and necessary for the diagnosis and treatment of illness or injury..." (See also Medicare Benefit Policy Manual, Chapter 16 - General Exclusions From Coverage, §20 - Services Not Reasonable and Necessary). APOE genotyping is not a diagnostic test for Alzheimer's disease but instead provides information regarding risk for developing the condition.* Therefore, *APOE* testing to assess risk for developing Alzheimer's disease is a statutory exclusion and considered **not medically necessary**. *Laboratories in IL, MN, WI, CT, NY, ME, MA, NH, RI, VT:* • National Government Services Inc. (NGS) LCD for Molecular Pathology Procedures (L35000) See the guideline specific to the gene within the LCD.
Chimerism/Short Tandem Repeat Marker Testing *(81265, 81266, 81267, 81268)*	*Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY* • Billing and Coding: Short Tandem Repeat (STR) Markers and Chimerism (CPT® codes 81265-81268) (A57843). *The companion LCD L36256 can be accessed from the article.*

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database* Back to Criteria
	Laboratories in CA and NV • Billing and Coding: Short Tandem Repeat (STR) Markers and Chimerism (CPT® codes 81265-81268) (A57842). *The companion LCD L35160 can be accessed from the article.*
Chromosomal Microarray (CMA) and Genomic CNV Testing for Inherited Disorders *(0209U, 81228, 81229)*	*Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • LCD attachment for L36256, Excluded Test List – as of 08/01/2016 *Laboratories in CA and NV:* • LCD attachment for L35160, Excluded Test List – as of 08/01/2016
DYPD, TYMS Testing	See Cross References section for Medicare Advantage medical policy, Laboratory and Genetic Testing for Use of 5-Fluorouracil (5-FU) in Patients with Cancer, Laboratory, Policy No. 64.
Envisia Genomic Classifier *(81554)* Veracyte, Inc., (CA)	MolDX: Envisia, Veracyte, Idiopathic Pulmonary Fibrosis Diagnostic Test (L37887) *Companion article A57419 can be accessed directly from the LCD.*
Fetal congenital abnormality biochemical assays with added Y chromosome test result *and other obstetric predictive-risk tests* Examples: • Maternal Fetal Screen | T1 + Y Chromosome℠ Eurofins NTD, LLC • Preeclampsia Screen | T1 + Y Chromosome℠ Eurofins NTD, LLC • PGIF Preeclampsia Screen (PerkinElmer Genetics, Inc.) (0243U) • PreTRM® (Sera Prognostics) (0247U) • PEPredictDx (OncoOmicsDx) (0390U) • Preeclampsia sFlt-1/PlGF Ratio (PERA) (Mayo Clinic) (0482U)	According to the DEX™ Change Healthcare Registry website, many of these tests are **non-covered** tests by MolDX for Medicare. Biochemical assays for prenatal screening of fetal congenital abnormalities may be considered medically necessary when reported with CPT codes 84163, 84704, 82105, 86336, 83520. Tests with the added component of fetal sex determination (Y chromosome) are considered **not medically necessary** under the *Social Security Act, §1862(a)(1)(A)*. Biochemical assays of analytes (free beta-hCG, PAPP-A, AFP, placental growth factor, and/or inhibin-A) without the Y chromosome testing are not typically reviewed and may be considered medically necessary. To be covered under the Medicare diagnostic laboratory testing benefit, a diagnostic test must (1) not be considered screening (testing in the absence of clinical signs and symptoms of disease) and (2) must be ordered by a physician who is treating the beneficiary and who will use the tests results in the management of a beneficiary's specific medical problem. Some of these tests are performed in the absence of clinical signs/symptoms, and the test results are not used in the management of a beneficiary's specific medical problem. Therefore, these tests are considered **not medically reasonable or necessary** for Medicare.

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database*	Back to Criteria
• sFlt-1/PlGF (Thermofisher) (0524U) **Note:** *Biochemical* assays differ from *chromosomal* genomic sequencing assays reported with 81420, 81422, 81507, etc. See separate rows for chromosomal testing.		
Genomic Unity® Tests *(coding varies)* Variantyx Inc. (MA) Examples: • Genomic Unity® AR Analysis • Genomic Unity® CACNA1A Analysis • Genomic Unity® CSTB Analysis • Genomic Unity® FXN Analysis • Genomic Unity® MECP2 Analysis • Genomic Unity® SMN1/2 Analysis • Genomic Unity® Cardiac Ion Channelopathies Analysis • Genomic Unity® Comprehensive Mitochondrial Disorders Analysis	Billing and Coding: Molecular Pathology Procedures (A56199) *The coverage policy L35000 can be accessed directly from the article.* There are non-coverage decisions for some single gene tests. For the following Genomic Unity tests, apply the same non-coverage rationale that this MAC uses for similar single gene tests: • Genomic Unity® AR Analysis – LCD and article address this individual gene by name (treat the same as CPT codes 81173, 81174, 81204). • Genomic Unity® FXN Analysis – LCD and article address this individual gene by name (treat the same as CPT codes 81284-81289). • Genomic Unity® MECP2 Analysis – LCD and article address this individual gene by name (treat the same as CPT codes 81302-81304). • Genomic Unity® SMN1/2 Analysis – the article addresses this individual gene by name (treat the same as CPT codes 81329, 81336, 81337). • Genomic Unity® Cardiac Ion Channelopathies Analysis - the article addresses similar panel tests (treat the same as CPT codes 81413, 81414) • Genomic Unity® Comprehensive Mitochondrial Disorders Analysis Coverage determination for the following tests requires documentation that clearly states how test results are actionable, and how they will promptly and directly be used for treatment decisions or diagnosis: • Genomic Unity® CACNA1A Analysis • Genomic Unity® CSTB Analysis	
Hereditary cardiomyopathy *other than arrhythmogenic right ventricular cardiomyopathy (81439, 81479)*	*Laboratories in IL, MN, WI, CT, NY, ME, MA, NH, RI, VT:* • Billing and Coding: Molecular Pathology Procedures (A56199) (Search CPT code)	

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database* Back to Criteria
HFE Gene Tests *(81256)*	*Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • MolDX: Molecular Diagnostic Tests (MDT) (L36256) *Companion article A57527 can be accessed directly from the LCD.* *Laboratories in CA and NV:* • MolDX: Molecular Diagnostic Tests (MDT) (L35160) *Companion article A57526 can be accessed directly from the LCD.* These LCDs require that tests complete a technology assessment to determine if a test meets Medicare's reasonable and necessary requirement. Tests that have completed this assessment are listed in the DEX™ registry. In addition, **clinical documentation must demonstrate how test results will be used** in the management or diagnosis of an illness or condition. *Laboratories in IL, MN, WI, CT, NY, ME, MA, NH, RI, VT:* Molecular Pathology Procedures (L35000) *See gene-specific guidance within the LCD. Companion article A56199 can be accessed directly from the LCD.*
Human Leukocyte Antigen (HLA) Typing *(81370-81383) (Note: This is different than testing described by NCD 190.1 and reported with CPT codes 86812-86826)*	***For HLA-A and HLA-B testing for medication selection, see Cross References.*** ***For transplant histocompatibility:*** *Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY, CA, HI, or NV:* • Billing and Coding: MolDX: HLA Testing for Transplant Histocompatibility (A57972) ***HLA-DQB1*06:02 Testing for Narcolepsy (81383)*** *Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • MolDX: HLA-DQB1*06:02 Testing for Narcolepsy (L36544) *Companion article A57527 can be accessed directly from the LCD.* *Laboratories in CA and NV:* • MolDX: HLA-DQB1*06:02 Testing for Narcolepsy (L36551) *Companion article A57527 can be accessed directly from the LCD.* ***For all other indications:*** *Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:*

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database* Back to Criteria
	• MolDX: Molecular Diagnostic Tests (MDT) (L36256). *Companion article A57527 can be accessed directly from the LCD.* *Laboratories in CA and NV:* • MolDX: Molecular Diagnostic Tests (MDT) (L35160). *Companion article A57526 can be accessed directly from the LCD.* These LCDs require that tests complete a technology assessment to determine if they meet Medicare's reasonable and necessary requirement. Tests that have completed this assessment are listed in the DEX™ registry. In addition, **clinical documentation must demonstrate how test results will be used** in the management or diagnosis of an illness or condition. *Laboratories in FL**, all indications***: Molecular Pathology Procedures for Human Leukocyte Antigen (HLA) Typing (L34518)
***Praxis Tests,* Praxis Genomics LLC, GA** • *Praxis Optical Genome Mapping* (0264U) • *Praxis Whole Genome Sequencing* (0265U) • *Praxis Transcriptome* (0266U) • *Praxis Combined Whole Genome Sequencing and Optical Genome Mapping* (0267U) • *Praxis Somatic Whole Genome Sequencing (0297U)* • *Praxis Somatic Transcriptome* (0298U) • *Praxis Somatic Optical Genome Mapping* (0299U) • *Praxis Somatic Combined Whole Genome Sequencing and Optical Genome Mapping (0300U)*	MolDX: The Palmetto LCD L35025 states reimbursement is only allowed for "approved tests… for dates of service consistent with the effective date of the coverage determination" after MolDX review. All of the tests by this laboratory are listed as "Not covered" in the DEX™ Diagnostics Exchange registry website. Therefore, these tests are considered **not medically reasonable or necessary** under the Palmetto GBA MolDX Program and the Social Security Act, §1862(a)(1)(A).

