Billing and Coding Article

Article - Billing and Coding: MolDX: Repeat Germline Testing (A57332)

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Noridian Healthcare Solutions, LLC	A and B MAC	02102 - MAC B	J - F	Alaska
Noridian Healthcare Solutions, LLC	A and B MAC	02201 - MAC A	J - F	Idaho
Noridian Healthcare Solutions, LLC	A and B MAC	02202 - MAC B	J - F	Idaho
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Noridian Healthcare Solutions, LLC	A and B MAC	03501 - MAC A	J - F	Utah
Noridian Healthcare Solutions, LLC	A and B MAC	03502 - MAC B	J - F	Utah
Noridian Healthcare Solutions, LLC	A and B MAC	03601 - MAC A	J - F	Wyoming
Noridian Healthcare Solutions, LLC	A and B MAC	03602 - MAC B	J - F	Wyoming

Article Information

General Information

Article ID A57332

Article Title	Fee schedules, relative value units, conversion factors and/or related
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CMS National Coverage Policy

Title XVIII of the Social Security Act, §1833(e). Prohibits Medicare payment for any claim which lacks the necessary information to process the claim.

CMS Internet-Only Manual, Pub. 100-04, Medicare Claims Processing Manual, Chapter 16, §50.5 Jurisdiction of Laboratory Claims, §60.1.1 Independent Laboratory Specimen Drawing, §60.2 Travel Allowance

CMS Internet-Only Manuals, Publication 100-04, Medicare Claims Processing Manual, Chapter 23, §10. Reporting ICD Diagnosis and Procedure Codes.

CMS Internet-Only Manual, Pub. 100-02, Medicare Benefit Policy Manual, Chapter 15, §80.1.2 A/B MAC (B) Contacts With Independent Clinical Laboratories

Article Guidance

Article Text

The information in this article contains billing, coding or other guidelines that complement the Local Coverage Determination (LCD) for MoIDX: Repeat Germline Testing L38353

Coding Information

CPT/HCPCS Codes

Group 1 Paragraph:

For the following $CPT^{(R)}$ codes associated with germline testing, a beneficiary may only be covered for one test per lifetime. This list does not imply coverage for any of the below codes; it identifies codes within scope of the Repeat Germline Testing Policy.

If non-duplicative testing is considered medically reasonable and necessary but utilizes the same billing codes as prior testing, please use a -77 modifier to trigger a medical review for coverage. This includes testing to identify different genetic content or information, such as different variant types or classes, or leveraging newer and more sensitive methodologies. Providers will be expected to provide a rationale for additional testing.

Group 1 Codes: (303 Codes)

CODE	DESCRIPTION
81105	HUMAN PLATELET ANTIGEN 1 GENOTYPING (HPA-1), ITGB3 (INTEGRIN, BETA 3 [PLATELET GLYCOPROTEIN IIIA], ANTIGEN CD61 [GPIIIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT, HPA-1A/B (L33P)
81106	HUMAN PLATELET ANTIGEN 2 GENOTYPING (HPA-2), GP1BA (GLYCOPROTEIN IB [PLATELET], ALPHA POLYPEPTIDE [GPIBA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT, HPA-2A/B (T145M)
81107	HUMAN PLATELET ANTIGEN 3 GENOTYPING (HPA-3), ITGA2B (INTEGRIN, ALPHA 2B [PLATELET GLYCOPROTEIN IIB OF IIB/IIIA COMPLEX], ANTIGEN CD41 [GPIIB]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT, HPA-3A/B (I843S)
81108	HUMAN PLATELET ANTIGEN 4 GENOTYPING (HPA-4), ITGB3 (INTEGRIN, BETA 3 [PLATELET GLYCOPROTEIN IIIA], ANTIGEN CD61 [GPIIIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT, HPA-4A/B (R143Q)
81109	HUMAN PLATELET ANTIGEN 5 GENOTYPING (HPA-5), ITGA2 (INTEGRIN, ALPHA 2 [CD49B, ALPHA 2 SUBUNIT OF VLA-2 RECEPTOR] [GPIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT (EG, HPA-5A/B [K505E])
81110	HUMAN PLATELET ANTIGEN 6 GENOTYPING (HPA-6W), ITGB3 (INTEGRIN, BETA 3 [PLATELET GLYCOPROTEIN IIIA, ANTIGEN CD61] [GPIIIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT, HPA-6A/B (R489Q)
81111	HUMAN PLATELET ANTIGEN 9 GENOTYPING (HPA-9W), ITGA2B (INTEGRIN, ALPHA

