Regence

Medical Policy Manual Genetic Testing, Policy No. 64

Evaluating the Utility of Genetic Panels

Effective: January 1, 2025

Next Review: July 2025
Last Review: January 2025

IMPORTANT REMINDER

Medical Policies are developed to provide guidance for members and providers regarding coverage in accordance with contract terms. Benefit determinations are based in all cases on the applicable contract language. To the extent there may be any conflict between the Medical Policy and contract language, the contract language takes precedence.

PLEASE NOTE: Contracts exclude from coverage, among other things, services or procedures that are considered investigational or cosmetic. Providers may bill members for services or procedures that are considered investigational or cosmetic. Providers are encouraged to inform members before rendering such services that the members are likely to be financially responsible for the cost of these services.

DESCRIPTION

Genetic panel tests evaluate many genes simultaneously, and have been developed for numerous indications, including hereditary cancer risk assessment, pharmacogenetics, and diagnosis of congenital disorders. Many panel tests include genes that do not have demonstrated clinical utility for their testing.

MEDICAL POLICY CRITERIA

Note: Where applicable, specific policies that have criteria and evidence used to review genetic panel tests are noted (see *Policy Cross-References* in the table below).

When there is not enough research to show that a gene and/or gene variant in a genetic panel test may be used to manage treatment decisions and improve net health outcomes, then the entire genetic panel test is considered **investigational**, including but not limited to the following (with or without any optional add-on genes or components):

Test Name	Laboratory	Policy Cross-Reference
Abnormal Genitalia/ Disorders of Sex Development Panel	Blueprint Genetics	None
Aeon Pain Management PGX Profile	Aeon Clinical Laboratories	GT10

Amyotrophic Lateral Sclerosis Advanced Evaluation Gene Panel	Athena Diagnostics	None
Amyotrophic Lateral Sclerosis Panel	Laboratory for Precision Diagnostics, University of Washington	None
Amyotrophic Lateral Sclerosis / Frontotemporal Lobar Degeneration Panel	GeneDx	None
Arthrogryposes Panel	Blueprint Genetics	None
ASD/ID Genetic Test Panel	Quadrant Laboratories	None
Ataxia Panel	Blueprint Genetics	None
Ataxia Repeat Expansion Panel	University of Chicago	None
Ataxia Complete Recessive Evaluation	Athena Diagnostics	None
Ataxia, Comprehensive Evaluation	Athena Diagnostics	None
Ataxia/Episodic Ataxia Disorders (including any add-on components, e.g., mtDNA, SCA, HTT, FRDA Repeat Expansion Analysis)	Labcorp/MNG Laboratories	None
Ataxia Xpanded Panel	GeneDx	None
Autism, Intellectual Disability, and Developmental Delay Panel	Cincinnati Children's Hospital	None
Autism Spectrum Disorders Panel	Prevention Genetics	None
AutismNext	Ambry Genetics™	None
Autism/ID and Autism/ID Xpanded Panel	GeneDx	None
Autoinflammatory Primary Immunodeficiency (PID) Gene Panel	Mayo Clinic	None
Autoinflammatory Syndrome Panel	Blueprint Genetics	None
Autosomal Dominant Thrombocytopenia Panel	Versiti	None
Bacterial Typing by Whole Genome Sequencing	Mayo Clinic	None
Bartter Syndrome Panel	Blueprint Genetics	None
Beacon Expanded Carrier Panels (with or without X-linked disorders)	Fulgent	GT81
Bleeding Disorders Panel	Prevention Genetics	None
Bone Marrow Failure Panel	Oregon Health & Science University, Knight Diagnostic Lab	None
Bone Marrow Failure Syndrome Panel	Blueprint Genetics	None

Bone Marrow Failure Syndromes	Cincinnati Children's Hospital	None
Bone Marrow Failure Syndromes Panel	Children’s Hospital of Philadelphia	None
BrainTumorNext	Ambry Genetics™	None
BRCANext-Expanded (with or without +RNAinsight)	Ambry Genetics™	GT02
BRCAPlus and BRCAPlus Expanded Panel	Ambry Genetics™	GT02
BROCA Cancer Risk Panel	University of Washington	GT02
CancerNext™ and CancerNext™ Expanded	Ambry Genetics™	None
CancerNext™ +RNAinsight™	Ambry Genetics™	None
CancerTYPE ID®	bioTheranostics	GT15
Cardiac Arrhythmia Panel	Laboratory for Precision Diagnostics, University of Washington	None
Cardiomyopathy Panel	Laboratory for Precision Diagnostics, University of Washington	None
Cardiomyopathy Panel	GeneDx	None
CardioNext	Ambry Genetics™	None
Carrier Screening Full Panel	Atlas Genomics	GT81
Cataract Panel Test	Blueprint Genetics	None
CentoNeuro Panel	Centogene	None
Cholestasis Panel	Oregon Health & Science University	None
Chronic Lymphocytic Leukemia (CLL) Panel	ARUP	None
Ciliopathies Panels	Oregon Health & Science University	None
Cleft Lip/Cleft Palate Panel	Prevention Genetics	None
Cleft Lip/Palate and Associated Syndromes Panel	Blueprint Genetics	None
CMNext Panel	Ambry Genetics™	None
Coagulation Disorder Panel	Versiti	None
Cobalamin/Propionate/Homocysteine Metabolism Related Disorders Panel	ARUP	None
ColoNext™ and ColoNext™ +RNAinsight™	Ambry Genetics™	GT06

Colorectal Cancer Panel	GeneDx	GT06
ColoSeq™ Lynch and Polyposis	University of Washington	GT06
Combined Cardiac Panel	GeneDx	None
Combined Hereditary Dementia and Amyotrophic Lateral Sclerosis Panel	Invitae	None
Common Cancer Management Panel	GeneDx	None
Comprehensive Arrhythmia/Cardiomyopathy Panel	Laboratory for Precision Diagnostics, University of Washington	None
Comprehensive Bleeding Disorder Panel	Versiti	None
Comprehensive Brain Malformation Panel	Prevention Genetics	None
Comprehensive Brain Malformations Panel	GeneDx	None
Comprehensive Breast Cancer Panel	Genetics Center	GT02
Comprehensive Common Cancer Panel	GeneDx	None
Comprehensive Congenital Heart Disease Panel	Prevention Genetics	None
Comprehensive Dystonia Panel	Labcorp/MNG Laboratories	None
Comprehensive Hematology and Hereditary Cancer Panel	Blueprint Genetics	None
Comprehensive Hereditary Cancer Panel	Blueprint Genetics	None
Comprehensive Hereditary Cancer Panel	Quest Diagnostics	None
Comprehensive Hereditary Spastic Paraplegia (HSP) Panel	GeneDx	None
Comprehensive Immune and Cytopenia Panel	Blueprint Genetics	None
Comprehensive Inherited Kidney Disease Panel	Prevention Genetics	None
Comprehensive Inherited Retinal Dystrophies Panel	Prevention Genetics	None
Comprehensive Ocular Disorders (includes RPGR ORF15) Panel	Prevention Genetics	None
Comprehensive Neuromuscular Panel	Prevention Genetics	None
Comprehensive Personalized Medicine Panel	Alpha Genomix Laboratories	GT10
Comprehensive Pharmacogenetic Panel	Advanced Genomics	GT10