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database* Back to Criteria
PrismRA® *(81599, previously 0456U)* Scipher Medicine® (CA)	MolDX: Molecular Biomarker Testing to Guide Targeted Therapy Selection in Rheumatoid Arthritis (L39467) *Companion article A59521 can be accessed directly from the LCD.*
PROMETHEUS® IBD sgi Diagnostic® Test *(coding varies)* Prometheus Laboratories (CA)	MolDX: Prometheus IBD sgi Diagnostic Policy (L37299) *Companion article A57516 can be accessed directly from the LCD.*
Red Blood Cell Molecular Phenotyping *(0001U, 81403, 81479)*	*Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • RETIRED: MolDX: Molecular RBC Phenotyping (L36171) *Companion article A57445 can be accessed directly from the LCD.* *Laboratories in CA and NV:* RETIRED: Noridian LCD (L36167) *Companion article A57444 can be accessed directly from the LCD.*
Repeat Germline Testing *(CPT coding varies)*	*For laboratories in NC, SC, AL, GA, VA, or WV*: • MolDX: Repeat Germline Testing (L38274) *Companion article* A58017 *can be accessed directly from the LCD.* *For laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, or WY*: • MolDX: Repeat Germline Testing (L38353) *Companion article A57332 can be accessed directly from the LCD.* *For laboratories in CA or NV*: • MolDX: Repeat Germline Testing (L38351) *Companion article A57331 can be accessed directly from the LCD.* *For laboratories in KY or OH*: • MolDX: Repeat Germline Testing (L38288) *Companion article A57141 can be accessed directly from the LCD.* *For laboratories in IA, KS, MO, NE, IN, or MI*: MolDX: Repeat Germline Testing (L38429) *Companion article A57100 can be accessed directly from the LCD.*
Reproductive Molecular Testing, including but not limited to: ***Fetal chromosomal aneuploidy/trisomy*** *(81420, 81507, 0252U, 0327U)*	Billing and Coding: Molecular Pathology Procedures (A56199) *(Laboratories in IL, MN, WI, CT, NY, ME, MA, NH, RI, or VT) (Group 3 Codes are considered "not medically necessary." If CPT code not found, apply general Medicare rationale below.)* For laboratories in all other states, ***or*** if a specific CPT code is not found in an LCD listed above, apply the following Medicare guidance: In order to be eligible for Medicare coverage, an item or service must fall within a statutory benefit category. In order to be paid under the

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE For Medicare Coverage Determinations and Articles, see the Medicare Coverage Database	Back to Criteria
Fetal chromosomal microdeletion testing *(81422)* ***Reproductive carrier screening*** *(81443, 81479, 0400U, 0449U)* ***Other reproductive testing*** *(0060U, 0253U, 0254U, 0341U, 0407U, 0488U, 0489U, 0494U, 0536U, 0552U, 0553U, 0554U, 0555U)* Examples: • *Harmony™ Prenatal Test* • *InformaSeq℠ Prenatal Test (Integrated Genetics)* • *MaterniT Genome (Labcorp)* • *Panorama Prenatal Panel (Natera)* • *Panorama Extended Panel (Natera)* • *Verifi® Prenatal Test (Illumina)* • *Vanadis NIPT (Perkin Elmer)* • *POC (Product of Conception) (Igenomix®)* • *ERA® (Endometrial Receptivity Analysis) (Igenomix®)* • *Rh Test (Natera)* • *SMART PGT-A (Pre-implantation Genetic Testing - Aneuploidy) (Igenomix®)* • *Spectrum PGT-M (Natera)* • *UNITY testing: Carrier Screen™, Fetal Antigen™ NIPT, Fetal Risk Screen™ (BillionToOne, Inc)*	diagnostic laboratory testing benefit, a diagnostic test must (1) not be considered screening (testing in the absence of clinical signs and symptoms of disease) and (2) must be ordered by a physician who is treating the beneficiary and who will use the tests results in the management of a beneficiary's specific medical problem. These tests are performed in the absence of clinical signs/symptoms, and the test results are not used in the management of a beneficiary's specific medical problem. Therefore, while these tests may provide useful information, they are not considered medically reasonable or necessary as they do not meet a Medicare benefit category and/or reasonable and necessary threshold for coverage, as required by the SSA §1862(a)(1)(A) and 42 CFR 410.32(a).	

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database* Back to Criteria
SMASH™ *(0156U)* Marvel Genomics™ (NY)	This test is considered **not medically reasonable or necessary**. Medicare considers tests for diseases or conditions that manifest signs or symptoms in childhood to be exclusions, as they are not usually relevant to the Medicare population. Under Medicare, testing is only considered reasonable and necessary when the test results directly impact treatment or management of the beneficiary.
Specimen Validity Testing *(81479, 84999, 0079U)* Examples: • *ToxLok* • *know error®*	*Laboratories in CA, such as ToxLok,(0079U):* • *Controlled Substance Monitoring and Drugs of Abuse Testing (L36668) (In the "Non-Covered Services" section, see the guidance specific to specimen validity testing)* ***For specimen validity testing in general:*** Title XVIII of the Social Security Act, Section 1862(a)(1)(A) states *" ...no Medicare payment shall be made for items or services which are not reasonable and necessary for the diagnosis and treatment of illness or injury..."* (See also Medicare Benefit Policy Manual, Chapter 16 - General Exclusions From Coverage, §20 - Services Not Reasonable and Necessary*).* Specimen validity tests (i.e., tests performed to confirm sample identity or measure the quality of a process) **are not medically necessary** for Medicare because they do not provide information to diagnose or treat a condition.
Thoracic Aortic Disease Genetic Testing *(81410, 81411, 81479) e.g., Marfan syndrome, Loey-Dietz syndrome)*	*Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • MolDX: Genetic Testing for Heritable Thoracic Aortic Disease (L39946) *Companion article A59870 can be accessed directly from the LCD.* *Laboratories in CA and NV:* • MolDX: Genetic Testing for Heritable Thoracic Aortic Disease (L39944) *Companion article A59868 can be accessed directly from the LCD.*
Thrombophilia and/or Hypercoagulability Testing *(F2, F5, F9, and/or MTHFR genes) (coding varies)*	***For F2, F5, and/or MTHFR:*** *Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • MolDX: Genetic Testing for Hypercoagulability / Thrombophilia (Factor V Leiden, Factor II Prothrombin, and MTHFR) (L36159) *Companion article A57424 can be accessed directly from the LCD.* *Laboratories in CA and NV:* • MolDX: Genetic Testing for Hypercoagulability / Thrombophilia (Factor V Leiden, Factor II Prothrombin, and MTHFR) (L36155) *Companion article A57423 can be accessed directly from the LCD.*

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database* Back to Criteria
	These LCDs state, "Genetic testing for these genes for all risk factors, signs, symptoms, diseases, or conditions, including cardiovascular risk assessment, are ***non-covered except for pregnant patients****."* ***For F9 (coagulation factor IX) testing:*** *Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • LCD attachment for L36256, Excluded Test List – as of 08/01/2016 *Laboratories in CA and NV:* • LCD attachment for L35160, Excluded Test List – as of 08/01/2016 *Laboratories in IL, MN, WI, CT, NY, ME, MA, NH, RI, VT:* Billing and Coding: Molecular Pathology Procedures (A56199)
TTR Gene Tests for Transthyretin Amyloidosis	*Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • MolDX: Molecular Testing for Identification and Management of Hereditary Transthyretin Amyloidosis (L39950) *Companion article A59874 can be accessed directly from the LCD.* *Laboratories in CA and NV:* • MolDX: Molecular Testing for Identification and Management of Hereditary Transthyretin Amyloidosis (L39948) *Companion article A59872 can be accessed directly from the LCD.*
Versiti Panels**,** Versiti Inc. (WI) • *Autosomal Dominant Thrombocytopenia Panel (0269U)* • *Coagulation Disorder Panel (0270U)* • *Congenital Neutropenia Panel (0271U)* • *Comprehensive Bleeding Disorders Panel (0272U)* • *Comprehensive Platelet Disorder Panel (0274U)* • *Inherited Thrombocytopenia Panel (0276U)*	Molecular Pathology Procedures (L35000) *This LCD states, "Testing assay(s) are Food and Drug Administration (FDA) approved/cleared or if LDT (lab developed test) or LDT protocol or FDA modified test(s) the laboratory documentation should support assay(s) of analytical validity and clinical utility."* Although this lab is not in a MolDX jurisdiction, these tests have completed the MolDX technical assessment and are recorded in the DEX™ Diagnostics Exchange registry as "not covered," indicating that they lack analytical validity and/or clinical utility. Therefore, these panels are considered **not medically reasonable or necessary** under the Social Security Act, §1862(a)(1)(A).