CODE	DESCRIPTION
	2B [PLATELET GLYCOPROTEIN IIB OF IIB/IIIA COMPLEX, ANTIGEN CD41] [GPIIB]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT, HPA-9A/B (V837M)
81112	HUMAN PLATELET ANTIGEN 15 GENOTYPING (HPA-15), CD109 (CD109 MOLECULE) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), GENE ANALYSIS, COMMON VARIANT, HPA-15A/B (S682Y)
81161	DMD (DYSTROPHIN) (EG, DUCHENNE/BECKER MUSCULAR DYSTROPHY) DELETION ANALYSIS, AND DUPLICATION ANALYSIS, IF PERFORMED
81162	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED), BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL SEQUENCE ANALYSIS AND FULL DUPLICATION/DELETION ANALYSIS (IE, DETECTION OF LARGE GENE REARRANGEMENTS)
81163	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED), BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81164	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED), BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL DUPLICATION/DELETION ANALYSIS (IE, DETECTION OF LARGE GENE REARRANGEMENTS)
81165	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81166	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL DUPLICATION/DELETION ANALYSIS (IE, DETECTION OF LARGE GENE REARRANGEMENTS)
81167	BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL DUPLICATION/DELETION ANALYSIS (IE, DETECTION OF LARGE GENE REARRANGEMENTS)
81171	AFF2 (ALF TRANSCRIPTION ELONGATION FACTOR 2 [FMR2]) (EG, FRAGILE X INTELLECTUAL DISABILITY 2 [FRAXE]) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81172	AFF2 (ALF TRANSCRIPTION ELONGATION FACTOR 2 [FMR2]) (EG, FRAGILE X INTELLECTUAL DISABILITY 2 [FRAXE]) GENE ANALYSIS; CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE AND METHYLATION STATUS)
81173	AR (ANDROGEN RECEPTOR) (EG, SPINAL AND BULBAR MUSCULAR ATROPHY, KENNEDY DISEASE, X CHROMOSOME INACTIVATION) GENE ANALYSIS; FULL GENE SEQUENCE
81174	AR (ANDROGEN RECEPTOR) (EG, SPINAL AND BULBAR MUSCULAR ATROPHY, KENNEDY DISEASE, X CHROMOSOME INACTIVATION) GENE ANALYSIS; KNOWN FAMILIAL VARIANT

CODE	DESCRIPTION
81177	ATN1 (ATROPHIN 1) (EG, DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81178	ATXN1 (ATAXIN 1) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81179	ATXN2 (ATAXIN 2) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81180	ATXN3 (ATAXIN 3) (EG, SPINOCEREBELLAR ATAXIA, MACHADO-JOSEPH DISEASE) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81181	ATXN7 (ATAXIN 7) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81182	ATXN8OS (ATXN8 OPPOSITE STRAND [NON-PROTEIN CODING]) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81183	ATXN10 (ATAXIN 10) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81184	CACNA1A (CALCIUM VOLTAGE-GATED CHANNEL SUBUNIT ALPHA1 A) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81185	CACNA1A (CALCIUM VOLTAGE-GATED CHANNEL SUBUNIT ALPHA1 A) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS; FULL GENE SEQUENCE
81186	CACNA1A (CALCIUM VOLTAGE-GATED CHANNEL SUBUNIT ALPHA1 A) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS; KNOWN FAMILIAL VARIANT
81187	CNBP (CCHC-TYPE ZINC FINGER NUCLEIC ACID BINDING PROTEIN) (EG, MYOTONIC DYSTROPHY TYPE 2) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81188	CSTB (CYSTATIN B) (EG, UNVERRICHT-LUNDBORG DISEASE) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81189	CSTB (CYSTATIN B) (EG, UNVERRICHT-LUNDBORG DISEASE) GENE ANALYSIS; FULL GENE SEQUENCE
81190	CSTB (CYSTATIN B) (EG, UNVERRICHT-LUNDBORG DISEASE) GENE ANALYSIS; KNOWN FAMILIAL VARIANT(S)
81200	ASPA (ASPARTOACYLASE) (EG, CANAVAN DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, E285A, Y231X)
81201	APC (ADENOMATOUS POLYPOSIS COLI) (EG, FAMILIAL ADENOMATOSIS POLYPOSIS [FAP], ATTENUATED FAP) GENE ANALYSIS; FULL GENE SEQUENCE
81202	APC (ADENOMATOUS POLYPOSIS COLI) (EG, FAMILIAL ADENOMATOSIS POLYPOSIS [FAP], ATTENUATED FAP) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS
81203	APC (ADENOMATOUS POLYPOSIS COLI) (EG, FAMILIAL ADENOMATOSIS POLYPOSIS