Comprehensive Platelet Disorder Panel	Versiti	None
Comprehensive Short Stature Syndrome Panel	Blueprint Genetics	None
Comprehensive Skeletal Dysplasias and Disorders Panel	Blueprint Genetics	None
Comprehensive Spinocerebellar Ataxia Repeat Expansion Panel	Labcorp/MNG Laboratories	None
Congenital Abnormalities of the Kidney Tract (CAKUT) Panel	Prevention Genetics	None
Congenital Adrenal Hyperplasia (CAH) Panel	Prevention Genetics	None
Congenital Adrenal Hyperplasia NGS Panel	Fulgent	None
Congenital Adrenal Hyperplasia Panel	Blueprint Genetics	None
Congenital Anomalies of the Gastrointestinal Tract Panel	Prevention Genetics	None
Congenital Central Hypoventilation Panel	Prevention Genetics	None
Congenital Diaphragmatic Hernia Panel	Prevention Genetics	None
Congenital Hypothyroidism and Thyroid Hormone Resistance Panel	Prevention Genetics	None
Congenital Limb Malformation Panel	Prevention Genetics	None
Congenital Myopathy and Muscular Dystrophy Panel	GeneDx	None
Congenital Stationary Night Blindness Panel	Prevention Genetics	None
Cornelia de Lange and Related Disorders Panel	Prevention Genetics	None
Cortical Brain Malformation Panel	GeneDx	None
Craniosynostosis NGS Panel	Connective Tissue Gene Tests (CTGT)	None
Craniosynostosis NGS Panel	Fulgent	None
Cystic Kidney and Liver Diseases Panel	GeneDx	None
Cystic Kidney Disease Panel	Blueprint Genetics	None
DecisionDx-UMSeq	Castle Biosciences	None
DetoxiGenomic® Profile Test	Genova® Diagnostics	GT10
Differences in Sex Development Sequencing	Seattle Children’s Hospital	None

Differences of Sex Development (DSD) Panel	Prevention Genetics	None
Distal Arthrogryposis Sequencing Panel	University of Chicago Genetics Services Laboratories	None
Distal Arthrogryposis Panel	Prevention Genetics	None
Dystonia and Choreatic Movement Disorder Panel	University of Washington	None
Dystonia Panel	GeneDx	None
Early Advantage Panel	NxGEN MDx	GT82
Ectrodactyly/Split Hand-Split Foot Malformation Panel	GeneDx	None
Empower Multi-Cancer and Multi-Cancer Expanded and Comprehensive Panels	Natera, Inc.	None
Episodic Pain Syndrome Sequencing Panel	Prevention Genetics	None
Familial Hemiplegic Migraine and Alternating Hemiplegia of Childhood Panel	Prevention Genetics	None
Female Infertility NGS Panel	Fulgent	None
Fibrinolytic Disorder Panel	Versiti	None
Foresight™ Carrier Screen Universal Panel and Universal Panel Plus	Myriad	GT81
Full Hereditary Cancer Panel	myTest Diagnostics	None
FusionPlex Pan-Heme Panel	Laboratory for Precision Diagnostics, University of Washington	GT59
GenArray™	GenPath Diagnostics	None
GeneAware Complete Panel	Miraca, Baylor Genetics	GT81
GeneDose™	Coriell Life Sciences	GT10
GeneSeq®: Cardio-Early-onset Coronary Artery Disease/Familial Hypercholesterolemia Profile	Labcorp	GT11
GeneSight® Psychotropic Genetic Testing	Assurex Health/Myriad	GT53
Genetic Platelet Disorders Panel	Labcorp	None
GeneticsNow® Comprehensive Germline Panel	GoPath	None
GeneTrails® Comprehensive Heme Panel (previously GeneTrails®	Oregon Health & Science Univ	GT59

Hematologic Malignancies 220 Gene Panel)		
Genomic Unity® Ataxia Repeat Expansion Analysis	Variantyx	None
Genomic Unity® Comprehensive Ataxia Analysis	Variantyx	None
Genomic Unity Movement Disorders Analysis	Variantyx	None
Genomind® Professional PGx Express™	Genomind LLC	GT53
Guideline-based Hereditary Cancer Panel	Quest Diagnostics	None
GxVision Comprehensive Inherited Cancer Gene Test	Otogenetics	None
Hematologic Malignancies Panel	Penn Medicine	GT59
Heme-STAMP	Stanford University	GT59
Hereditary Breast Cancer Panel	Quest Diagnostics	GT02
Hereditary Cancer Predisposition Panel	Magee-Women's Hospital	None
Hereditary Hemochromatosis Panel	Prevention Genetics	None
Hereditary Hemorrhagic Telangiectasia and Vascular Malformations Gene Panel	Asante Lab	None
Hereditary Leukemia Panel	Blueprint Genetics	None
Hereditary Ovarian Cancer Panel	Prevention Genetics	GT02
HopeSeq HemeComplete	City of Hope National Medical Center	GT59
Horizon™ 27	Natera, Inc.	GT81
Horizon™ 106	Natera, Inc.	GT81
Horizon™ 274	Natera, Inc.	GT81
Horizon™ 421	Natera, Inc.	GT81
HSP, Comprehensive Evaluation	Athena Diagnostics	None
Hydrocephalus Panel	Prevention Genetics	None
Hydrops Sequencing Panel	Greenwood Genetics	None
Hyperparathyroidism Panel	Blueprint Genetics	None
Hypoglycemia Panel - Expanded	Prevention Genetics	None
Hypogonadotropic Hypogonadism/ Kallmann Syndrome Panel	Prevention Genetics	None
Hypogonadotropic Hypogonadism Panel	GeneDx	None
InheriGen Panel and InheriGen Plus	GenPath Diagnostics	GT81

Inherited Bone Marrow Failure Panel	Prevention Genetics	None
Inherited Pancreatic Cancer Panel	Oregon Health & Science University, Knight Diagnostic Lab	None
Inherited Thrombocytopenia Panel	Versiti	None
Inheritest Ashkenazi Jewish Carrier Screening Panel	LabCorp/Integrated Genetics	GT81
Inheritest Carrier Screen, Comprehensive Panel	LabCorp/Integrated Genetics	GT81
Inheritest 100 PLUS Panel, 300 PLUS Panel and 500 PLUS Panel	LabCorp/Integrated Genetics	GT81
Intellectual Disability, Epilepsy, and Autism (IDEA) Panel	Prevention Genetics	None
Invitae Amyotrophic Lateral Sclerosis Panel	Invitae	None
Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel	Invitae	None
Invitae Arrhythmia Comprehensive Panel	Invitae	None
Invitae Autoinflammatory and Autoimmunity Syndromes Panel	Invitae	None
Invitae Bone Marrow Failure Syndromes Panel	Invitae	None
Invitae Brain Malformations Panel	Invitae	None
Invitae Breast and Gyn Cancers Guidelines-Based Panel	Invitae	None
Invitae Breast Cancer Guidelines-Based Panel	Invitae	GT02
Invitae Breast Cancer Panel	Invitae	GT02
Invitae Broad Carrier Screen	Invitae	GT81
Invitae Cancer Screen	Invitae	None
Invitae Cataracts Panel	Invitae	None
Invitae Cerebral Palsy Spectrum Disorders Panel	Invitae	None
Invitae Cholestasis Panel	Invitae	None
Invitae Ciliopathies Panel	Invitae	None
Invitae Colorectal Cancer Panel	Invitae	None
Invitae Common Hereditary Cancer Panel	Invitae	GT02

Invitae Comprehensive Carrier Screen	Invitae	GT81
Invitae Comprehensive Lipidemia Panel	Invitae	None
Invitae Comprehensive Muscular Dystrophy Panel	Invitae	None
Invitae Comprehensive Myopathy Panel	Invitae	None
Invitae Comprehensive Neuromuscular Disorders Panel	Invitae	None
Invitae Comprehensive Neuropathies Panel	Invitae	None
Invitae Comprehensive Porphyrias Panel	Invitae	None
Invitae Congenital Anomalies of Kidney and Urinary Tract (CAKUT) Panel	Invitae	None
Invitae Congenital Heart Defects and Heterotaxy Panel	Invitae	None
Invitae Congenital Heart Disease Panel	Invitae	None
Invitae Congenital Muscular Dystrophy Panel	Invitae	None
Invitae Congenital Myasthenic Syndrome Panel	Invitae	None
Invitae Cornelia de Lange and Related Disorders Panel	Invitae	None
Invitae Cystic Kidney Disease Panel	Invitae	None
Invitae Dystonia Comprehensive Panel	Invitae	None
Invitae Ectodermal Dysplasias and Related Disorders Panel	Invitae	None
Invitae Elevated Phenylalanine (Hyperphenylalaninemia) Panel	Invitae	None
Invitae Elevated Very Long chain Fatty Acids Panel (including X-ALD)	Invitae	None
Invitae Epidermolysis Bullosa and Palmoplantar Keratoderma Panel	Invitae	None
Invitae Expanded Renal Disease Panel	Invitae	None
Invitae Familial Hemiplegic Migraine Panel	Invitae	None
Invitae Frontotemporal Dementia Panel	Invitae	GT01
Invitae Gastric Cancer Panel	Invitae	None
Invitae Glaucoma Panel	Invitae	None