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database*	Back to Criteria
• *Platelet Function Disorder Panel (0277U)* • *Thrombosis Panel (0278U)* • *Red Cell Genotyping Panel (0282U)*		
Versiti Fibrinolytic Disorder Panel *(0273U)* Versiti (WI)	Molecular Pathology Procedures (L35000) *(Applies to the indicated performing laboratory).* *According to this LCD, coverage for panel tests is limited to only the genes or tests that are reasonable and necessary to obtain information for therapeutic decision making. This panel test includes several gene tests with specific limited coverage criteria available and is therefore considered not medically reasonable or necessary.*	
Whole Exome and Whole Genome Sequencing *(81415, 81416, 81417, 81425, 81426, 81427, 81479, 0036U, 0094U, 0212U, 0213U, 0214U, 0215U, 0335U, 0336U, 0425U, 0426U, 0532U, 0567U, 0582U, 0583U)*	*Laboratories in AK, ID, OR, WA, UT, AZ, MT, ND, SD, WY:* • LCD attachment for L36256, Excluded Test List – as of 08/01/2016 *Laboratories in CA and NV:* • LCD attachment for L35160, Excluded Test List – as of 08/01/2016 *Laboratories in IL, MN, WI, CT, NY, ME, MA, NH, RI, or VT:* Billing and Coding: Molecular Pathology Procedures (A56199) *Non-coverage of genome and exome analysis includes the EXaCT-1 Whole Exome Test, 0036U. All whole exome and whole genome sequencing tests are considered non-covered, regardless of CPT code, until LCDs or articles indicate otherwise.*	
Miscellaneous Non-Covered Tests:		
• ***ScoliScore™ Transgenomic*** *(0004M)* • ***HeproDX™*** *(0006M)* GoPath Laboratories, LLC • ***NETest*** *(0007M) Wren Laboratories, LLC*	Due to the prior non-coverage and in the absence of a coverage determination by MolDX (Excluded Test List – as of 08/01/2016), these tests are considered **not medically reasonable and necessary** until a MolDX review is complete and coverage is indicated by MolDX or Noridian.	
• ***Apifiny®*** *(0021U)* Armune BioScience, Inc. (MI) • ***Augusta Optical Genome Mapping*** *(0260U)* Bionano	The MolDX Program requires labs to submit a technology assessment (TA) to provide evidence of analytical and clinical validity, and clinical utility. Reimbursement is only allowed for "approved tests… for dates of service consistent with the effective date of the coverage determination" after MolDX review. The results of these TAs are published on the DEX™ Registry, with a "covered"	

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE **For Medicare Coverage Determinations and Articles, see the** *Medicare Coverage Database*	Back to Criteria
Genomics (GA) • ***Avise Lupus*** *(0312U) and Avise CTD (81599)* Exagen, Inc. (CA) • ***HCMFirst*** *(81479) and* ***HHTNext*** *(81479)* Ambry Genetics (CA) • ***LVNCNext™*** *(81479)* Ambry Genetics (CA) • ***Lyme Borrelia Nanotrap® Urine Antigen Test*** *(0316U)* Galaxy Diagnostics (NC) • ***Lyme ImmunoBlots IgM and IgG Tests*** *(0041U, 0042U)* IgeneX (CA) • ***Macula Risk® PGx*** *(81479)* and ***Vita Risk*** *(0205U)* Arctic Medical Laboratories (MI) • ***Mind.Px*** *(0258U)* Mindera (CA) • ***NASHnext™ (NIS4®)*** *(0468U)* Labcorp • ***OncobiotaLUNG*** *(0395U)* Micronoma (CA) • ***RhythmFirst*** *(81479)* Ambry Genetics (CA) • ***Tick-Borne Relapsing Fever Borrelia ImmunoBlots IgM and IgG*** *(0043U, 0044U)* IgeneX (CA)	or “not covered” determination. According to the DEX™ Registry, these tests are listed as “not covered” or they do not have a coverage determination and are therefore considered **not medically necessary**.	
• ***MindX Blood Tests*** *(Memory 0289U, Pain 0290U, Mood*	*Title XVIII of the Social Security Act, Section 1862(a)(1)(A)* states " ...no Medicare payment shall be made for items or services which are not reasonable and necessary for the diagnosis and treatment of	

TEST INFORMATION	MEDICARE RATIONALE / REFERENCE For Medicare Coverage Determinations and Articles, see the Medicare Coverage Database	Back to Criteria
0291U, Stress *0292U, Suicidality* *0293U, Longevity* *0294U, Anxiety* *0437U)* MindX Sciences • ***NaviDKD*** *(0384U)* Journey Biosciences • ***PredictSure IBD*** *(0203U)* KSL Diagnostics • ***PromarkerD*** *(0385U) Sonic Reference Laboratory*	illness or injury..." *(See also Medicare Benefit Policy Manual, Chapter 16 - General Exclusions From Coverage, §20 - Services Not Reasonable and Necessary).* The purpose of these tests is to predict the risk of developing a disease or condition. They are not diagnostic tests. Therefore, they are considered excluded tests services.	

POLICY GUIDELINES

Important Notes Regarding Diagnostic Laboratory and Genetic Testing Services

Medicare and Medical Necessity

According to Medicare guidelines, Medicare coverage is contingent upon the services meeting certain requirements to determine medical necessity. To be considered a covered service, Medicare requires that the service in question:

- Fall within a defined Medicare benefit category.[1,2]
- Not be excluded from coverage by statute, regulation, National Coverage Determination, (NCD), or Local Coverage Determination (LCD).[2]
- Be considered medically necessary, as required per the Social Security Act, §1862(a)(1)(A). This means the service must be considered reasonable and necessary in the diagnosis or treatment of an illness or injury, or to rule out or confirm a suspected diagnosis because the patient has signs and/or symptoms;[1,3,4] This also means services determined to be not medically necessary for any reason (including lack of safety and efficacy because it is an investigational service) are non-covered.[5]
- Be ordered by a physician who is treating the beneficiary.[6,7]
- Provide data that would be directly used in the management of a beneficiary's specific medical problem.[6,7]

For the referring physician to effectively manage their patient's specific medical problem using genetic or molecular diagnostic testing, the genetic tests performed must be used to assist in the management/treatment of the beneficiary. Therefore, it is important for referring physicians to be familiar with all specific genetic tests they order to ensure all test result components are clinically actionable.

In addition to the above Medicare requirements, when making coverage decision policies, under Chapter 13 of the Medicare Program Integrity Manual, Medicare allows contractors to consider a service "reasonable and necessary" when the service is appropriate for the member's condition. This includes appropriateness in duration, frequency, and that the service is furnished in accordance with accepted standards of medical practice for the condition, furnished in a setting appropriate to the medical needs and condition, ordered and furnished by qualified personnel, that the service meets, but does not exceed, the medical need; and is at least as beneficial as an existing and available medically appropriate alternative.[21]

Services Excluded from Coverage

Tests performed in the absence of signs, symptoms, complaints, personal history of disease, or injury are not covered, except when there is a statutory provision that explicitly covers a specific screening test. Tests that confirm a diagnosis or known information, and tests to determine risk for developing a disease or condition are also excluded test services.(8-11)

Molecular Diagnostic Services Program (MolDX)

The Medicare Molecular Diagnostic Services Program (MolDX) was developed in 2011 to identify and establish coverage and reimbursement for molecular diagnostic tests, and is maintained by Palmetto GBA. Palmetto evaluates genetic tests to determine analytical and clinical validity and clinical utility, as well as confirming that each test meets Medicare criteria (described below). Palmetto MolDX guidelines provide assessments and indicate coverage or non-coverage of the test.[12-15]

The MolDX program will affect diagnostic services reported with the following CPT/HCPCS codes: (13)

Code Category/Description	2018 MolDx Code Range
Tier 1	81161-81383
Tier 2	81400-81408
Genomic Sequencing Procedures	81410-81471
Molecular Multianalyte Assays (MAAA)	81490-81595
MAAA Admin. Codes	MAAA codes for molecular tests
Immunology	86152-86153
PLA	PLA codes for molecular tests
Cytology	88120-88121
The following NOC codes are in scope for molecular tests only	81479, 81599, 84999, 85999, 86849, 87999

For Testing Performed by a Laboratory Outside of the Medicare Advantage Organization's (MAO) Service Area

"A MAC outside of the plan's service area sometimes has exclusive jurisdiction over a Medicare covered item or service. In some instances, one Medicare A/B MAC processes all of the claims for a particular Medicare-covered item or service for all Medicare beneficiaries around the country. This generally occurs when there is only one supplier of a particular item, medical device or diagnostic test (for example: certain pathology and lab tests furnished by independent laboratories). In this situation, MA plans must follow the coverage requirements or LCD of the MAC that enrolled the supplier and processes all of the Medicare claims for that item, test or service."[15]

In addition, “Jurisdiction of claims for laboratory services furnished by an independent laboratory normally lies with the carrier serving the area in which the laboratory test is performed. However, there are some situations where a regional or national lab chain jurisdiction is with a single carrier.”[16]

REQUIRED DOCUMENTATION

The following information is required in order to determine medical necessity and potential Medicare coverage for a genetic or molecular diagnostic test. *[See Title XVIII of the Social Security Act, §1833(e), which states no payment may be made unless information necessary to determine payment has been submitted)]*

1. The specific name of the genetic or molecular diagnostic test or panel;
    a. The DEX Z-code as assigned by DEX™ Diagnostics Exchange and/or a copy of the decision letter by the MolDX Program would also be beneficial in making timely and efficient coverage determinations;
2. Name and location of the performing laboratory;
3. The exact gene(s) and/or variants being tested, if applicable;
4. Applicable CPT and/or HCPCS code(s);
5. Brief explanation of how the results of genetic testing are necessary to guide treatment decisions relevant to the member’s personal medical history. Tests performed for the following purposes are a few examples:
    - Diagnose an illness when signs/symptoms are displayed; or
    - Rule out a diagnosis when signs/symptoms are displayed; or
    - Guide treatment planning for a previously diagnosed illness (i.e., whether to perform surgery or choose between medication options, etc.); and,
6. Medical records relevant to the testing being performed. This includes:
    - History and physical examinations by the referring physician;
    - Conventional testing and outcomes; and
    - Conservative treatment provided, if applicable.