CODE	DESCRIPTION
	[FAP], ATTENUATED FAP) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81204	AR (ANDROGEN RECEPTOR) (EG, SPINAL AND BULBAR MUSCULAR ATROPHY, KENNEDY DISEASE, X CHROMOSOME INACTIVATION) GENE ANALYSIS; CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE OR METHYLATION STATUS)
81205	BCKDHB (BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE) (EG, MAPLE SYRUP URINE DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, R183P, G278S, E422X)
81209	BLM (BLOOM SYNDROME, RECQ HELICASE-LIKE) (EG, BLOOM SYNDROME) GENE ANALYSIS, 2281DEL6INS7 VARIANT
81212	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED), BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; 185DELAG, 5385INSC, 6174DELT VARIANTS
81215	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; KNOWN FAMILIAL VARIANT
81216	BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81217	BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; KNOWN FAMILIAL VARIANT
81220	CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; COMMON VARIANTS (EG, ACMG/ACOG GUIDELINES)
81221	CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS
81222	CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81223	CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; FULL GENE SEQUENCE
81224	CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; INTRON 8 POLY-T ANALYSIS (EG, MALE INFERTILITY)
81225	CYP2C19 (CYTOCHROME P450, FAMILY 2, SUBFAMILY C, POLYPEPTIDE 19) (EG, DRUG METABOLISM), GENE ANALYSIS, COMMON VARIANTS (EG, *2, *3, *4, *8, *17)
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)

CODE	DESCRIPTION
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
81230	CYP3A4 (CYTOCHROME P450 FAMILY 3 SUBFAMILY A MEMBER 4) (EG, DRUG METABOLISM), GENE ANALYSIS, COMMON VARIANT(S) (EG, *2, *22)
81231	CYP3A5 (CYTOCHROME P450 FAMILY 3 SUBFAMILY A MEMBER 5) (EG, DRUG METABOLISM), GENE ANALYSIS, COMMON VARIANTS (EG, *2, *3, *4, *5, *6, *7)
81232	DPYD (DIHYDROPYRIMIDINE DEHYDROGENASE) (EG, 5-FLUOROURACIL/5-FU AND CAPECITABINE DRUG METABOLISM), GENE ANALYSIS, COMMON VARIANT(S) (EG, *2A, *4, *5, *6)
81233	BTK (BRUTON'S TYROSINE KINASE) (EG, CHRONIC LYMPHOCYTIC LEUKEMIA) GENE ANALYSIS, COMMON VARIANTS (EG, C481S, C481R, C481F)
81234	DMPK (DM1 PROTEIN KINASE) (EG, MYOTONIC DYSTROPHY TYPE 1) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EXPANDED) ALLELES
81238	F9 (COAGULATION FACTOR IX) (EG, HEMOPHILIA B), FULL GENE SEQUENCE
81239	DMPK (DM1 PROTEIN KINASE) (EG, MYOTONIC DYSTROPHY TYPE 1) GENE ANALYSIS; CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE)
81240	F2 (PROTHROMBIN, COAGULATION FACTOR II) (EG, HEREDITARY HYPERCOAGULABILITY) GENE ANALYSIS, 20210G>A VARIANT
81241	F5 (COAGULATION FACTOR V) (EG, HEREDITARY HYPERCOAGULABILITY) GENE ANALYSIS, LEIDEN VARIANT
81242	FANCC (FANCONI ANEMIA, COMPLEMENTATION GROUP C) (EG, FANCONI ANEMIA, TYPE C) GENE ANALYSIS, COMMON VARIANT (EG, IVS4+4A>T)
81243	FMR1 (FRAGILE X MESSENGER RIBONUCLEOPROTEIN 1) (EG, FRAGILE X SYNDROME, X-LINKED INTELLECTUAL DISABILITY [XLID]) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81244	FMR1 (FRAGILE X MESSENGER RIBONUCLEOPROTEIN 1) (EG, FRAGILE X SYNDROME, X-LINKED INTELLECTUAL DISABILITY [XLID]) GENE ANALYSIS; CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE AND PROMOTER METHYLATION STATUS)
81247	G6PD (GLUCOSE-6-PHOSPHATE DEHYDROGENASE) (EG, HEMOLYTIC ANEMIA, JAUNDICE), GENE ANALYSIS; COMMON VARIANT(S) (EG, A, A-)
81248	G6PD (GLUCOSE-6-PHOSPHATE DEHYDROGENASE) (EG, HEMOLYTIC ANEMIA,