Invitae Hereditary Amyotrophic Lateral Sclerosis, Frontotemporal Dementia and Alzheimer Disease Panel	Invitae	None
Invitae Hereditary Breast and Gyn Cancers Panel	Invitae	GT02
Invitae Hereditary Cerebral Small Vessel Disease Panel	Invitae	None
Invitae Hereditary Hemochromatosis Panel	Invitae	None
Invitae Hereditary Nervous System/Brain Cancer Panel	Invitae	None
Invitae Hereditary Parkinson’s Disease and Parkinsonism Panel	Invitae	None
Invitae Hereditary Sarcoma Panel	Invitae	None
Invitae Hereditary Spastic Paraplegia Comprehensive Panel	Invitae	None
Invitae Hereditary Thrombophilia Panel	Invitae	None
Invitae Hereditary Thyroid Cancer Panel	Invitae	None
Invitae Hyperammonemia Panel	Invitae	None
Invitae Hypoglycemia Panel	Invitae	None
Invitae Hypogonadotrophic Hypogonadism Panel	Invitae	None
Invitae Hypoparathyroidism Panel	Invitae	None
Invitae Hypophosphatemia Panel	Invitae	None
Invitae Inborn Errors of Immunity and Cytopenias Panel	Invitae	None
Invitae Inherited Platelet Disorders Including Thrombocytopenia Panel	Invitae	None
Invitae Inherited Retinal Disorders Panel	Invitae	None
Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	Invitae	None
Invitae Limb and Digital Malformations Panel	Invitae	None
Invitae Melanoma Panel	Invitae	GT08
Invitae Metabolic Newborn Screening Confirmation Panel	Invitae	None
Invitae Methylmalonic Acidemia and Homocystinuria Panel	Invitae	None

Invitae Microphthalmia, Anophthalmia, Coloboma (MAC) and Anterior Segment Dysgenesis Panel	Invitae	None
Invitae Monogenic Diabetes Panel	Invitae	None
Invitae Multi-Cancer Panel and Multi-Cancer+RNA Panel	Invitae	None
Invitae Myelodysplastic Syndrome/Leukemia Panel	Invitae	None
Invitae Neonatal Respiratory Distress Panel	Invitae	None
Invitae Nephrolithiasis Panel	Invitae	None
Invitae Nephrotic Syndrome and Focal Segmental Glomerulosclerosis (FSGS) Panel	Invitae	None
Invitae Neurodegeneration with Brain Iron Accumulation Panel	Invitae	None
Invitae Neurodevelopmental Disorders (NDD) Panel	Invitae	None
Invitae Organic Acidemias Panel	Invitae	None
Invitae Overgrowth and Macrocephaly Syndromes Panel	Invitae	None
Invitae Overgrowth Syndromes Panel	Invitae	None
Invitae Pancreatic Cancer Panel	Invitae	None
Invitae Pediatric Solid Tumors Panel	Invitae	None
Invitae Periodic Paralysis Panel	Invitae	None
Invitae Phagocytic Disorders Including Neutropenia Panel	Invitae	None
Invitae Primary Immunodeficiency Panel	Invitae	None
Invitae Progressive Renal Disease Panel	Invitae	None
Invitae Prostate Cancer Panel	Invitae	None
Invitae Pulmonary Arterial Hypertension Panel	Invitae	None
Invitae RASopathies and Noonan Spectrum Disorders Panel	Invitae	None
Invitae Renal Tubular Disorders Panel	Invitae	None
Invitae Renal/Urinary Tract Cancers Panel	Invitae	None
Invitae Rett and Angelman Syndromes and Related Disorders Panel	Invitae	None

Invitae Rhabdomyolysis and Metabolic Myopathy Panel	Invitae	None
Invitae Skeletal Disorders Panel	Invitae	None
Kabuki Syndrome Panel	Prevention Genetics	None
Leukodystrophy and Leukoencephalopathy Panel	Blueprint Genetics	None
Leukodystrophy and Leukoencephalopathy Panel	Prevention Genetics	None
Leukoencephalopathy NGS Panel	Fulgent	None
Limb Abnormalities and Reduction Defects Panel	GeneDx	None
LipidSeq™	Boston Heart Diagnostics	GT11
Lipodystrophy NGS Panel	Fulgent	None
Low Bone Mass Panel	Baylor Genetics	None
Lymphoid Gene Panel by NGS	University of Washington	None
Metabolic Myopathies Panel	University of Washington	None
Metabolic Myopathies, Rhabdomyolysis, and Exercise Intolerance Panel	Prevention Genetics	None
Metabolic Myopathy and Rhabdomyolysis Panel	Blueprint Genetics	None
Metabolic Myopathy Panel	GeneDx	None
Microcephaly Panel and Microcephaly Xpanded Panel	GeneDx	None
Microcephaly Sequencing Panel	University of Chicago Genetics Services Laboratory	None
Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis Panel	Blueprint Genetics	None
Microphthalmia/Anophthalmia/Coloboma Panel	Prevention Genetics	None
Migraine and Stroke Panel	Oregon Health & Science University, Knight Diagnostic Lab	None
Migraine Panel	Blueprint Genetics	None
MODY Panel	Blueprint Genetics	None
Movement Disorder Ataxia Panel	Laboratory for Precision Diagnostics, University of Washington	None
MVL Vision Panel	Molecular Vision Laboratory	None

MyAML® 194 Targeted NGS Gene Panel	Invivoscribe	GT59
MyGenVar Pharmacogenomics Test	Geisinger Medical Laboratory	GT10
myMRD NGS Panel	Lab for Personalized Molecular Medicine	None
Myopathies and Myotonia, Muscular Dystrophies and Limb Girdle Panel	Laboratory for Precision Diagnostics, University of Washington	None
Myotonic Syndrome Advanced Evaluation Panel	Athena Diagnostics	None
myRisk™ Hereditary Cancer Panel (Update myRisk™)	Myriad	None
Nephrolithiasis Panel	Blueprint Genetics	None
Nephrotic Syncrome (NS)/Focal Segmental Glomerulosclerosis (FSGS) Panel	Prevention Genetics	None
Nephrotic Syndrome Panel	Blueprint Genetics	None
Neuromuscular Disorders Panel	GeneDx	None
Neuro-ophthalmology Panel	Blueprint Genetics	None
Neurotransmitter Metabolism Deficiency	Labcorp/MNG Laboratories	GT65
Neurotransmitter Metabolism Deficiency NGS Panel	Fulgent	None
NGS Hematology Molecular Profile	Sonora Quest Laboratories	GT59
Non-Immune Hydrops Fetalis Panel	Prevention Genetics	None
Non-NF1 RASopathy Panel	University of Alabama	None
NxGen MDx Hereditary Cancer Panel	NxGen MDx	None
NxGen Super Panel	NxGen MDx	GT81
OI and Genetic Bone Disorders Panel	Laboratory for Precision Diagnostics, University of Washington	None
OmniSeq® Immune Report Card	OmniSeq®	None
Optic Atrophy Panel	Blueprint Genetics	None
Optic Atrophy Panel	Prevention Genetics	None
Osteopetrosis and Dense Bone Dysplasia NGS/Del Dup Comprehensive Panel	Connective Tissue Gene Tests (CTGT)	None
Overgrowth and Macrocephaly Syndromes Panel	Prevention Genetics	None
Pan Cardiomyopathy Panel	Prevention Genetics	