CROSS REFERENCES

1. Genetic and Molecular Diagnostics – Testing for Inherited Cancer Risk, Genetic Testing, Policy No. M-02
2. Pharmacogenomics (PGx) Testing, Genetic Testing, Policy No. M-10
3. Genetic and Molecular Diagnostics – Testing for Cancer Diagnosis, Prognosis, and Treatment Selection, Genetic Testing, Policy No. M-83
4. Molecular Panel Testing for Identification of Microorganisms, Genetic Testing, Policy No. M-85
5. Laboratory Tests for Organ Transplant Rejection, Laboratory, Policy No. M-51
6. COVID-19 Testing, Laboratory, Policy No. M-74
7. Biomarkers for Cardiovascular Disease, Laboratory, Policy No. M-78

REFERENCES

1. Medicare Coverage Determination Process
2. Medicare Managed Care Manual, Ch. 4 - Benefits and Beneficiary Protections, §10.2 - Basic Rule
3. Title XVIII of the Social Security Act, §1862(a)(1)(A)
4. Medicare Benefit Policy Manual, Chapter 16 - General Exclusions From Coverage, §20 - Services Not Reasonable and Necessary

5. Medicare Claims Processing Manual, Chapter 23 - Fee Schedule Administration and Coding Requirements, §30 - Services Paid Under the Medicare Physician's Fee Schedule, Subsection A
6. 42 CFR §410.32(a)
7. Medicare Benefit Policy Manual, Ch. 15 – Covered Medical and Other Health Services, §80.1 - Clinical Laboratory Services
8. Federal Register / Vol. 66, No. 226 / Friday, November 23, 2001
9. Medicare Claims Processing Manual, Chapter 16 – Laboratory Services, §120.1, Negotiated Rulemaking Implementation, see section regarding "Clarification of the Use of the Term 'Screening' or 'Screen'"
10. Medicare National Coverage Determinations (NCD) Coding Policy Manual and Change Report January 2013
11. Palmetto GBA MolDX Program
12. Noridian Healthcare Solutions - Palmetto GBA MolDX Program for Jurisdiction F
13. Molecular Diagnostics Program (MolDX®) Manual
14. Palmetto GBA Molecular Diagnostic Tests and Medicare web page
15. Medicare Managed Care Manual, Ch. 4 - Benefits and Beneficiary Protections, §90.4.1 - MACS with Exclusive Jurisdiction over a Medicare Item or Service
16. Medicare Claims Processing Manual, Chapter 1 - General Billing Requirements, §10.1.5.4 - Independent Laboratories
17. Novitas Article, Biomarkers for Oncology (A52986) *(This reference can be found on the Medicare Coverage Database website)*
18. Retired Noridian Article, Molecular Genetic Testing (A52932)
19. Palmetto GBA MolDX: Molecular Test Panel Edit Alert
20. Medicare Claims Processing Manual, Chapter 16 - Laboratory Services, §50.5 - Jurisdiction of Laboratory Claims
21. Medicare Program Integrity Manual, Chapter 13 – Local Coverage Determinations, §13.5.4 - Reasonable and Necessary Provision in an LCD

CODING

NOTE: CPT codes 81400-81408 are nonspecific and many of the tests represented by these codes are not covered by Medicare.[18] To determine coverage for testing, the name of the test being performed must be included.

For laboratories in the health plan's service area, instructions regarding the reporting of next generation sequencing (NGS), targeted tumor panels, or other panel testing, see the Noridian article, *MolDX: Defining panel services in MolDX* (A59678) for definitions and coding expectations.

In addition, HCPCS S-codes are not payable by Medicare, and therefore, are not payable for the health plan's Medicare Advantage members.

Codes	Number	Description
CPT	0004M	Scoliosis, DNA analysis of 53 single nucleotide polymorphisms (SNPs), using saliva, prognostic algorithm reported as a risk score (ScoliScoreTM)
	0001U	Red blood cell antigen typing, DNA, human erythrocyte antigen gene analysis of 35 antigens from 11 blood groups, utilizing whole blood, common RBC alleles reported
	0035U	Neurology (prion disease), cerebrospinal fluid, detection of prion protein by quaking induced conformational conversion, qualitative

0039U	Autoimmunity (Systemic Lupus Erythematosus, SLE), detection of high avidity antidsDNA antibodies, ELISA assay, serum, reported as quantitative number for therapeutic decision making
0041U	Borrelia burgdorferi, antibody detection of 5 recombinant protein groups, by immunoblot, IgM
0042U	Borrelia burgdorferi, antibody detection of 5 recombinant protein groups, by immunoblot, IgG
0043U	Tick-borne relapsing fever Borrelia group, antibody detection to 4 recombinant protein groups, by immunoblot, IgM
0044U	Tick-borne relapsing fever Borrelia group, antibody detection to 4 recombinant protein groups, by immunoblot, IgG
0060U	Twin zygosity, genomic targeted sequence analysis of chromosome 2, using circulating cell-free fetal DNA in maternal blood
0068U	Candida species panel (C. albicans, C. glabrata, C. parapsilosis, C. kruseii, C tropicalis, and C. auris), amplified probe technique with qualitative report of the presence or absence of each species
0078U	Pain management (opioid-use disorder) genotyping panel, 16 common variants (ie, ABCB1, COMT, DAT1, DBH, DOR, DRD1, DRD2, DRD4, GABA, GAL, HTR2A, HTTLPR, MTHFR, MUOR, OPRK1, OPRM1), buccal swab or other germline tissue sample, algorithm reported as positive or negative risk of opioid-use disorder (Deleted 10/1/2024)
0079U	Comparative DNA analysis using multiple selected single-nucleotide polymorphisms (SNPs), urine and buccal DNA, for specimen identity verification
0094U	Genome (eg, unexplained constitutional or heritable disorder or syndrome), rapid sequence analysis
0105U	Nephrology (chronic kidney disease), multiplex electrochemiluminescent immunoassay (ECLIA) of tumor necrosis factor receptor 1A, receptor superfamily 2 (TNFR1, TNFR2), and kidney injury molecule-1 (KIM-1) combined with longitudinal clinical data, including APOL1 genotype if available, and plasma (isolated fresh or frozen), algorithm reported as probability score for rapid kidney function decline (RKFD)
0156U	Copy number (eg, intellectual disability, dysmorphology), sequence analysis
0203U	Autoimmune (inflammatory bowel disease), mRNA, gene expression profiling by quantitative RT-PCR, 17 genes (15 target and 2 reference genes), whole blood, reported as a continuous risk score and classification of inflammatory bowel disease aggressiveness
0205U	Ophthalmology (age-related macular degeneration), analysis of 3 gene variants (2 CFH gene, 1 ARMS2 gene), using PCR and MALDI-TOF, buccal swab, reported as positive or negative for neovascular age-related macular-degeneration risk associated with zinc supplements
0209U	Cytogenomic constitutional (genome-wide) analysis, interrogation of genomic regions for copy number, structural changes and areas of homozygosity for chromosomal abnormalities
0212U	Rare diseases (constitutional/heritable disorders), whole genome and mitochondrial DNA sequence analysis, including small sequence changes, deletions, duplications, short tandem repeat gene expansions, and variants in non-uniquely mappable regions, blood or saliva, identification and categorization of genetic variants, proband
0213U	Rare diseases (constitutional/heritable disorders), whole genome and mitochondrial DNA sequence analysis, including small sequence changes, deletions, duplications, short tandem repeat gene expansions, and variants in non-uniquely mappable regions, blood or saliva, identification and

	categorization of genetic variants, each comparator genome (eg, parent, sibling)
0214U	Rare diseases (constitutional/heritable disorders), whole exome and mitochondrial DNA sequence analysis, including small sequence changes, deletions, duplications, short tandem repeat gene expansions, and variants in non-uniquely mappable regions, blood or saliva, identification and categorization of genetic variants, proband
0215U	;saliva, identification and categorization of genetic variants, each comparator exome (eg, parent, sibling)
0216U	Neurology (inherited ataxias), genomic DNA sequence analysis of 12 common genes including small sequence changes, deletions, duplications, short tandem repeat gene expansions, and variants in non-uniquely mappable regions, blood or saliva, identification and categorization of genetic variant
0217U	Neurology (inherited ataxias), genomic DNA sequence analysis of 51 genes including small sequence changes, deletions, duplications, short tandem repeat gene expansions, and variants in non-uniquely mappable regions, blood or saliva, identification and categorization of genetic variants
0218U	Neurology (muscular dystrophy), DMD gene sequence analysis, including small sequence changes, deletions, duplications, and variants in non-uniquely mappable regions, blood or saliva, identification and characterization of genetic variants
0230U	AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation), full sequence analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-uniquely mappable regions
0231U	CACNA1A (calcium voltage-gated channel subunit alpha 1A) (eg, spinocerebellar ataxia), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) gene expansions, mobile element insertions, and variants in non-uniquely mappable regions
0232U	CSTB (cystatin B) (eg, progressive myoclonic epilepsy type 1A, Unverricht-Lundborg disease), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-uniquely mappable regions
0233U	FXN (frataxin) (eg, Friedreich ataxia), gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-uniquely mappable regions
0234U	MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, mobile element insertions, and variants in non-uniquely mappable regions
0236U	SMN1 (survival of motor neuron 1, telomeric) and SMN2 (survival of motor neuron 2, centromeric) (eg, spinal muscular atrophy) full gene analysis, including small sequence changes in exonic and intronic regions, duplications and deletions, and mobile element insertions
0237U	Cardiac ion channelopathies (eg, Brugada syndrome, long QT syndrome, short QT syndrome, catecholaminergic polymorphic ventricular tachycardia), genomic sequence analysis panel including ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, and SCN5A, including small