CODE	DESCRIPTION
	JAUNDICE), GENE ANALYSIS; KNOWN FAMILIAL VARIANT(S)
81249	G6PD (GLUCOSE-6-PHOSPHATE DEHYDROGENASE) (EG, HEMOLYTIC ANEMIA, JAUNDICE), GENE ANALYSIS; 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	GJB2 (GAP JUNCTION PROTEIN, BETA 2, 26KDA, CONNEXIN 26) (EG, NONSYNDROMIC HEARING LOSS) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS
81254	GJB6 (GAP JUNCTION PROTEIN, BETA 6, 30KDA, CONNEXIN 30) (EG, NONSYNDROMIC HEARING LOSS) GENE ANALYSIS, COMMON VARIANTS (EG, 309KB [DEL(GJB6-D13S1830)] AND 232KB [DEL(GJB6-D13S1854)])
81255	HEXA (HEXOSAMINIDASE A [ALPHA POLYPEPTIDE]) (EG, TAY-SACHS DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, 1278INSTATC, 1421+1G>C, G269S)
81256	HFE (HEMOCHROMATOSIS) (EG, HEREDITARY HEMOCHROMATOSIS) GENE ANALYSIS, COMMON VARIANTS (EG, C282Y, H63D)
81257	HBA1/HBA2 (ALPHA GLOBIN 1 AND ALPHA GLOBIN 2) (EG, ALPHA THALASSEMIA, HB BART HYDROPS FETALIS SYNDROME, HBH DISEASE), GENE ANALYSIS; COMMON DELETIONS OR VARIANT (EG, SOUTHEAST ASIAN, THAI, FILIPINO, MEDITERRANEAN, ALPHA3.7, ALPHA4.2, ALPHA20.5, CONSTANT SPRING)
81258	HBA1/HBA2 (ALPHA GLOBIN 1 AND ALPHA GLOBIN 2) (EG, ALPHA THALASSEMIA, HB BART HYDROPS FETALIS SYNDROME, HBH DISEASE), GENE ANALYSIS; KNOWN FAMILIAL VARIANT
81259	HBA1/HBA2 (ALPHA GLOBIN 1 AND ALPHA GLOBIN 2) (EG, ALPHA THALASSEMIA, HB BART HYDROPS FETALIS SYNDROME, HBH DISEASE), GENE ANALYSIS; FULL GENE SEQUENCE
81260	IKBKAP (INHIBITOR OF KAPPA LIGHT POLYPEPTIDE GENE ENHANCER IN B-CELLS, KINASE COMPLEX-ASSOCIATED PROTEIN) (EG, FAMILIAL DYSAUTONOMIA) GENE ANALYSIS, COMMON VARIANTS (EG, 2507+6T>C, R696P)
81269	HBA1/HBA2 (ALPHA GLOBIN 1 AND ALPHA GLOBIN 2) (EG, ALPHA THALASSEMIA, HB BART HYDROPS FETALIS SYNDROME, HBH DISEASE), GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81271	HTT (HUNTINGTIN) (EG, HUNTINGTON DISEASE) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81274	HTT (HUNTINGTIN) (EG, HUNTINGTON DISEASE) GENE ANALYSIS;