Pan Cardiomyopathy Panel	Seattle Children's Hospital/Personalized Medicine Partners	None
PancNext™	Ambry Genetics™	None
Pancreatic Cancer Panel	GeneDx	None
Pediatric Cancer Panel	Prevention Genetics	None
Peroxisomal Disorders Panel	Prevention Genetics	None
Personalized Medicine Panel Comprehensive Panel	Capstone Healthcare	GT10
Personalized Medication Panel	UpFront Laboratories	GT10
Phagocytic Primary Immunodeficiency (PID) Gene Panel	Mayo Clinic	None
Pharmacogenetics PGx	Lineagen	GT10
Pharmacogenomics Panel	Quest Diagnostics	GT10
Platelet Disorders, Comprehensive Gene Panel	Mayo Clinic	None
Platelet Disorders Gene Sequencing Panel	Cincinnati Children's Human Genetics- Cytogenetics and Molecular Genetics Laboratories	None
Platelet Disorders Panel	Oregon Health & Science University	None
Platelet Function Disorder Panel	Versiti	None
Platelet Genex Functional Defect Panel	Machaon Diagnostics	None
Polycystic Kidney Disease Panel	Blueprint Genetics	None
Polycystic Liver Disease Panel	Prevention Genetics	None
Pontocerebellar Hypoplasia Panel	GeneDx	None
Porphyria Gene Panel	Blueprint Genetics	None
Premature Ovarian Failure Panel	Blueprint Genetics	None
Premature Ovarian Failure Panel	Prevention Genetics	None
Preventest	GeneID	None
Primary Antibody Deficiency Panel	ARUP	None
Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia (PCD) Panel	Blueprint Genetics	None
Primary Immunodeficiency Panel	Blueprint Genetics	None
Professional PGx and Professional PGx Express (CORE and FULL)	Genomind	GT53
ProstateNext™	Ambry Genetics™	GT17

ProstateNow®	GoPath	None
Psych HealthPGx Panel	RPRD Diagnostics	GT53
Psychiatric/Mental Health Panel	GeneID	GT53
Psychotropic Drug Profile	GeneTrait Laboratories	GT53
ProstateNext +RNAinsight™	Ambry Genetics™	GT17
Psychiatry/ADHD Panel	Alpha Genomix Laboratories	GT53
Pulmonary Arterial Hypertension Panel	ARUP	None
Pulmonary Arterial Hypertension Panel	GeneDx	None
PyloriAR™/AmHPR® H. pylori Antibiotic Resistance NGS Panel	American Molecular Labs	None
Qherit 381 Diseases, Male	Quest Diagnostics	GT81
Qherit 421 Diseases, Female	Quest Diagnostics	GT81
Qherit Extended (both Female and Male versions)	Quest Diagnostics	GT81
Qherit Plus, Female	Quest Diagnostics	GT81
RenalNext™ and RenalNext™+RNAinsight	Ambry Genetics™	None
Renasight Kidney Gene Panel	Natera, Inc.	None
Response Pharmacogenetics Testing	LabSolutions	GT10
Retinal Dystrophy Panel	Blueprint Genetics	None
Retinal Dystrophy Panel	Laboratory for Precision Diagnostics, University of Washington	None
Rett/Angelman Syndrome Sequencing Panel	Greenwood Genetic Center	None
Rett/Angelman Syndrome Sequencing Panel	Seattle Children’s Hospital	None
Rett/Angelman Syndrome Panel	GeneDx	None
RightMed® Panels and Gene Report/Medication Report (including the Mental Health, PGx16, and Comprehensive Tests with or without F2 and F5)	OneOme	GT10/GT53
Riskguard™	Exact Sciences	None
RNA Heme Fusion Panel	Seattle Children’s Hospital	None
Sarcoma Targeted Gene Fusion Panel	Mayo Clinic	None
Skeletal Disorders and Joint Problems Panel	Prevention Genetics	None
Somatic Overgrowth Panel	Washington University	None

Spastic Paraplegia	Labcorp/MNG Laboratories	None
Spastic Paraplegia (NGS Panel and Copy Number Analysis + mtDNA)	Labcorp/MNG Laboratories	None
Spinocerebellar Ataxia Repeat Expansion Panel	Labcorp/MNG Laboratories	None
Stargardt Disease (STGD) and Macular Dystrophies Panel	Prevention Genetics	None
Stroke, Cerebral Hemorrhage, Hemiplegia, and Migraines Panel	Prevention Genetics	None
Sudden Cardiac Arrest Panel	Prevention Genetics	None
Syndromic Autism Sequencing Panel	Greenwood Genetic Center	None
Tempus nP	Tempus	GT53
Tempus xG and xG+	Tempus	None
Thrombocytopenia Panel	Blueprint Genetics	None
Thrombosis Panel	Versiti	None
UroSeq	Know Error	None
VACTERL Association and Related Disorders Panel	Prevention Genetics	None
VanSeq Expanded Sequencing Panel	Seattle Children's Hospital	None
Vascular Malformations Panel	ARUP	None
VistaSeq Breast Cancer Panel	LabCorp	GT02
VistaSeq Hereditary Cancer Panel	LabCorp	None
VistaSeq Pancreatic Cancer Panel	LabCorp	None
VistaSeq Renal Cell Cancer Panel	LabCorp	None
Vitreoretinopathy NGS Panel	Connective Tissue Gene Tests (CTGT)	None
Vitreoretinopathy Panel	Molecular Vision Laboratory	None
Vitreoretinopathy Panel and Vitreoretinopathy Panel Plus	Blueprint Genetics	None
X-linked Intellectual Disability	Greenwood Genetic Center	None
YouScript® Personalized Prescribing System	Invitae	GT10

NOTE: A summary of the supporting rationale for the policy criteria is at the end of the policy.

LIST OF INFORMATION NEEDED FOR REVIEW

In order to determine the clinical utility of gene test(s), all of the following information must be submitted for review:

1. Name of the genetic test(s) or panel test
2. Name of the performing laboratory and/or genetic testing organization (more than one may be listed)
3. The exact gene(s) and/or variant(s) being tested
4. Relevant billing codes
5. Brief description of how the genetic test results will guide clinical decisions that would not otherwise be made in the absence testing
6. Medical records related to this genetic test, if available:
   - History and physical exam
   - Conventional testing and outcomes
   - Conservative treatment provided

CROSS REFERENCES

1. Medical Policy Manual: Genetic Testing Section Table of Contents

BACKGROUND

New genetic technology, such as next generation sequencing and chromosomal microarray, has led to the ability to examine many genes simultaneously.[1] This in turn has resulted in a proliferation of genetic panels. The intended use for these panels is variable. For example, for the diagnosis of hereditary disorders, a clinical diagnosis may already be established, and genetic testing is performed to determine whether there is a hereditary condition, and/or to determine the specific variant that is present. In other cases, there is a clinical syndrome (phenotype) with a broad number of potential diagnoses and genetic testing is used to make a specific diagnosis. For cancer panels, there are also different intended uses. Some panels may be intended to determine whether a known cancer is part of a hereditary cancer syndrome. Other panels may include somatic variants in a tumor biopsy specimen that may help identify a cancer type or subtype and/or help select best treatment.

Panels using next generation technology are currently available in the areas of cancer, cardiovascular disease, neurologic disease, psychiatric conditions, and for reproductive testing.[2-4] These panels are intuitively attractive to use in clinical care because they can screen for numerous variants within a single or multiple genes quickly, and may lead to greater efficiency in the work-up of genetic disorders. It is also possible that these “bundled” gene tests can be performed more cost effectively than direct sequencing, although this may not be true in all cases. However, panel testing also provides information on genetic variants that are of unclear clinical significance or which would not lead to changes in patient management.

One potential challenge of genetic panel testing is the availability of a large amount of ancillary genetic information, much of which has uncertain clinical consequences and management strategies. Identification of variants for which the clinical management is uncertain may lead to unnecessary follow-up testing and procedures, all of which have their own inherent risks.