	sequence changes in exonic and intronic regions, deletions, duplications, mobile element insertions, and variants in non-uniquely mappable regions
0243U	Obstetrics (preeclampsia), biochemical assay of placental-growth factor, time-resolved fluorescence immunoassay, maternal serum, predictive algorithm reported as a risk score for preeclampsia
0247U	Obstetrics (preterm birth), insulin-like growth factor–binding protein 4 (IBP4), sex hormone–binding globulin (SHBG), quantitative measurement by LC-MS/MS, utilizing maternal serum, combined with clinical data, reported as predictive-risk stratification for spontaneous preterm birth
0252U	Fetal aneuploidy short tandem–repeat comparative analysis, fetal DNA from products of conception, reported as normal (euploidy), monosomy, trisomy, or partial deletion/duplications, mosaicism, and segmental aneuploidy
0253U	Reproductive medicine (endometrial receptivity analysis), RNA gene expression profile, 238 genes by next-generation sequencing, endometrial tissue, predictive algorithm reported as endometrial window of implantation (eg, pre-receptive, receptive, post-receptive)
0254U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using embryonic DNA genomic sequence analysis for aneuploidy, and a mitochondrial DNA score in euploid embryos, results reported as normal (euploidy), monosomy, trisomy, or partial deletion/duplications, mosaicism, and segmental aneuploidy, per embryo tested
0258U	Autoimmune (psoriasis), mRNA, next generation sequencing, gene expression profiling of 50-100 genes, skin-surface collection using adhesive patch, algorithm reported as likelihood of response to psoriasis biologics
0260U	Rare diseases (constitutional/heritable disorders), identification of copy number variations, inversions, insertions, translocations, and other structural variants by optical genome mapping
0264U	Rare diseases (constitutional/heritable disorders), identification of copy number variations, inversions, insertions, translocations, and other structural variants by optical genome mapping
0265U	Rare constitutional and other heritable disorders, whole genome and mitochondrial DNA sequence analysis, blood, frozen and formalin-fixed paraffin embedded (FFPE) tissue, saliva, buccal swabs or cell lines, identification of single nucleotide and copy number variants
0266U	Unexplained constitutional or other heritable disorders or syndromes, tissue specific gene expression by whole transcriptome and next-generation sequencing, blood, formalin-fixed paraffin embedded (FFPE) tissue or fresh frozen tissue, reported as presence or absence of splicing or expression changes
0267U	Rare constitutional and other heritable disorders, identification of copy number variations, inversions, insertions, translocations, and other structural variants by optical genome mapping and whole genome sequencing
0269U	Hematology (autosomal dominant congenital thrombocytopenia), genomic sequence analysis of 22 genes, blood, buccal swab, or amniotic fluid
0270U	Hematology (congenital coagulation disorders), genomic sequence analysis of 20 genes, blood, buccal swab, or amniotic fluid
0271U	Hematology (congenital neutropenia), genomic sequence analysis of 24 genes, blood, buccal swab, or amniotic fluid
0272U	Hematology (genetic bleeding disorders), genomic sequence analysis of 60 genes, and duplication/deletion of PLAU, blood, buccal swab, or amniotic fluid, comprehensive

0273U	Hematology (genetic hyperfibrinolysis, delayed bleeding), analysis of 9 genes (F13A1, F13B, FGA, FGB, FGG, SERPINA1, SERPINE1, SERPINF2, by nextgeneration sequencing and PLAU by array comparative genomic hybridization), blood, buccal swab, or amniotic fluid
0274U	Hematology (genetic platelet disorders), genomic sequence analysis of 62 genes, and duplication/deletion of PLAU, blood, buccal swab, or amniotic fluid
0276U	Hematology (inherited thrombocytopenia), genomic sequence analysis of 42 genes, blood, buccal swab, or amniotic fluid
0277U	Hematology (genetic platelet function disorder), genomic sequence analysis of 40 genes, and duplication/deletion of PLAU, blood, buccal swab, or amniotic fluid
0278U	Hematology (genetic thrombosis), genomic sequence analysis of 14 genes, blood, buccal swab, or amniotic fluid
0282U	Red blood cell antigen typing, DNA, genotyping of 12 blood group system genes to predict 44 red blood cell antigen phenotypes
0289U	Neurology (Alzheimer disease), mRNA, gene expression profiling by RNA sequencing of 24 genes, whole blood, algorithm reported as predictive risk score
0290U	Pain management, mRNA, gene expression profiling by RNA sequencing of 36 genes, whole blood, algorithm reported as predictive risk score
0291U	Psychiatry (mood disorders), mRNA, gene expression profiling by RNA sequencing of 144 genes, whole blood, algorithm reported as predictive risk score
0292U	Psychiatry (stress disorders), mRNA, gene expression profiling by RNA sequencing of 72 genes, whole blood, algorithm reported as predictive risk score
0293U	Psychiatry (suicidal ideation), mRNA, gene expression profiling by RNA sequencing of 54 genes, whole blood, algorithm reported as predictive risk score
0294U	Longevity and mortality risk, mRNA, gene expression profiling by RNA sequencing of 18 genes, whole blood, algorithm reported as predictive risk score
0312U	Autoimmune diseases (eg, systemic lupus erythematosus [SLE]), analysis of 8 IgG autoantibodies and 2 cell-bound complement activation products using enzyme-linked immunosorbent immunoassay (ELISA), flow cytometry and indirect immunofluorescence, serum, or plasma and whole blood, individual components reported along with an algorithmic SLE-likelihood assessment
0316U	Borrelia burgdorferi (Lyme disease), OspA protein evaluation, urine
0327U	Fetal aneuploidy (trisomy 13, 18, and 21), DNA sequence analysis of selected regions using maternal plasma, algorithm reported as a risk score for each trisomy, includes sex reporting, if performed
0335U	Rare diseases (constitutional/heritable disorders), whole genome sequence analysis, including small sequence changes, copy number variants, deletions, duplications, mobile element insertions, uniparental disomy (UPD), inversions, aneuploidy, mitochondrial genome sequence analysis with heteroplasmy and large deletions, short tandem repeat (STR) gene expansions, fetal sample, identification and categorization of genetic variants (Do not report 0335U in conjunction with 81425, 0212U)
0336U	Rare diseases (constitutional/heritable disorders), whole genome sequence analysis, including small sequence changes, copy number variants, deletions, duplications, mobile element insertions, uniparental disomy (UPD), inversions, aneuploidy, mitochondrial genome sequence analysis with heteroplasmy and

	large deletions, short tandem repeat (STR) gene expansions, blood or saliva, identification and categorization of genetic variants, each comparator genome (eg, parent) (Do not report 0336U in conjunction with 81426, 0213U)
0341U	Fetal aneuploidy DNA sequencing comparative analysis, fetal DNA from products of conception, reported as normal (euploidy), monosomy, trisomy, or partial deletion/duplication, mosaicism, and segmental aneuploid
0355U	APOL1 (apolipoprotein L1) (eg, chronic kidney disease), risk variants (G1, G2)
0378U	RFC1 (replication factor C subunit 1), repeat expansion variant analysis by traditional and repeat-primed PCR, blood, saliva, or buccal swab
0384U	Nephrology (chronic kidney disease), carboxymethyllysine, methylglyoxal hydroimidazolone, and carboxyethyl lysine by liquid chromatography with tandem mass spectrometry (LCMS/MS) and HbA1c and estimated glomerular filtration rate (GFR), with risk score reported for predictive progression to high-stage kidney disease
0385U	Nephrology (chronic kidney disease), apolipoprotein A4 (ApoA4), CD5 antigen-like (CD5L), and insulin-like growth factor binding protein 3 (IGFBP3) by enzyme-linked immunoassay (ELISA), plasma, algorithm combining results with HDL, estimated glomerular filtration rate (GFR) and clinical data reported as a risk score for developing diabetic kidney disease
0389U	Pediatric febrile illness (Kawasaki disease [KD]), interferon alphainducible protein 27 (IFI27) and mast cell-expressed membrane protein 1 (MCEMP1), RNA, using reverse transcription polymerase chain reaction (RT-qPCR), blood, reported as a risk score for KD
0390U	Obstetrics (preeclampsia), kinase insert domain receptor (KDR), Endoglin (ENG), and retinol-binding protein 4 (RBP4), by immunoassay, serum, algorithm reported as a risk score
0393U	Neurology (eg, Parkinson disease, dementia with Lewy bodies), cerebrospinal fluid (CSF), detection of misfolded α-synuclein protein by seed amplification assay, qualitative
0396U	Obstetrics (pre-implantation genetic testing), evaluation of 300000 DNA single-nucleotide polymorphisms (SNPs) by microarray, embryonic tissue, algorithm reported as a probability for single-gene germline conditions (Deleted 10/1/2024)
0399U	Neurology (cerebral folate deficiency), serum, detection of anti-human folate receptor IgG binding antibody and blocking autoantibodies by enzyme-linked immunoassay (ELISA), qualitative, and blocking autoantibodies, using a functional blocking assay for IgG or IgM, quantitative, reported as positive or not detected
0400U	Obstetrics (expanded carrier screening), 145 genes by next generation sequencing, fragment analysis and multiplex ligation dependent probe amplification, DNA, reported as carrier positive or negative
0407U	Nephrology (diabetic chronic kidney disease [CKD]), multiplex electrochemiluminescent immunoassay (ECLIA) of soluble tumor necrosis factor receptor 1 (sTNFR1), soluble tumor necrosis receptor 2 (sTNFR2), and kidney injury molecule 1 (KIM-1) combined with clinical data, plasma, algorithm reported as risk for progressive decline in kidney function
0417U	Rare diseases (constitutional/heritable disorders), whole mitochondrial genome sequence with heteroplasmy detection and deletion analysis, nuclearencoded mitochondrial gene analysis of 335 nuclear genes, including sequence changes, deletions, insertions, and copy number variants analysis,