CODE	DESCRIPTION
	CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE)
81283	IFNL3 (INTERFERON, LAMBDA 3) (EG, DRUG RESPONSE), GENE ANALYSIS, RS12979860 VARIANT
81284	FXN (FRATAXIN) (EG, FRIEDREICH ATAXIA) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EXPANDED) ALLELES
81285	FXN (FRATAXIN) (EG, FRIEDREICH ATAXIA) GENE ANALYSIS; CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE)
81286	FXN (FRATAXIN) (EG, FRIEDREICH ATAXIA) GENE ANALYSIS; FULL GENE SEQUENCE
81289	FXN (FRATAXIN) (EG, FRIEDREICH ATAXIA) GENE ANALYSIS; KNOWN FAMILIAL VARIANT(S)
81290	MCOLN1 (MUCOLIPIN 1) (EG, MUCOLIPIDOSIS, TYPE IV) GENE ANALYSIS, COMMON VARIANTS (EG, IVS3-2A>G, DEL6.4KB)
81291	MTHFR (5,10-METHYLENETETRAHYDROFOLATE REDUCTASE) (EG, HEREDITARY HYPERCOAGULABILITY) GENE ANALYSIS, COMMON VARIANTS (EG, 677T, 1298C)
81292	MLH1 (MUTL HOMOLOG 1, COLON CANCER, NONPOLYPOSIS TYPE 2) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81293	MLH1 (MUTL HOMOLOG 1, COLON CANCER, NONPOLYPOSIS TYPE 2) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS
81294	MLH1 (MUTL HOMOLOG 1, COLON CANCER, NONPOLYPOSIS TYPE 2) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81295	MSH2 (MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81296	MSH2 (MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS
81297	MSH2 (MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81298	MSH6 (MUTS HOMOLOG 6 [E. COLI]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81299	MSH6 (MUTS HOMOLOG 6 [E. COLI]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL

CODE	DESCRIPTION
	VARIANTS
81300	MSH6 (MUTS HOMOLOG 6 [E. COLI]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81302	MECP2 (METHYL CPG BINDING PROTEIN 2) (EG, RETT SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81303	MECP2 (METHYL CPG BINDING PROTEIN 2) (EG, RETT SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANT
CODE	DESCRIPTION
81304	MECP2 (METHYL CPG BINDING PROTEIN 2) (EG, RETT SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81306	NUDT15 (NUDIX HYDROLASE 15) (EG, DRUG METABOLISM) GENE ANALYSIS, COMMON VARIANT(S) (EG, *2, *3, *4, *5, *6)
81307	PALB2 (PARTNER AND LOCALIZER OF BRCA2) (EG, BREAST AND PANCREATIC CANCER) GENE ANALYSIS; FULL GENE SEQUENCE
81309	PIK3CA (PHOSPHATIDYLINOSITOL-4, 5-BIPHOSPHATE 3-KINASE, CATALYTIC SUBUNIT ALPHA) (EG, COLORECTAL AND BREAST CANCER) GENE ANALYSIS, TARGETED SEQUENCE ANALYSIS (EG, EXONS 7, 9, 20)
81312	PABPN1 (POLY[A] BINDING PROTEIN NUCLEAR 1) (EG, OCULOPHARYNGEAL MUSCULAR DYSTROPHY) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81317	PMS2 (POSTMEIOTIC SEGREGATION INCREASED 2 [S. CEREVISIAE]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81318	PMS2 (POSTMEIOTIC SEGREGATION INCREASED 2 [S. CEREVISIAE]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS
81319	PMS2 (POSTMEIOTIC SEGREGATION INCREASED 2 [S. CEREVISIAE]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS
81321	PTEN (PHOSPHATASE AND TENSIN HOMOLOG) (EG, COWDEN SYNDROME, PTEN HAMARTOMA TUMOR SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81322	PTEN (PHOSPHATASE AND TENSIN HOMOLOG) (EG, COWDEN SYNDROME, PTEN HAMARTOMA TUMOR SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANT
81323	PTEN (PHOSPHATASE AND TENSIN HOMOLOG) (EG, COWDEN SYNDROME, PTEN HAMARTOMA TUMOR SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANT
81324	PMP22 (PERIPHERAL MYELIN PROTEIN 22) (EG, CHARCOT-MARIE-TOOTH,