Additionally, the design and composition of genetic panel tests have not been standardized. Composition of the panels is variable, and different commercial products for the same condition may test different sets of genes. The make-up of the panel is determined by the specific lab that has developed the test. In addition, the composition of any individual panel is likely to change over time, as new variants are discovered and added to the existing panels.

GENETIC COUNSELING

Due to the complexity of interpreting genetic test results, patients should receive pre- and post-test genetic counseling from a qualified professional when testing is performed to diagnose or predict susceptibility for inherited diseases. The benefits and risks of genetic testing should be fully disclosed to individuals prior to testing, and counseling concerning the test results should be provided.

REGULATORY STATUS

The majority of genetic panel tests are laboratory derived tests that are not subject to U.S. Food and Drug Administration (FDA) approval. Clinical laboratories may develop and validate tests in-house ("home-brew") and market them as a laboratory service; such tests must meet the general regulatory standards of the Clinical Laboratory Improvement Act (CLIA). The laboratory offering the service must be licensed by CLIA for high-complexity testing.

Note: Separate Medical Policies may apply to some specific genetic tests and panels not addressed in the criteria below. See the Genetic Testing Section of the Medical Policy Manual Table of Contents for additional genetic testing policies.

EVIDENCE SUMMARY

Human Genome Variation Society (HGVS) nomenclature[5] is used to describe variants found in DNA and serves as an international standard. It is being implemented for genetic testing medical evidence review updates starting in 2017. According to this nomenclature, the term "variant" is used to describe a change in a DNA or protein sequence, replacing previously-used terms, such as "mutation." Pathogenic variants are variants associated with disease, while benign variants are not. The majority of genetic changes have unknown effects on human health, and these are referred to as variants of uncertain significance.

Genetic cancer susceptibility panels utilizing next generation sequencing are best evaluated in the framework of a diagnostic test, as the test provides diagnostic information that assists in treatment decisions. The clinical utility of genetic panel testing refers to the likelihood that the panel will result in improved health outcomes.
For positive test results, the health benefits are related to interventions that reduce the risk of developing the disease, earlier or more intensive screening to detect and treat early disease symptoms, or interventions to improve quality of life.

- Alternatively, negative test results may prevent unnecessary intensive monitoring, invasive tests or procedures, or ineffective therapies.

For genetic panels that test for a broad number of variants, some components of the panel may be indicated based on the patient's clinical presentation and/or family history, while other components may not be indicated. The impact of test results related to non-indicated variants must be well-defined and take into account the possibility that the information may cause harm by leading to additional unnecessary interventions that would not otherwise be considered based on the patient's clinical presentation and/or family history.

Therefore, the focus of the following review is on evidence from well-designed controlled trials or large cohort studies that demonstrate the clinical utility of each panel test, i.e., the ability of results from the comprehensive genetic panels to:

1. Guide decisions in the clinical setting related to either treatment, management, or prevention; and

2. Improve health outcomes as a result of those decisions.

A limited body of literature exists on the potential clinical utility of available next generation sequencing (NGS) panels.

NONRANDOMIZED STUDIES

Desmond (2015) reported on an observational study assessing whether testing of hereditary cancer gene variants other than BRCA1/2 altered clinical management in a prospectively collected cohort of 1046 patients from three institutions who were negative for BRCA1/2.[6] Patients were tested with the 29-gene Hereditary Cancer Syndromes test (Invitae) or the 25-gene MyRisk test (Myriad Genetics). The investigators evaluated the likelihood of a post-test change in management considering gene-specific consensus management guidelines, gene-associated cancer risks, and personal and family history. Of this cohort, 40 patients (3.8%, 95% CI 2.8% to 5.2%) harbored deleterious variants, most commonly in moderate-risk breast and ovarian cancer genes and Lynch syndrome genes. Among 63 variant-positive patients, 20 were found to harbor variants in high-risk genes associated with detailed NCCN management guidelines which would change the pretest recommendations for screening and/or preventive surgery. However, the most common variants found were those in genes associated with low or moderately increased breast cancer risk (40 of 63 patients), where a change in management would be recommended for these patients in a minority of cases (10 of 40), involving either increased screening or preventive surgery. Since this study only reported anticipated changes in management, these variant-positive patients were not provided with these post-test recommendations. The investigators conceded that the potential clinical effect reported in this cohort is likely to apply only to an appropriately ascertained cohort, thereby limiting the generalizability of the results.

Kurian (2014) evaluated the information from a NGS panel of 42 cancer associated genes in women who had been previously referred for clinical BRCA1/2 testing after clinical evaluation of hereditary breast and ovarian cancer from 2002 to 2012.[7] The authors aimed to assess concordance of the results of the panel with prior clinical sequencing, the prevalence of potentially clinically actionable results, and the downstream effects on cancer screening and risk reduction. Potentially actionable results were defined as pathogenic variants that cause recognized hereditary caner syndromes or have a published association with a two-fold or greater relative risk of breast cancer compared to average risk women. In total, 198 women participated in the study. Of these, 174 had breast cancer and 57 carried 59 germline BRCA variants. Testing with the panel confirmed 57 of 59 of the pathogenic BRCA variants; of the two others, one was detected but reclassified as a VUS and the other was a large insertion that would not be picked up by NGS panel testing. Of the women who tested negative for *BRCA* variants (n=141), 16 had pathogenic variants in other genes (11.4%). The affected genes were *ATM* (n=2), *BLM* (n=1), *CDH1* (n=1), *CDKN2A* (n=1), *MLH1* (n=1), *MUTYH* (n=5), *NBN* (n=2), *PRSS1* (n=1), and *SLX4* (n=2). Eleven of these variants had been previously reported in the literature and five were novel. 80% of the women with pathogenic variants in the non BRCA1/2 genes had a personal history of breast cancer. Overall, a total of 428 VUS were identified in 39 genes, among 175 patients.

Six women with variants in *ATM*, *BLM*, *CDH1*, *NBN* and *SLX4* were advised to consider annual breast MRIs because of an estimated doubling of breast cancer risk, and six with variants in CDH1, MLH1 and MUTYH were advised to consider frequent colonoscopy and/or endoscopic gastroduodenoscopy (once every 1 to 2 years) due to estimated increases in

gastrointestinal cancer risk. One patient with a MLH1 variant consistent with Lynch syndrome underwent risk-reducing salpingo-oophorectomy and early colonoscopy which identified a tubular adenoma that was excised (she had previously undergone hysterectomy for endometrial carcinoma).

Mauer (2014) reported a single academic center's genetics program's experience with NGS panels for cancer susceptibility.[8] The authors conducted a retrospective review of the outcomes and clinical indications for the ordering of Ambry's next generation sequencing panels (BreastNext, OvaNext, ColoNext, and CancerNext) for patients seen for cancer genetics counseling from April 2012 to January 2013. Of 1,521 new patients seen for cancer genetics counseling, 1,233 (81.1%) had genetic testing. Sixty of these patients (4.9% of the total) had a next generation sequencing panel ordered, 54 of which were ordered as a second-tier test after single-gene testing was performed. Ten tests were cancelled due to out-of-pocket costs or previously identified variants. Of the 50 tests obtained, five were found to have a deleterious result (10%, compared with 131 [10.6%] of the 1,233 single-gene tests ordered at the same center during the study time frame). The authors report that of the 50 completed tests, 30 (60%) did not affect management decisions, 15 (30%) introduced uncertainty regarding the patients' cancer risks, and five (10%) directly influenced management decisions.

A number of other studies have evaluated the impact of panel testing on clinical management of a variety of conditions, including prostate cancer,[9] breast and/or ovarian cancer,[10-13] and non-specific hereditary cancers,[14] as well as genetic profiling of tumor tissue to guide cancer treatment.[15, 16] While some of these studies noted specific changes in medical management resulting from the testing, none of them evaluated whether these changes led to improvements in patient outcomes.