	blood or saliva, identification and categorization of mitochondrial disorder–associated genetic variants
0425U	Genome (eg, unexplained constitutional or heritable disorder or syndrome), rapid sequence analysis, each comparator genome (eg, parents, siblings)
0426U	Genome (eg, unexplained constitutional or heritable disorder or syndrome), ultra-rapid sequence analysis
0437U	Psychiatry (anxiety disorders), mRNA, gene expression profiling by RNA sequencing of 15 biomarkers, whole blood, algorithm reported as predictive risk score
0441U	Infectious disease (bacterial, fungal or viral infection), semiquantitative biomechanical assessment (via deformability cytometry), whole blood, with algorithmic analysis and result reported as an index.
0442U	Infectious disease (respiratory infection), Myxovirus resistance protein A (MxA) and C-reactive protein (CRP), fingerstick whole blood specimen, each biomarker reported as present or absent
0446U	Autoimmune diseases (systemic lupus erythematosus) [SLE], analysis of 10 cytokine soluble mediator biomarkers by immunoassay, plasma, individual components reported with an algorithmic risk score for current disease activity
0447U	Autoimmune diseases (systemic lupus erythematosus [SLE]), analysis of 11 cytokine soluble mediator biomarkers by immunoassay, plasma, individual components reported with an algorithmic prognostic risk score for developing a clinical flare
0449U	Carrier screening for severe inherited conditions (eg, cystic fibrosis, spinal muscular atrophy, beta hemoglobinopathies [including sickle cell disease], alpha thalassemias) regardless of race or self-identified ancestry, genomic sequence analysis of 5 genes (CFTR, SMN1, HBB, HBA1, HBA2)
0456U	Autoimmune (rheumatoid arthritis), next-generation sequencing (NGS), gene expression testing of 19 genes, whole blood, with analysis of anticyclic citrullinated peptides (CCP) levels, combined with sex, patient global assessment, and body mass index (BMI), algorithm reported as a score that predicts nonresponse to tumor necrosis factor inhibitor (TNFi) therapy (Deleted 1/1/2025)
0457U	Perfluoroalkyl substances (PFAS) (eg, perfluorooctanoic acid, perfluorooctane sulfonic acid), 9 PFAS compounds by LC-MS/MS, plasma or serum, quantitative
0468U	Hepatology (nonalcoholic steatohepatitis [NASH]), miR-34a5p, alpha 2-macroglobulin, YKL40, HbA1c, serum and whole blood, algorithm reported as a single score for NASH activity and fibrosis
0482U	Obstetrics (preeclampsia), biochemical assay of soluble fms-like tyrosine kinase 1 (sFlT-1) and placental growth factor (PIGF), serum, ratio reported for sFlT-1/PIGF, with risk of progression for preeclampsia with severe features within 2 weeks
0488U	Obstetrics (fetal antigen noninvasive prenatal test), cell-free DNA sequence analysis for detection of fetal presence or absence of 1 or more of the Rh, C, c, D, E, Duffy (Fya), or Kell (K) antigen in alloimmunized pregnancies, reported as selected antigen(s) detected or not detected
0489U	Obstetrics (single-gene noninvasive prenatal test), cellfree DNA sequence analysis of 1 or more targets (eg, CFTR, SMN1, HBB, HBA1, HBA2) to identify paternally inherited pathogenic variants, and relative mutation-dosage analysis based on molecular counts to determine fetal inheritance of maternal mutation, algorithm reported as a fetal risk score for the condition (eg, cystic fibrosis, spinal muscular atrophy, beta hemoglobinopathies [including sickle cell disease], alpha thalassemia)

0494U	Red blood cell antigen (fetal RhD gene analysis), next-generation sequencing of circulating cell-free DNA (cfDNA) of blood in pregnant individuals known to be RhD negative, reported as positive or negative
0500U	Autoinflammatory disease (VEXAS syndrome), DNA, UBA1 gene mutations, targeted variant analysis (M41T, M41V, M41L, c.118-2A>C, c.118-1G>C, c.118- 9_118-2del, S56F, S621C)
0502U	Human papillomavirus (HPV), E6/E7 markers for high-risk types (16, 18, 31, 33, 35, 39, 45, 51, 52, 56, 58, 59, 66, and 68), cervical cells, branched-chain capture hybridization, reported as negative or positive for high risk for HPV
0524U	Obstetrics (preeclampsia), sFlt-1/PlGF ratio, immunoassay, utilizing serum or plasma, reported as a value
0527U	Herpes simplex virus (HSV) types 1 and 2 and Varicella zoster virus (VZV), amplified probe technique, each pathogen reported as detected or not detected
0532U	Rare diseases (constitutional disease/hereditary disorders), rapid whole genome and mitochondrial DNA sequencing for singlenucleotide variants, insertions/deletions, copy number variations, peripheral blood, buffy coat, saliva, buccal or tissue sample, results reported as positive or negative
0536U	Red blood cell antigen (fetal RhD), PCR analysis of exon 4 of RHD gene and housekeeping control gene GAPDH from whole blood in pregnant individuals at 10+ weeks gestation known to be RhD negative, reported as fetal RhD status
0542U	Nephrology (renal transplant), urine, nuclear magnetic resonance (NMR) spectroscopy measurement of 84 urinary metabolites, combined with patient data, quantification of BK virus (human polyomavirus 1) using real-time PCR and serum creatinine, algorithm reported as a probability score for allograft injury status
0552U	Reproductive medicine (preimplantation genetic assessment), analysis for known genetic disorders from trophectoderm biopsy, linkage analysis of diseasecausing locus, and when possible, targeted mutation analysis for known familial variant, reported as low-risk or high-risk for familial genetic disorder
0553U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using DNA genomic sequence analysis from embryonic trophectoderm for structural rearrangements, aneuploidy, and a mitochondrial DNA score, results reported as normal/balanced (euploidy/balanced), unbalanced structural rearrangement, monosomy, trisomy, segmental aneuploidy, or mosaic, per embryo tested
0554U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using DNA genomic sequence analysis from trophectoderm biopsy for aneuploidy, ploidy, a mitochondrial DNA score, and embryo quality control, results reported as normal (euploidy), monosomy, trisomy, segmental aneuploidy, triploid, haploid, or mosaic, with quality control results reported as contamination detected or inconsistent cohort when applicable, per embryo tested
0555U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using DNA genomic sequence analysis from embryonic trophectoderm for structural rearrangements, aneuploidy, ploidy, a mitochondrial DNA score, and embryo quality control, results reported as normal/balanced (euploidy/balanced), unbalanced structural rearrangement, monosomy, trisomy, segmental aneuploidy, triploid, haploid, or mosaic, with quality control results reported as contamination detected or inconsistent cohort when applicable, per embryo tested

0567U	Rare diseases (constitutional/heritable disorders), whole-genome sequence analysis combination of short and long reads, for single-nucleotide variants, insertions/deletions and characterized intronic variants, copy-number variants, duplications/deletions, mobile element insertions, runs of homozygosity, aneuploidy, and inversions, mitochondrial DNA sequence and deletions, short tandem repeat genes, methylation status of selected regions, blood, saliva, amniocentesis, chorionic villus sample or tissue, identification and categorization of genetic variants
0579U	Nephrology (diabetic chronic kidney disease), enzyme linked immunosorbent assay (ELISA) of apolipoprotein A4 (APOA4), CD5 antigen-like (CD5L) combined with estimated glomerular filtration rate (GFR), age, plasma, algorithm reported as a risk score for kidney function decline
0582U	Rare diseases (constitutional disease/hereditary disorders), rapid whole genome DNA sequencing for single nucleotide variants, insertions/deletions, copy number variations, blood, saliva, tissue sample, variants reported
0583U	Rare diseases (constitutional disease/hereditary disorders), rapid whole genome comparator DNA sequencing for single-nucleotide variants, insertions/deletions, copy number variations, blood, saliva, tissue sample, variants reported with proband results (List separately in addition to code for primary procedure)
0584U	Neurology (prion disease), cerebrospinal fluid, detection of prion protein by quaking induced conformational conversion, qualitative
0588U	Infectious disease (bacterial or viral), 32 genes (29 informative and 3 housekeeping), immune response mRNA, gene expression profiling by split well multiplex reverse transcription loop-mediated isothermal amplification (RT LAMP), whole blood, reported as continuous risk scores for likelihood of bacterial and viral infection and likelihood of severe illness within the next 7 days
0589U	Perfluoroalkyl substances (PFAS) (eg, perfluorooctanoic acid, perfluorooctane sulfonic acid), 24 PFAS compounds by high-performance liquid chromatography with tandem mass spectrometry (LCMS/MS), plasma or serum, quantitative
0594U	Infectious disease (sepsis), semiquantitative measurement of pancreatic stone protein concentration, whole blood, reported as risk of sepsis
0598U	Gastroenterology (irritable bowel syndrome), IgG antibodies to 18 food items by microarray-based immunoassay, whole blood or serum, report as elevated (positive) or normal (negative) antibody levels
81105	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), gene analysis, common variant, HPA-1A/B (L33P)
81106	Human platelet antigen 2 genotyping (HPA-2), GP1BA (glycoprotein ib [platelet], alpha polypeptide [GPIBA]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), gene analysis, common variant, HPA-2A/B (T145M)
81107	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), gene analysis, common variant, HPA-3A/B (I843S)
81108	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), gene analysis, common variant, HPA-4A/B (R143Q)

