



Evaluating the Utility of Genetic Panels

Effective: January 1, 2025

Next Review: July 2025

Last Review: December 2024

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 |

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| 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 |

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| 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 |

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| 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 |

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| 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 |

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| Differences of Sex Development (DSD) Panel | Prevention Genetics | None |
| Distal Arthrogyriposis Sequencing Panel | University of Chicago Genetics Services Laboratories | None |
| Distal Arthrogyriposis 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 |

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| 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 |

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| 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 |

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| 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 Porphyrins 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 |

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| 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 |

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| 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 |

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| 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 |

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| 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 | |

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| 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 |

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| 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 |

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| 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 cancer 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 iii _a], antigen CD61 [gp _{iii_a}]) (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/iii _a 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 adenomatous 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 | <i>BCR/ABL 1 (t(9;22))</i> (eg, chronic myelogenous leukemia) translocation analysis; major breakpoint, qualitative or quantitative |
| | 81207 | ;minor breakpoint, qualitative or quantitative |
| | 81208 | ;other breakpoint, qualitative or quantitative |
| | 81209 | <i>BLM (Bloom syndrome, RecQ helicase-like)</i> (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 | <i>CFTR (cystic fibrosis transmembrane conductance regulator)</i> (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 | <i>F2 (prothrombin, coagulation factor II)</i> (eg, hereditary hypercoagulability) gene analysis, 20210G>A variant |
| | 81241 | <i>F5 (coagulation factor V)</i> (eg, hereditary hypercoagulability) gene analysis, Leiden variant |
| | 81242 | <i>FANCC (Fanconi anemia, complementation group C)</i> (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A>T) |
| | 81243 | <i>FMR1 (Fragile X messenger ribonucleoprotein 1)</i> (eg, fragile X syndrome, X-linked intellectual disability [XLID]) gene analysis; evaluation to detect abnormal (eg, expanded) alleles |
| | 81244 | <i>FMR1 (fragile X messenger ribonucleoprotein 1)</i> (eg, fragile X syndrome, X-linked intellectual disability [XLID]) gene analysis; characterization of alleles (eg, expanded size and promoter methylation status) |
| | 81245 | <i>FLT3 (fms-related tyrosine kinase 3)</i> (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 | <i>GJB2 (gap junction protein, beta 2, 26kDa, connexin 26)</i> (eg, nonsyndromic hearing loss) gene analysis; full gene sequence |
| | 81253 | ;known familial variant |
| | 81254 | <i>GJB6 (gap junction protein, beta 6, 30kDa, connexin 30)</i> (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 | <i>HFE (hemochromatosis)</i> (eg, hereditary hemochromatosis) gene analysis, common variants (eg, C282Y, H63D) |
| | 81257 | <i>HBA1/HBA2 (alpha globin 1 and alpha globin 2)</i> (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 | <i>IGH@ (Immunoglobulin heavy chain locus)</i> (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 | <i>IGH@ (Immunoglobulin heavy chain locus)</i> (eg, leukemia and lymphoma, B-cell), variable region somatic mutation analysis |
| | 81264 | <i>IGK@ (Immunoglobulin kappa light chain locus)</i> (eg, leukemia and lymphoma, B-cell), gene rearrangement analysis, evaluation to detect abnormal clonal population(s) |
| | 81270 | <i>JAK2 (Janus kinase 2)</i> (eg, myeloproliferative disorder) gene analysis, p.Val617Phe (V617F) variant |
| | 81272 | <i>KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog)</i> (eg, gastrointestinal stromal tumor [GIST], acute myeloid leukemia, melanoma), gene analysis, targeted sequence analysis (eg, exons 8, 11, 13, 17, 18) |
| | 81273 | <i>KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog)</i> (eg, mastocytosis), gene analysis, D816 variant(s) |
| | 81275 | <i>KRAS (Kirsten rat sarcoma viral oncogene homolog)</i> (eg, carcinoma) gene analysis; variants in exon 2 (eg, codons 12 and 13) |
| | 81276 | <i>KRAS (Kirsten rat sarcoma viral oncogene homolog)</i> (eg, carcinoma) gene analysis; additional variant(s) (eg, codon 61, codon 146) |
| | 81287 | <i>MGMT (O-6-methylguanine-DNA methyltransferase)</i> (eg, glioblastoma multiforme), promoter methylation analysis |
| | 81288 | <i>MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2)</i> (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; promoter methylation analysis |
| | 81290 | <i>MCOLN1 (mucolipin 1)</i> (eg, Mucopolipidosis, type IV) gene analysis, common variants (eg, IVS3-2A>G, del6.4kb) |
| | 81291 | <i>MTHFR (5,10-methylenetetrahydrofolate reductase)</i> (eg, hereditary hypercoagulability) gene analysis, common variants (eg, 677T, 1298C) |
| | 81292 | <i>MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2)</i> (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis |
| | 81293 | ;known familial variants |
| | 81294 | ;duplication/deletion variants |
| | 81295 | <i>MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1)</i> (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis |
| | 81296 | ;known familial variants |
| | 81297 | ;duplication/deletion variants |
| | 81298 | <i>MSH6 (mutS homolog 6 [E. coli])</i> (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis |
| | 81299 | ;known familial variants |
| | 81300 | ;duplication/deletion variants |
| | 81302 | <i>MECP2 (methyl CpG binding protein 2)</i> (eg, Rett syndrome) gene analysis; full sequence analysis |
| | 81303 | ;known familial variants |
| | 81304 | ;duplication/deletion variants |