PRACTICE GUIDELINE SUMMARY

AMERICAN SOCIETY OF CLINICAL ONCOLOGY

A 2015 update of a policy statement on genetic and genomic testing for cancer susceptibility from the American Society of Clinical Oncology (ASCO) addresses the application of next-generation sequencing.[17] According to this statement:

> ASCO recognizes that concurrent multigene testing (i.e., panel testing) may be efficient in circumstances that require evaluation of multiple high-penetrance genes of established clinical utility as possible explanations for a patient's personal or family history of cancer. Depending on the specific genes included on the panel employed, panel testing may also identify mutations in genes associated with moderate or low cancer risks and mutations in high-penetrance genes that would not have been evaluated on the basis of the presenting personal or family history. Multigene panel testing will also identify variants of uncertain significance (VUS) in a substantial proportion of patient cases. ASCO affirms that it is sufficient for cancer risk assessment to evaluate genes of established clinical utility that are suggested by the patient's personal and/or family history. Because of the current uncertainties and knowledge gaps, providers with particular expertise in cancer risk assessment should be involved in the ordering and interpretation of multigene panels that include genes of uncertain clinical utility and genes not suggested by the patient's personal and/or family history.
>
> This type of testing may be particularly useful in situations where there are multiple high-penetrance genes associated with a specific cancer, the prevalence of actionable

mutations in one of several genes is high, and it is difficult to predict which gene may be mutated on the basis of phenotype or family history.

So far, there is little consensus as to which genes should be included on panels offered for cancer susceptibility testing- this heterogeneity presents a number of challenges. All panels include high-penetrance genes that are known to cause autosomal-dominant predisposition syndromes, but often include genes that are not necessarily linked to the disease for which the testing is being offered. There is uncertainty regarding the appropriate risk estimates and management strategies for families with unexpected mutations in high-penetrance genes when there is no evidence of the associated syndrome. Clinical utility remains the fundamental issue with respect to testing for mutations in moderate penetrance genes. It is not yet clear whether clinical management should change based on the presence or absence of a mutation. There is insufficient evidence at the present time to conclusively demonstrate the clinical utility of testing for moderate-penetrance mutations, and no guidelines exist to assist oncology providers. Early experience with panel-based testing indicates that a substantial proportion of tests identify a VUS in 1 or more genes, and VUSs are more common in broad-panel testing both because of the number of genes tested and because of the limited understanding of the range of normal variation in some of these genes.

NATIONAL COMPREHENSIVE CANCER NETWORK

The National Comprehensive Cancer Network (NCCN) guidelines on genetic/familial high-risk assessment for breast, ovarian, and pancreatic cancer (v.1.2023)[18] state the following regarding multi-gene testing:

- An individual's personal and/or family history may be explained by more than one inherited cancer syndrome; thus, phenotype-directed testing based on personal and family history through a tailored multi-gene panel test is often more efficient and cost-effective and increases the yield of detecting a P/LP [pathogenic/likely pathogenic] variant in a gene that will impact medical management for the individual or their at-risk family members.
- There may also be a role for multi-gene testing in individuals who have tested negative for a single syndrome, but whose personal or family history remains suggestive of an inherited susceptibility.
- Some individuals may carry P/LP germline variants in more than one cancer susceptibility gene; thus, consideration of a multi-gene panel for individuals already known to carry a single P/LP germline variant from phenotype-directed testing may be considered on a case-by-case basis, based on the degree of suspicion for there being additional variants.
- Multi-gene testing can include "intermediate" penetrant (moderate-risk) genes. For many of these genes, there are limited data on the degree of cancer risk, and there may currently be no clear guidelines on risk management for carriers of P/LP variants. Not all genes included on available multi-gene tests will change risk management compared to that based on other risk factors such as family history.
- It may be possible to refine risks associated with both moderate and high-penetrance genes, taking into account the influence of gene/gene or gene/environment interactions. In addition, certain P/LP variants in a gene may pose higher or lower risk than other P/LP variants in that same gene. This information should be taken into consideration when assigning risks and management recommendations for individuals and their at-risk relatives.

- P/LP variants in many breast, ovarian, pancreatic, and prostate cancer susceptibility genes involved in DNA repair may be associated with rare autosomal recessive conditions, thus posing risks to offspring if the partner is also a carrier.
- As more genes are tested, there is an increased likelihood of finding VUS, mosaicism, and clonal hematopoiesis of indeterminate potential (CHIP).
- There are significant limitations in interpretation of polygenic risk scores (PRSs). PRS should not be used for clinical management at this time and use is recommended in the context of a clinical trial, ideally including more diverse populations.

SUMMARY

Genetic test panels are available for many clinical conditions. Genetic test panels may be focused to a few genes or include a large number of genes. The advantage of genetic test panels is the ability to analyze many genes simultaneously, potentially improving the breadth and efficiency of the genetic workup. A disadvantage of genetic test panels is that the results may provide information on genetic variants that are of unclear clinical significance, or which would not lead to changes in patient management. These results may potentially cause harm by leading to additional unnecessary interventions and anxiety that would not otherwise be considered based on the patient's clinical presentation and/or family history. There is not enough research to show that the genetic panels listed in the policy criteria can lead to better health outcomes for patients. When there is not enough research to show that all genes and/or gene variants in a genetic test panel may be useful for guiding patient management to improve health outcomes, the entire genetic test panel is considered investigational.

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CODES

NOTE: There are few specific codes for molecular pathology testing by panels. If the specific analyte is listed with a CPT code, the specific CPT code should be reported. If the specific analyte is not listed with a specific CPT code, unlisted code 81479 should be reported. The unlisted code would be reported once to represent all of the unlisted analytes in the panel.

Codes	Number	Description
CPT	0008U	Helicobacter pylori detection and antibiotic resistance, DNA, 16S and 23S rRNA, gyrA, pbp1, rdxA and rpoB, next generation sequencing, formalin-fixed paraffin embedded or fresh tissue or fecal sample, predictive, reported as

Codes	Number	Description
		positive or negative for resistance to clarithromycin, fluoroquinolones, metronidazole, amoxicillin, tetracycline and rifabutin
	0010U	Infectious disease (bacterial), strain typing by whole genome sequencing, phylogenetic-based report of strain relatedness, per submitted isolate
	0029U	Drug metabolism (adverse drug reactions and drug response), targeted sequence analysis (ie, CYP1A2, CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, SLCO1B1, VKORC1 and rs12777823)
	0030U	Drug metabolism (warfarin drug response), targeted sequence analysis (ie, CYP2C9, CYP4F2, VKORC1, rs12777823)
	0033U	HTR2A (5-hydroxytryptamine receptor 2A), HTR2C (5-hydroxytryptamine receptor 2C) (eg, citalopram metabolism) gene analysis, common variants (ie, HTR2A rs7997012 [c.614-2211T>C], HTR2C rs3813929 [c.-759C>T] and rs1414334 [c.551-3008C>G])
	0050U	Targeted genomic sequence analysis panel, acute myelogenous leukemia, DNA analysis, 194 genes, interrogation for sequence variants, copy number variants or rearrangements
	0101U	Hereditary colon cancer disorders (eg, Lynch syndrome, PTEN hamartoma syndrome, Cowden syndrome, familial adenomatosis polyposis); genomic sequence analysis panel utilizing a combination of NGS, Sanger, MLPA and array CGH, with MRNA analytics to resolve variants of unknown significance when indicated [15 genes (sequencing and deletion/duplication), EPCAM and GREM1 (deletion/duplication only)]
	0102U	Hereditary breast cancer-related disorders (eg, hereditary breast cancer, hereditary ovarian cancer, hereditary endometrial cancer); genomic sequence analysis panel utilizing a combination of NGS, Sanger, MLPA and array CGH, with MRNA analytics to resolve variants of unknown significance when indicated [17 genes (sequencing and deletion/duplication)]
	0103U	Hereditary ovarian cancer (eg, hereditary ovarian cancer, hereditary endometrial cancer); genomic sequence analysis panel utilizing a combination of NGS, Sanger, MLPA and array CGH, with MRNA analytics to resolve variants of unknown significance when indicated [24 genes (sequencing and deletion/duplication); EPCAM (deletion/duplication only)]
	0129U	Hereditary breast cancer–related disorders (eg, hereditary breast cancer, hereditary ovarian cancer, hereditary endometrial cancer), genomic sequence analysis and deletion/duplication analysis panel (ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, and TP53)
	0130U	Hereditary colon cancer disorders (eg, Lynch syndrome, PTEN hamartoma syndrome, Cowden syndrome, familial adenomatosis polyposis), targeted mRNA sequence analysis panel (APC, CDH1, CHEK2, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, and TP53) (List separately in addition to code for primary procedure) (Use 0130U in conjunction with 81435, 0101U)
	0131U	Hereditary breast cancer–related disorders (eg, hereditary breast cancer, hereditary ovarian cancer, hereditary endometrial cancer), targeted mRNA sequence analysis panel (13 genes) (List separately in addition to code for primary procedure) (Use 0131U in conjunction with 81162, 81432, 0102U)
	0132U	Hereditary ovarian cancer–related disorders (eg, hereditary breast cancer, hereditary ovarian cancer, hereditary endometrial cancer), targeted mRNA sequence analysis panel (17 genes) (List separately in addition to code for primary procedure) (Use 0132U in conjunction with 81162, 81432, 0103U)
	0133U	Hereditary prostate cancer–related disorders, targeted mRNA sequence analysis panel (11 genes) (List separately in addition to code for primary procedure) (Use 0133U in conjunction with 81162)