	81109	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), gene analysis, common variant (eg, HPA-5A/B (K505E))
	81110	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), gene analysis, common variant, HPA-6A/B (R489Q)
	81111	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), gene analysis, common variant, HPA-9A/B (V837M)
	81112	Human platelet antigen 15 genotyping (HPA-15), CD109 (CD109 molecule) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), gene analysis, common variant, HPA-15A/B (S682Y)
	81161	DMD (dystrophin) (e.g., Duchenne/Becker muscular dystrophy) deletion analysis and duplication analysis, if performed
	81171	AFF2 (ALF transcription elongation factor 2 [FMR2]) (eg, fragile X intellectual disability 2 [FRAXE]) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
	81172	AFF2 (ALF transcription elongation factor 2 [FMR2]) (eg, fragile X intellectual disability 2 [FRAXE]) gene analysis; characterization of alleles (eg, expanded size and methylation status)
	81173	AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; full gene sequence
	81174	AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; known familial variant
	81177	ATN1 (atrophin 1) (eg, dentatorubral-pallidoluysian atrophy) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81178	ATXN1 (ataxin 1) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81179	ATXN2 (ataxin 2) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81180	ATXN3 (ataxin 3) (eg, spinocerebellar ataxia, Machado-Joseph disease) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81181	ATXN7 (ataxin 7) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81182	ATXN8OS (ATXN8 opposite strand [non-protein coding]) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81183	ATXN10 (ataxin 10) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81184	CACNA1A (calcium voltage-gated channel subunit alpha1 A) (eg, spinocerebellar ataxia) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
	81185	CACNA1A (calcium voltage-gated channel subunit alpha1 A) (eg, spinocerebellar ataxia) gene analysis; full gene sequence
	81186	CACNA1A (calcium voltage-gated channel subunit alpha1 A) (eg, spinocerebellar ataxia) gene analysis; known familial variant
	81187	CNBP (CCHC-type zinc finger nucleic acid binding protein) (eg, myotonic dystrophy type 2) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
	81188	CSTB (cystatin B) (eg, Unverricht-Lundborg disease) gene analysis; evaluation to detect abnormal (eg, expanded) alleles

81189	CSTB (cystatin B) (eg, Unverricht-Lundborg disease) gene analysis; full gene sequence
81190	CSTB (cystatin B) (eg, Unverricht-Lundborg disease) gene analysis; known familial variant(s)
81200	ASPA (aspartoacylase) (eg, Canavan disease) gene analysis, common variants (eg, E285A, Y231X)
81204	AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; characterization of alleles (eg, expanded size or methylation status)
81205	BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (eg, maple syrup urine disease) gene analysis, common variants (eg, R183P, G278S, E422X)
81209	BLM (Bloom syndrome, RecQ helicase-like) (eg, Bloom syndrome) gene analysis, 2281del6ins7 variant
81220	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)
81221	;known familial variants
81222	;duplication/deletion variants
81223	;full gene sequence
81224	;ntron 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	;interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants for chromosomal abnormalities
81234	DMPK (DM1 protein kinase) (eg, myotonic dystrophy type 1) gene analysis; evaluation to detect abnormal (expanded) alleles
81238	F9 (coagulation factor IX) (eg, hemophilia B), full gene sequence
81239	DMPK (DM1 protein kinase) (eg, myotonic dystrophy type 1) gene analysis; characterization of alleles (eg, expanded size)
81240	F2 (prothrombin, coagulation factor II) (eg, hereditary hypercoagulability) gene analysis, 20210G>A variant
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 messenger ribonucleoprotein 1) (eg, fragile X syndrome, X-linked intellectual disability [XLID]) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81244	FMR1 (fragile X messenger ribonucleoprotein 1) (eg, fragile X syndrome, X-linked intellectual disability [XLID]) gene analysis; characterization of alleles (eg, expanded size and promoter methylation status)
81247	G6PD (glucose-6-phosphate dehydrogenase) (eg, hemolytic anemia, jaundice), gene analysis; common variant(s) (eg, a, a-)
81248	;known familial variant(s)
81249	;full gene sequence
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)

81252	GJB2 (gap junction protein, beta 2, 26kDa, connexin 26) (eg, nonsyndromic hearing loss) gene analysis; full gene sequence
81253	;known familial variants
81254	GJB6 (gap junction protein, beta 6, 30kDa, connexin 30) (eg, nonsyndromic hearing loss) gene analysis, common variants (eg, 309kb [del(GJB6-D13S1830)] and 232kb [del(GJB6-D13S1854)])
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)
81258	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; known familial variant
81259	;full gene sequence
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)
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	;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	;with cell selection (eg, CD3, CD33), each cell type
81269	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; duplication/deletion variants
81271	HTT (huntingtin) (eg, Huntington disease) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81274	HTT (huntingtin) (eg, Huntington disease) gene analysis; characterization of alleles (eg, expanded size)
81284	FXN (frataxin) (eg, Friedreich ataxia) gene analysis; evaluation to detect abnormal (expanded) alleles
81285	FXN (frataxin) (eg, Friedreich ataxia) gene analysis; characterization of alleles (eg, expanded size)
81286	FXN (frataxin) (eg, Friedreich ataxia) gene analysis; full gene sequence
81289	FXN (frataxin) (eg, Friedreich ataxia) gene analysis; known familial variant(s)
81290	MCOLN1 (mucolipin 1) (eg, Mucolipidosis, 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)
81302	MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; full sequence analysis
81303	;known familial variant

81304	;duplication/deletion variants
81312	PABPN1 (poly[A] binding protein nuclear 1) (eg, oculopharyngeal muscular dystrophy) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81324	PMP22 (peripheral myelin protein 22) (eg, Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; duplication/deletion analysis
81325	;full sequence analysis
81326	;known familial variant
81329	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; dosage/deletion analysis (eg, carrier testing), includes SMN2 (survival of motor neuron 2, centromeric) analysis, if performed
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)
81333	TGFBI (transforming growth factor beta-induced) (eg, corneal dystrophy) gene analysis, common variants (eg, R124H, R124C, R124L, R555W, R555Q)
81336	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; full gene sequence
81337	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; known familial sequence variant(s)
81343	PPP2R2B (protein phosphatase 2 regulatory subunit Bbeta) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81344	TBP (TATA box binding protein) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81349	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and loss-of-heterozygosity variants, low-pass sequencing analysis
81350	UGT1A1 (UDP glucuronosyltransferase 1 family, polypeptide A1) (eg, hyperbilirubinemia [Gilbert syndrome]), gene analysis, common variants (eg, *28, *36, *37)
81361	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); common variant(s) (eg, HBS, HBC, HBE)
81362	;known familial variant(s)
81363	;duplication/deletion variant(s)
81364	;full gene sequence
81370	HLA Class I and II typing, low resolution (eg, antigen equivalents); HLA-A, -B, -C, -DRB1/3/4/5, and -DQB1
81371	;HLA-A, -B, and -DRB1 (eg, verification typing)
81372	HLA Class I typing, low resolution (eg, antigen equivalents); complete (ie, HLA-A, -B, and -C)
81373	;one locus (eg, HLA-A, -B, or -C), each
81374	;one antigen equivalent (eg, B*27), each
81375	HLA Class II typing, low resolution (eg, antigen equivalents); HLA-DRB1/3/4/5 and -DQB1

81376	;one locus (eg, HLA-DRB1, -DRB3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1), each
81377	;one 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	;one locus (eg, HLA-A, -B, or -C), each
81381	;one allele or allele group (eg, B*57:01P), each
81382	HLA Class II typing, high resolution (ie, alleles or allele groups); one locus (eg, HLA-DRB1, -DRB3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1), each
81383	;one allele or allele group (eg, HLA-DQB1*06:02P), each
81400	Molecular pathology procedure, Level 1
81401	Molecular pathology procedure, Level 2
81402	Molecular pathology procedure, Level 3
81403	Molecular pathology procedure, Level 4
81404	Molecular pathology procedure, Level 5
81405	Molecular pathology procedure, Level 6
81406	Molecular pathology procedure, Level 7
81407	Molecular pathology procedure, Level 8
81408	Molecular pathology procedure, Level 9
81410	Aortic dysfunction or dilation (eg, Marfan syndrome, Loeys Dietz syndrome, Ehler Danlos syndrome type IV, arterial tortuosity syndrome); genomic sequence analysis panel, must include sequencing of at least 9 genes, including FBN1, TGFBR1, TGFBR2, COL3A1, MYH11, ACTA2, SLC2A10, SMAD3, and MYLK
81411	;duplication/deletion analysis panel, must include analyses for TGFBR1, TGFBR2, MYH11, and COL3A1
81412	Ashkenazi Jewish associated disorders (eg, Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia group C, Gaucher disease, Tay-Sachs disease), genomic sequence analysis panel, must include sequencing of at least 9 genes, including ASPA, BLM, CFTR, FANCC, GBA, HEXA, IKBKAP, MCOLN1, and SMPD1
81413	Cardiac ion channelopathies (eg, Brugada syndrome, long QT syndrome, short QT syndrome, catecholaminergic polymorphic ventricular tachycardia); genomic sequence analysis panel, must include sequencing of at least 10 genes, including ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, and SCN5A
81414	;duplication/deletion gene analysis panel, must include analysis of at least 2 genes, including KCNH2 and KCNQ1
81415	Exome (eg, unexplained constitutional or heritable disorder or syndrome); sequence analysis
81416	;sequence analysis, each comparator exome (eg, parents, siblings) (List separately in addition to code for primary procedure)
81417	;re-evaluation of previously obtained exome sequence (eg, updated knowledge or unrelated condition/syndrome)
81419	Epilepsy genomic sequence analysis panel, must include analyses for ALDH7A1, CACNA1A, CDKL5, CHD2, GABRG2, GRIN2A, KCNQ2, MECP2, PCDH19, POLG, PRRT2, SCN1A, SCN1B, SCN2A, SCN8A, SLC2A1, SLC9A6, STXBP1, SYNGAP1, TCF4, TPP1, TSC1, TSC2, and ZEB2
81420	Fetal chromosomal aneuploidy (eg, trisomy 21, monosomy X) genomic sequence analysis panel, circulating cell-free fetal DNA in maternal blood, must include analysis of chromosomes 13, 18, and 21