| Codes | Number | Description |
|-------|--------|--|
| | 81310 | <i>NPM1</i> (<i>nucleophosmin</i>) (eg, acute myeloid leukemia) gene analysis, exon 12 variants |
| | 81311 | <i>NRAS</i> (<i>neuroblastoma RAS viral [v-ras] oncogene homolog</i>) (eg, colorectal carcinoma), gene analysis, variants in exon 2 (eg, codons 12 and 13) and exon 3 (eg, codon 61) |
| | 81314 | <i>PDGFRA</i> (<i>platelet-derived growth factor receptor, alpha polypeptide</i>) (eg, gastrointestinal stromal tumor [GIST]), gene analysis, targeted sequence analysis (eg, exons 12, 18) |
| | 81315 | <i>PML/RARalpha</i> , (<i>t(15;17)</i>), (<i>promyelocytic leukemia/retinoic acid receptor alpha</i>) (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 | <i>PMS2</i> (<i>postmeiotic segregation increased 2 [S. cerevisiae]</i>) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis |
| | 81318 | ;known familial variants |
| | 81319 | ;duplication/deletion variants |
| | 81321 | <i>PTEN</i> (<i>phosphatase and tensin homolog</i>) (eg, Cowden syndrome, <i>PTEN</i> hamartoma tumor syndrome) gene analysis; full sequence analysis |
| | 81322 | ;known familial variants |
| | 81323 | ;duplication/deletion variants |
| | 81324 | <i>PMP22</i> (<i>peripheral myelin protein 22</i>) (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 | <i>SMPD1</i> (<i>sphingomyelin phosphodiesterase 1, acid lysosomal</i>) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330) |
| | 81331 | <i>SNRPN/UBE3A</i> (<i>small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A</i>) (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis |
| | 81332 | <i>SERPINA1</i> (<i>serpin peptidase inhibitor, clade A, alpha-1 antiproteinase, antitrypsin, member 1</i>) (eg, alpha-1-antitrypsin deficiency), gene analysis, common variants (eg, *S and *Z) |
| | 81340 | <i>TRB@</i> (<i>T cell antigen receptor, beta</i>) (eg, leukemia and lymphoma), gene rearrangement analysis to detect abnormal clonal population(s); using amplification methodology (eg, polymerase chain reaction) |
| | 81342 | <i>TRG@</i> (<i>T cell antigen receptor, gamma</i>) (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population(s) |
| | 81349 | Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities |
| | 81350 | <i>UGT1A1</i> (<i>UDP glucuronosyltransferase 1 family, polypeptide A1</i>) (eg, drug metabolism, hereditary unconjugated hyperbilirubinemia [Gilbert syndrome]) gene analysis, common variants (eg, *28, *36, *37) |
| | 81355 | <i>VKORC1</i> (<i>vitamin K epoxide reductase complex, subunit 1</i>) (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 |
| | 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, SUGL1, 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, mucopolipidosis 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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