Codes	Number	Description
	0134U	Hereditary pan cancer (eg, hereditary breast and ovarian cancer, hereditary endometrial cancer, hereditary colorectal cancer), targeted mRNA sequence analysis panel (18 genes) (List separately in addition to code for primary procedure) (Use 0134U in conjunction with 81162, 81432, 81435)
	0135U	Hereditary gynecological cancer (eg, hereditary breast and ovarian cancer, hereditary endometrial cancer, hereditary colorectal cancer), targeted mRNA sequence analysis panel (12 genes) (List separately in addition to code for primary procedure) (Use 0135U in conjunction with 81162)
	0171U	Targeted genomic sequence analysis panel, acute myeloid leukemia, myelodysplastic syndrome, and myeloproliferative neoplasms, DNA analysis, 23 genes, interrogation for sequence variants, rearrangements and minimal residual disease, reported as presence/absence
	0173U	Psychiatry (ie, depression, anxiety), genomic analysis panel, includes variant analysis of 14 genes
	0175U	Psychiatry (eg, depression, anxiety), genomic analysis panel, variant analysis of 15 genes
	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
	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
	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 next generation 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
	0347U	Drug metabolism or processing (multiple conditions), whole blood or buccal specimen, DNA analysis, 16 gene report, with variant analysis and reported phenotypes
	0348U	Drug metabolism or processing (multiple conditions), whole blood or buccal specimen, DNA analysis, 25 gene report, with variant analysis and reported phenotypes

Codes	Number	Description
	0349U	Drug metabolism or processing (multiple conditions), whole blood or buccal specimen, DNA analysis, 27 gene report, with variant analysis, including reported phenotypes and impacted gene-drug interactions
	0350U	Drug metabolism or processing (multiple conditions), whole blood or buccal specimen, DNA analysis, 27 gene report, with variant analysis and reported phenotypes
	0460U	Oncology, whole blood or buccal, DNA single-nucleotide polymorphism (SNP) genotyping by real-time PCR of 24 genes, with variant analysis and reported phenotypes
	0461U	Oncology, pharmacogenomic analysis of single-nucleotide polymorphism (SNP) genotyping by real-time PCR of 24 genes, whole blood or buccal swab, with variant analysis, including impacted gene-drug interactions and reported phenotypes
	0474U	Hereditary pan-cancer (eg, hereditary sarcomas, hereditary endocrine tumors, hereditary neuroendocrine tumors, hereditary cutaneous melanoma), genomic sequence analysis panel of 88 genes with 20 duplications/deletions using next generation sequencing (NGS), Sanger sequencing, blood or saliva, reported as positive or negative for germline variants, each gene
	0475U	Hereditary prostate cancer related disorders, genomic sequence analysis panel using next-generation sequencing (NGS), Sanger sequencing, multiplex ligation-dependent probe amplification (MLPA), and array comparative genomic hybridization (CGH), evaluation of 23 genes and duplications/deletions when indicated, pathologic mutations reported with a genetic risk score for prostate cancer
	0476U	Drug metabolism, psychiatry (eg, major depressive disorder, general anxiety disorder, attention deficit hyperactivity disorder [ADHD], schizophrenia), whole blood, buccal swab, and pharmacogenomic genotyping of 14 genes and CYP2D6 copy number variant analysis and reported phenotypes
	0477U	Drug metabolism, psychiatry (eg, major depressive disorder, general anxiety disorder, attention deficit hyperactivity disorder [ADHD], schizophrenia), whole blood, buccal swab, and pharmacogenomic genotyping of 14 genes and CYP2D6 copy number variant analysis, including impacted gene-drug interactions and reported phenotypes
	0516U	Drug metabolism, whole blood, pharmacogenomic genotyping of 40 genes and CYP2D6 copy number variant analysis, reported as metabolizer status
	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)

Codes	Number	Description
	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)
	81170	ABL1 (ABL proto-oncogene 1, non-receptor tyrosine kinase) (eg, acquired imatinib tyrosine kinase inhibitor resistance), gene analysis, variants in the kinase domain
	81175	ASXL1 (additional sex combs like 1, transcriptional regulator) (eg, myelodysplastic syndrome, myeloproliferative neoplasms, chronic myelomonocytic leukemia), gene analysis; full gene sequence
	81176	;targeted sequence analysis (eg, EXON 12)
	81200	ASPA (aspartoacylase) (eg, Canavan disease) gene analysis, common variants
	81201	APC (adenomatous polyposis coli) (eg, familial adenomatosis polyposis [FAP], attenuated FAP) gene analysis; full gene sequence
	81202	;known familial variants
	81203	;duplication/deletion variants
	81205	BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (eg, Maple syrup urine disease) gene analysis, common variants (eg, R183P, G278S, E422X)
	81206	*BCR/ABL1 (t(9;22))* (eg, chronic myelogenous leukemia) translocation analysis; major breakpoint, qualitative or quantitative
	81207	;minor breakpoint, qualitative or quantitative
	81208	;other breakpoint, qualitative or quantitative
	81209	*BLM (Bloom syndrome, RecQ helicase-like)* (eg, Bloom syndrome) gene analysis, 2281del6ins7 variant
	81210	BRAF (B-Raf proto-oncogene, serine/threonine kinase) (eg, colon cancer, melanoma), gene analysis, V600 variant(s)
	81218	CEBPA (CCAAT/enhancer binding protein [C/EBP], alpha) (eg, acute myeloid leukemia), gene analysis, full gene sequence
	81219	CALR (calreticulin) (eg, myeloproliferative disorders), gene analysis, common variants in exon 9
	81220	*CFTR (cystic fibrosis transmembrane conductance regulator)* (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)
	81221	;known familial variant
	81222	;duplication/deletion variants
	81223	;full gene sequence
	81224	;intron 8 poly-T analysis (eg, male infertility)
	81225	CYP2C19 (cytochrome P450, family 2, subfamily C, polypeptide 19) (eg, drug metabolism), gene analysis, common variants (eg, *2, *3, *4, *8, *17)

Codes	Number	Description
	81226	CYP2D6 (cytochrome P450, family 2, subfamily D, polypeptide 6) (eg, drug metabolism), gene analysis, common variants (eg, *2, *3, *4, *5, *6, *9, *10, *17, *19, *29, *35, *41, *1XN, *2XN, *4XN)
	81227	CYP2C9 (cytochrome P450, family 2, subfamily C, polypeptide 9) (eg, drug metabolism), gene analysis, common variants (eg, *2, *3, *5, *6)
	81228	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number variants, comparative genomic hybridization [CGH] microarray analysis
	81229	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants, comparative genomic hybridization (CGH) microarray analysis
	81235	EGFR (epidermal growth factor receptor) (eg, non-small cell lung cancer) gene analysis, common variants (eg, exon 19 LREA deletion, L858R, T790M, G719A, G719S, L861Q)
	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)
	81245	*FLT3 (fms-related tyrosine kinase 3)* (eg, acute myeloid leukemia), gene analysis; internal tandem duplication (ITD) variants (ie, exons 14, 15)
	81246	;tyrosine kinase domain (TKD) variants (eg, D835, I836)
	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 variant
	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