81422	Fetal chromosomal microdeletion(s) genomic sequence analysis (eg, DiGeorge syndrome, Cri-du-chat syndrome), circulating cell-free fetal DNA in maternal blood
81425	Genome (eg, unexplained constitutional or heritable disorder or syndrome); sequence analysis
81426	;sequence analysis, each comparator genome (eg, parents, siblings) (List separately in addition to code for primary procedure)
81427	;re-evaluation of previously obtained genome sequence (eg, updated knowledge or unrelated condition/syndrome)
81430	Hearing loss (eg, nonsyndromic hearing loss, Usher syndrome, Pendred syndrome); genomic sequence analysis panel, must include sequencing of at least 60 genes, including CDH23, CLRN1, GJB2, GPR98, MTRNR1, MYO7A, MYO15A, PCDH15, OTOF, SLC26A4, TMC1, TMPRSS3, USH1C, USH1G, USH2A, and WFS1
81431	;duplication/deletion analysis panel, must include copy number analyses for STRC and DFNB1 deletions in GJB2 and GJB6 genes
81434	Hereditary retinal disorders (eg, retinitis pigmentosa, Leber congenital amaurosis, cone-rod dystrophy), genomic sequence analysis panel, must include sequencing of at least 15 genes, including ABCA4, CNGA1, CRB1, EYS, PDE6A, PDE6B, PRPF31, PRPH2, RDH12, RHO, RP1, RP2, RPE65, RPGR, and USH2A
81439	Inherited cardiomyopathy (eg, hypertrophic cardiomyopathy, dilated cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy) genomic sequence analysis panel, must include sequencing of at least 5 cardiomyopathy-related genes, including DSG2, MYBPC3, MYH7, PKP2, and TTN
81440	Nuclear encoded mitochondrial genes (eg, neurologic or myopathic phenotypes), genomic sequence panel, must include analysis of at least 100 genes, including BCS1L, C10orf2, COQ2, COX10, DGUOK, MPV17, OPA1, PDSS2, POLG, POLG2, RRM2B, SCO1, SCO2, SLC25A4, SUCLA2, SUCLG1, TAZ, TK2, and TYMP
81441	Inherited bone marrow failure syndromes (IBMFS) (eg, Fanconi anemia, dyskeratosis congenita, Diamond-Blackfan anemia, Shwachman-Diamond syndrome, GATA2 deficiency syndrome, congenital amegakaryocytic thrombocytopenia) sequence analysis panel, must include sequencing of at least 30 genes, including BRCA2, BRIP1, DKC1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA1, GATA2, MPL, NHP2, NOP10, PALB2, RAD51C, RPL11, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS7, SBDS, TERT, and TINF2
81442	Noonan spectrum disorders (eg, Noonan syndrome, cardio-facio-cutaneous syndrome, Costello syndrome, LEOPARD syndrome, Noonan-like syndrome), genomic sequence analysis panel, must include sequencing of at least 12 genes, including BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, and SOS1
81443	Genetic testing for severe inherited conditions (eg, cystic fibrosis, Ashkenazi Jewish-associated disorders [eg, Bloom syndrome, Canavan disease, Fanconi anemia type C, mucolipidosis type VI, Gaucher disease, Tay-Sachs disease], beta hemoglobinopathies, phenylketonuria, galactosemia), genomic sequence analysis panel, must include sequencing of at least 15 genes (eg, ACADM, ARSA, ASPA, ATP7B, BCKDHA, BCKDHB, BLM, CFTR, DHCR7, FANCC, G6PC, GAA, GALT, GBA, GBE1, HBB, HEXA, IKBKAP, MCOLN1, PAH)

81448	Hereditary peripheral neuropathies (eg, Charcot-Marie-Tooth, spastic paraplegia), genomic sequence analysis panel, must include sequencing of at least 5 peripheral neuropathy-related genes (eg, BSCL2, GJB1, MFN2, MPZ, REEP1, SPAST, SPG11, SPTLC1)
81460	Whole mitochondrial genome (eg, Leigh syndrome, mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes [MELAS], myoclonic epilepsy with ragged-red fibers [MERFF], neuropathy, ataxia, and retinitis pigmentosa [NARP], Leber hereditary optic neuropathy [LHON]), genomic sequence, must include sequence analysis of entire mitochondrial genome with heteroplasmy detection
81465	Whole mitochondrial genome large deletion analysis panel (eg, Kearns-Sayre syndrome, chronic progressive external ophthalmoplegia), including heteroplasmy detection, if performed
81470	X-linked intellectual disability (XLID) (eg, syndromic and non-syndromic XLID); genomic sequence analysis panel, must include sequencing of at least 60 genes, including ARX, ATRX, CDKL5, FGD1, FMR1, HUWE1, IL1RAPL, KDM5C, L1CAM, MECP2, MED12, MID1, OCRL, RPS6KA3, and SLC16A2
81471	;duplication/deletion gene analysis, must include analysis of at least 60 genes, including ARX, ATRX, CDKL5, FGD1, FMR1, HUWE1, IL1RAPL, KDM5C, L1CAM, MECP2, MED12, MID1, OCRL, RPS6KA3, and SLC16A2
81479	Unlisted molecular pathology procedure
81493	Coronary artery disease, mRNA, gene expression profiling by real-time RT-PCR of 23 genes, utilizing whole peripheral blood, algorithm reported as a risk score
81506	Endocrinology (type 2 diabetes), biochemical assays of seven analytes (glucose, HbA1c, insulin, hs-CRP, adiponectin, ferritin, interleukin 2-receptor alpha), utilizing serum or plasma, algorithm reporting a risk score
81507	Fetal aneuploidy (trisomy 21, 18, and 13) DNA sequence analysis of selected regions using maternal plasma, algorithm reported as a risk score for each trisomy
81508	Fetal congenital abnormalities, biochemical assays of two proteins (PAPP-A, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score
81509	Fetal congenital abnormalities, biochemical assays of three proteins (PAPP-A, hCG [any form], DIA), utilizing maternal serum, algorithm reported as a risk score
81510	Fetal congenital abnormalities, biochemical assays of three analytes (AFP, uE3, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score
81511	Fetal congenital abnormalities, biochemical assays of four analytes (AFP, uE3, hCG [any form], DIA) utilizing maternal serum, algorithm reported as a risk score (may include additional results from previous biochemical testing)
81512	Fetal congenital abnormalities, biochemical assays of five analytes (AFP, uE3, total hCG, hyperglycosylated hCG, DIA) utilizing maternal serum, algorithm reported as a risk score
81513	Infectious disease, bacterial vaginosis, quantitative real-time amplification of RNA markers for Atopobium vaginae, Gardnerella vaginalis, and Lactobacillus species, utilizing vaginal-fluid specimens, algorithm reported as a positive or negative result for bacterial vaginosis
81514	Infectious disease, bacterial vaginosis and vaginitis, quantitative real-time amplification of DNA markers for Gardnerella vaginalis, Atopobium vaginae, Megasphaera type 1, Bacterial Vaginosis Associated Bacteria-2 (BVAB-2), and Lactobacillus species (L. crispatus and L. jensenii), utilizing vaginal-fluid specimens, algorithm reported as a positive or negative for high likelihood of

		bacterial vaginosis, includes separate detection of Trichomonas vaginalis and/or Candida species (C. albicans, C. tropicalis, C. parapsilosis, C. dubliniensis), Candida glabrata, Candida krusei, when reported
	81554	Pulmonary disease (idiopathic pulmonary fibrosis [IPF]), mRNA, gene expression analysis of 190 genes, utilizing transbronchial biopsies, diagnostic algorithm reported as categorical result (eg, positive or negative for high probability of usual interstitial pneumonia [UIP])
	81599	Unlisted multianalyte assay with algorithmic analysis
	84999	Unlisted chemistry procedure
	88271	Molecular cytogenetics; DNA probe, each (eg, FISH)
	88273	;chromosomal in situ hybridization, analyze 10-30 cells (eg, for microdeletions)
	88274	;interphase in situ hybridization, analyze 25-99 cells
	88275	;interphase in situ hybridization, analyze 100-300 cells
	88291	Cytogenetics and molecular cytogenetics, interpretation and report
HCPCS	G0452	Molecular pathology procedure; physician interpretation and report