CODE	DESCRIPTION
	HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES) GENE ANALYSIS; DUPLICATION/DELETION ANALYSIS
81325	PMP22 (PERIPHERAL MYELIN PROTEIN 22) (EG, CHARCOT-MARIE-TOOTH, HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES) GENE ANALYSIS; FULL SEQUENCE ANALYSIS
81326	PMP22 (PERIPHERAL MYELIN PROTEIN 22) (EG, CHARCOT-MARIE-TOOTH, HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES) GENE ANALYSIS; KNOWN FAMILIAL VARIANT
81328	SLCO1B1 (SOLUTE CARRIER ORGANIC ANION TRANSPORTER FAMILY, MEMBER 1B1) (EG, ADVERSE DRUG REACTION), GENE ANALYSIS, COMMON VARIANT(S) (EG, *5)
81329	SMN1 (SURVIVAL OF MOTOR NEURON 1, TELOMERIC) (EG, SPINAL MUSCULAR ATROPHY) GENE ANALYSIS; DOSAGE/DELETION ANALYSIS (EG, CARRIER TESTING), INCLUDES SMN2 (SURVIVAL OF MOTOR NEURON 2, CENTROMERIC) ANALYSIS, IF PERFORMED
81330	SMPD1 (SPHINGOMYELIN PHOSPHODIESTERASE 1, ACID LYSOSOMAL) (EG, NIEMANN-PICK DISEASE, TYPE A) GENE ANALYSIS, COMMON VARIANTS (EG, R496L, L302P, FSP330)
81331	SNRPN/UBE3A (SMALL NUCLEAR RIBONUCLEOPROTEIN POLYPEPTIDE N AND UBIQUITIN PROTEIN LIGASE E3A) (EG, PRADER-WILLI SYNDROME AND/OR ANGELMAN SYNDROME), METHYLATION ANALYSIS
81332	SERPINA1 (SERPIN PEPTIDASE INHIBITOR, CLADE A, ALPHA-1 ANTIPROTEINASE, ANTITRYPSIN, MEMBER 1) (EG, ALPHA-1-ANTITRYPSIN DEFICIENCY), GENE ANALYSIS, COMMON VARIANTS (EG, *S AND *Z)
81333	TGFBI (TRANSFORMING GROWTH FACTOR BETA-INDUCED) (EG, CORNEAL DYSTROPHY) GENE ANALYSIS, COMMON VARIANTS (EG, R124H, R124C, R124L, R555W, R555Q)
81335	TPMT (THIOPURINE S-METHYLTRANSFERASE) (EG, DRUG METABOLISM), GENE ANALYSIS, COMMON VARIANTS (EG, *2, *3)
81336	SMN1 (SURVIVAL OF MOTOR NEURON 1, TELOMERIC) (EG, SPINAL MUSCULAR ATROPHY) GENE ANALYSIS; FULL GENE SEQUENCE
81337	SMN1 (SURVIVAL OF MOTOR NEURON 1, TELOMERIC) (EG, SPINAL MUSCULAR ATROPHY) GENE ANALYSIS; KNOWN FAMILIAL SEQUENCE VARIANT(S)
81343	PPP2R2B (PROTEIN PHOSPHATASE 2 REGULATORY SUBUNIT BBETA) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81344	TBP (TATA BOX BINDING PROTEIN) (EG, SPINOCEREBELLAR ATAXIA) GENE ANALYSIS, EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES
81346	TYMS (THYMIDYLATE SYNTHETASE) (EG, 5-FLUOROURACIL/5-FU DRUG