Codes	Number	Description
		or variant (eg, Southeast Asian, Thai, Filipino, Mediterranean, alpha3.7, alpha4.2, alpha20.5, and Constant Spring)
	81260	IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (eg, familial dysautonomia) gene analysis, common variants (eg, 2507+6T>C, R696P)
	81261	*IGH@ (Immunoglobulin heavy chain locus)* (eg, leukemias and lymphomas, B-cell), gene rearrangement analysis to detect abnormal clonal population(s); amplified methodology (eg, polymerase chain reaction)
	81262	;direct probe methodology (eg, Southern blot)
	81263	*IGH@ (Immunoglobulin heavy chain locus)* (eg, leukemia and lymphoma, B-cell), variable region somatic mutation analysis
	81264	*IGK@ (Immunoglobulin kappa light chain locus)* (eg, leukemia and lymphoma, B-cell), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)
	81270	*JAK2 (Janus kinase 2)* (eg, myeloproliferative disorder) gene analysis, p.Val617Phe (V617F) variant
	81272	*KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (eg, gastrointestinal stromal tumor [GIST], acute myeloid leukemia, melanoma), gene analysis, targeted sequence analysis (eg, exons 8, 11, 13, 17, 18)*
	81273	*KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (eg, mastocytosis), gene analysis, D816 variant(s)*
	81275	KRAS (Kirsten rat sarcoma viral oncogene homolog) (eg, carcinoma) gene analysis; variants in exon 2 (eg, codons 12 and 13)
	81276	KRAS (Kirsten rat sarcoma viral oncogene homolog) (eg, carcinoma) gene analysis; additional variant(s) (eg, codon 61, codon 146)
	81287	*MGMT (O-6-methylguanine-DNA methyltransferase)* (eg, glioblastoma multiforme), promoter methylation analysis
	81288	MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; promoter methylation analysis
	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)
	81292	MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
	81293	;known familial variants
	81294	;duplication/deletion variants
	81295	MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
	81296	;known familial variants
	81297	;duplication/deletion variants
	81298	MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
	81299	;known familial variants
	81300	;duplication/deletion variants
	81302	MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; full sequence analysis
	81303	;known familial variants
	81304	;duplication/deletion variants

Codes	Number	Description
	81310	*NPM1 (nucleophosmin)* (eg, acute myeloid leukemia) gene analysis, exon 12 variants
	81311	*NRAS (neuroblastoma RAS viral [v-ras] oncogene homolog) (eg, colorectal carcinoma), gene analysis, variants in exon 2 (eg, codons 12 and 13) and exon 3 (eg, codon 61)*
	81314	*PDGFRA (platelet-derived growth factor receptor, alpha polypeptide) (eg, gastrointestinal stromal tumor [GIST]), gene analysis, targeted sequence analysis (eg, exons 12, 18)*
	81315	*PML/RARalpha, (t(15;17))*, *(promyelocytic leukemia/retinoic acid receptor alpha)* (eg, promyelocytic leukemia) translocation analysis; common breakpoints (eg, intron 3 and intron 6), qualitative or quantitative
	81316	;single breakpoint (eg, intron 3, intron 6 or exon 6), qualitative or quantitative
	81317	PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
	81318	;known familial variants
	81319	;duplication/deletion variants
	81321	*PTEN (phosphatase and tensin homolog)* (eg, Cowden syndrome, *PTEN* hamartoma tumor syndrome) gene analysis; full sequence analysis
	81322	;known familial variants
	81323	;duplication/deletion variants
	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 variants
	81330	SMPD1(sphingomyelin phosphodiesterase 1, acid lysosomal) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330)
	81331	*SNRPN/UBE3A (small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A)* (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis
	81332	*SERPINA1 (serpin peptidase inhibitor, clade A, alpha-1 antiproteinase, antitrypsin, member 1)* (eg, alpha-1-antitrypsin deficiency), gene analysis, common variants (eg, *S and *Z)
	81340	*TRB@ (T cell antigen receptor, beta)* (eg, leukemia and lymphoma), gene rearrangement analysis to detect abnormal clonal population(s); using amplification methodology (eg, polymerase chain reaction)
	81342	*TRG@ (T cell antigen receptor, gamma)* (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)
	81349	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities
	81350	*UGT1A1 (UDP glucuronosyltransferase 1 family, polypeptide A1)* (eg, drug metabolism, hereditary unconjugated hyperbilirubinemia [Gilbert syndrome]) gene analysis, common variants (eg, *28, *36, *37)
	81355	*VKORC1 (vitamin K epoxide reductase complex, subunit 1)* (eg, warfarin metabolism), gene analysis, common variant(s) (eg, -1639G>A, c.173+1000C>T)
	81400	Molecular pathology procedure, Level 1
	81401	Molecular pathology procedure, Level 2
	81402	Molecular pathology procedure, Level 3

Codes	Number	Description
	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
	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
	81432	Hereditary breast cancer-related disorders (eg, hereditary breast cancer, hereditary ovarian cancer, hereditary endometrial cancer); genomic sequence analysis panel, must include sequencing of at least 10 genes, always including BRCA1, BRCA2, CDH1, MLH1, MSH2, MSH6, PALB2, PTEN, STK11, and TP53
	~~81433~~	~~Hereditary breast cancer-related disorders (eg, hereditary breast cancer, hereditary ovarian cancer, hereditary endometrial cancer); duplication/deletion analysis panel, must include analyses for BRCA1, BRCA2, MLH1, MSH2, and STK11~~ (Deleted 01/01/2025)
	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
	81437	Hereditary neuroendocrine tumor-related disorders (eg, medullary thyroid carcinoma, parathyroid carcinoma, malignant pheochromocytoma or paraganglioma), genomic sequence analysis panel, 5 or more genes, interrogation for sequence variants and copy number variants; genomic sequence analysis panel, must include sequencing of at least 6 genes, including MAX, SDHB, SDHC, SDHD, TMEM127, and VHL
	~~81438~~	~~Hereditary neuroendocrine tumor disorders (eg, medullary thyroid carcinoma, parathyroid carcinoma, malignant pheochromocytoma or paraganglioma); duplication/deletion analysis panel, must include analyses for SDHB, SDHC, SDHD, and VHL~~ (Deleted 01/01/2025)
	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,

Codes	Number	Description
		NHP2, NOP10, PALB2, RAD51C, RPL11, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS7, SBDS, TERT, and TINF2
	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)
	81450	Hematolymphoid neoplasm or disorder, genomic sequence analysis panel, 5-50 genes, interrogation for sequence variants, and copy number variants or rearrangements, or isoform expression or mRNA expression levels, if performed; DNA analysis or combined DNA and RNA analysis
	81451	Hematolymphoid neoplasm or disorder, genomic sequence analysis panel, 5-50 genes, interrogation for sequence variants, and copy number variants or rearrangements, or isoform expression or mRNA expression levels, if performed; RNA analysis
	81455	Solid organ or hematolymphoid neoplasm or disorder, 51 or greater genes, genomic sequence analysis panel, interrogation for sequence variants and copy number variants or rearrangements, or isoform expression or mRNA expression levels, if performed; DNA analysis or combined DNA and RNA analysis
	81456	Solid organ or hematolymphoid neoplasm or disorder, 51 or greater genes, genomic sequence analysis panel, interrogation for sequence variants and copy number variants or rearrangements, or isoform expression or mRNA expression levels, if performed; RNA analysis
	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
	81599	Unlisted multianalyte assay with algorithmic analysis
HCPCS	None	

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