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CODE	DESCRIPTION
	METABOLISM), GENE ANALYSIS, COMMON VARIANT(S) (EG, TANDEM REPEAT VARIANT)
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)
81361	HBB (HEMOGLOBIN, SUBUNIT BETA) (EG, SICKLE CELL ANEMIA, BETA THALASSEMIA, HEMOGLOBINOPATHY); COMMON VARIANT(S) (EG, HBS, HBC, HBE)
81362	HBB (HEMOGLOBIN, SUBUNIT BETA) (EG, SICKLE CELL ANEMIA, BETA THALASSEMIA, HEMOGLOBINOPATHY); KNOWN FAMILIAL VARIANT(S)
81363	HBB (HEMOGLOBIN, SUBUNIT BETA) (EG, SICKLE CELL ANEMIA, BETA THALASSEMIA, HEMOGLOBINOPATHY); DUPLICATION/DELETION VARIANT(S)
81364	HBB (HEMOGLOBIN, SUBUNIT BETA) (EG, SICKLE CELL ANEMIA, BETA THALASSEMIA, HEMOGLOBINOPATHY); FULL GENE SEQUENCE
81410	AORTIC DYSFUNCTION OR DILATION (EG, MARFAN SYNDROME, LOEYS DIETZ SYNDROME, EHLER DANLOS SYNDROME TYPE IV, ARTERIAL TORTUOSITY SYNDROME); GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 9 GENES, INCLUDING FBN1, TGFBR1, TGFBR2, COL3A1, MYH11, ACTA2, SLC2A10, SMAD3, AND MYLK
81411	AORTIC DYSFUNCTION OR DILATION (EG, MARFAN SYNDROME, LOEYS DIETZ SYNDROME, EHLER DANLOS SYNDROME TYPE IV, ARTERIAL TORTUOSITY SYNDROME); DUPLICATION/DELETION ANALYSIS PANEL, MUST INCLUDE ANALYSES FOR TGFBR1, TGFBR2, MYH11, AND COL3A1
81412	ASHKENAZI JEWISH ASSOCIATED DISORDERS (EG, BLOOM SYNDROME, CANAVAN DISEASE, CYSTIC FIBROSIS, FAMILIAL DYSAUTONOMIA, FANCONI ANEMIA GROUP C, GAUCHER DISEASE, TAY-SACHS DISEASE), GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 9 GENES, INCLUDING ASPA, BLM, CFTR, FANCC, GBA, HEXA, IKBKAP, MCOLN1, AND SMPD1
81413	CARDIAC ION CHANNELOPATHIES (EG, BRUGADA SYNDROME, LONG QT SYNDROME, SHORT QT SYNDROME, CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA); GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 10 GENES, INCLUDING ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, AND SCN5A
81414	CARDIAC ION CHANNELOPATHIES (EG, BRUGADA SYNDROME, LONG QT SYNDROME, SHORT QT SYNDROME, CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA); DUPLICATION/DELETION GENE ANALYSIS PANEL, MUST INCLUDE ANALYSIS OF AT LEAST 2 GENES, INCLUDING KCNH2 AND KCNQ1
81415	EXOME (EG, UNEXPLAINED CONSTITUTIONAL OR HERITABLE DISORDER OR

CODE	DESCRIPTION
	SYNDROME); SEQUENCE ANALYSIS
81418	DRUG METABOLISM (EG, PHARMACOGENOMICS) GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE TESTING OF AT LEAST 6 GENES, INCLUDING CYP2C19, CYP2D6, AND CYP2D6 DUPLICATION/DELETION ANALYSIS
81419	EPILEPSY GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE ANALYSES FOR ALDH7A1, CACNA1A, CDKL5, CHD2, GABRG2, GRIN2A, KCNQ2, MECP2, PCDH19, POLG, PRRT2, SCN1A, SCN1B, SCN2A, SCN8A, SLC2A1, SLC9A6, STXBP1, SYNGAP1, TCF4, TPP1, TSC1, TSC2, AND ZEB2
81425	GENOME (EG, UNEXPLAINED CONSTITUTIONAL OR HERITABLE DISORDER OR SYNDROME); SEQUENCE ANALYSIS
81430	HEARING LOSS (EG, NONSYNDROMIC HEARING LOSS, USHER SYNDROME, PENDRED SYNDROME); GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 60 GENES, INCLUDING CDH23, CLRN1, GJB2, GPR98, MTRNR1, MYO7A, MYO15A, PCDH15, OTOF, SLC26A4, TMC1, TMPRSS3, USH1C, USH1G, USH2A, AND WFS1
81431	HEARING LOSS (EG, NONSYNDROMIC HEARING LOSS, USHER SYNDROME, PENDRED SYNDROME); DUPLICATION/DELETION ANALYSIS PANEL, MUST INCLUDE COPY NUMBER ANALYSES FOR STRC AND DFNB1 DELETIONS IN GJB2 AND GJB6 GENES
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
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
81435	HEREDITARY COLON CANCER DISORDERS (EG, LYNCH SYNDROME, PTEN HAMARTOMA SYNDROME, COWDEN SYNDROME, FAMILIAL ADENOMATOSIS POLYPOSIS); GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 10 GENES, INCLUDING APC, BMPR1A, CDH1, MLH1, MSH2, MSH6, MUTYH, PTEN, SMAD4, AND STK11
81436	HEREDITARY COLON CANCER DISORDERS (EG, LYNCH SYNDROME, PTEN HAMARTOMA SYNDROME, COWDEN SYNDROME, FAMILIAL ADENOMATOSIS

CODE	DESCRIPTION
	POLYPOSIS); DUPLICATION/DELETION ANALYSIS PANEL, MUST INCLUDE ANALYSIS OF AT LEAST 5 GENES, INCLUDING MLH1, MSH2, EPCAM, SMAD4, AND STK11
81437	HEREDITARY NEUROENDOCRINE TUMOR DISORDERS (EG, MEDULLARY THYROID CARCINOMA, PARATHYROID CARCINOMA, MALIGNANT PHEOCHROMOCYTOMA OR PARAGANGLIOMA); 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
81439	HEREDITARY CARDIOMYOPATHY (EG, HYPERTROPHIC CARDIOMYOPATHY, DILATED CARDIOMYOPATHY, ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY), GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 5 CARDIOMYOPATHY-RELATED GENES (EG, DSG2, MYBPC3, MYH7, PKP2, TTN)
81440	NUCLEAR ENCODED MITOCHONDRIAL GENES (EG, NEUROLOGIC OR MYOPATHIC PHENOTYPES), GENOMIC SEQUENCE PANEL, MUST INCLUDE ANALYSIS OF AT LEAST 100 GENES, INCLUDING BCS1L, C100RF2, COQ2, COX10, DGUOK, MPV17, OPA1, PDSS2, POLG, POLG2, RRM2B, SCO1, SCO2, SLC25A4, SUCLA2, SUCLG1, TAZ, TK2, AND TYMP
81441	INHERITED BONE MARROW FAILURE SYNDROMES (IBMFS) (EG, FANCONI ANEMIA, DYSKERATOSIS CONGENITA, DIAMOND-BLACKFAN ANEMIA, SHWACHMAN- DIAMOND SYNDROME, GATA2 DEFICIENCY SYNDROME, CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA) SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 30 GENES, INCLUDING BRCA2, BRIP1, DKC1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA1, GATA2, MPL, NHP2, NOP10, PALB2, RAD51C, RPL11, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS7, SBDS, TERT, AND TINF2
81442	NOONAN SPECTRUM DISORDERS (EG, NOONAN SYNDROME, CARDIO-FACIO- CUTANEOUS SYNDROME, COSTELLO SYNDROME, LEOPARD SYNDROME, NOONAN- LIKE SYNDROME), GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 12 GENES, INCLUDING BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, AND SOS1
81443	GENETIC TESTING FOR SEVERE INHERITED CONDITIONS (EG, CYSTIC FIBROSIS, ASHKENAZI JEWISH-ASSOCIATED DISORDERS [EG, BLOOM SYNDROME, CANAVAN DISEASE, FANCONI ANEMIA TYPE C, MUCOLIPIDOSIS TYPE VI, GAUCHER DISEASE, TAY-SACHS DISEASE], BETA HEMOGLOBINOPATHIES, PHENYLKETONURIA, GALACTOSEMIA), GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 15 GENES (EG, ACADM, ARSA, ASPA, ATP7B, BCKDHA, BCKDHB, BLM, CFTR, DHCR7, FANCC, G6PC, GAA, GALT, GBA, GBE1, HBB, HEXA, IKBKAP, MCOLN1, PAH)

CODE	DESCRIPTION
81448	HEREDITARY PERIPHERAL NEUROPATHIES (EG, CHARCOT-MARIE-TOOTH, SPASTIC PARAPLEGIA), GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 5 PERIPHERAL NEUROPATHY-RELATED GENES (EG, BSCL2, GJB1, MFN2, MPZ, REEP1, SPAST, SPG11, SPTLC1)
81460	WHOLE MITOCHONDRIAL GENOME (EG, LEIGH SYNDROME, MITOCHONDRIAL ENCEPHALOMYOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES [MELAS], MYOCLONIC EPILEPSY WITH RAGGED-RED FIBERS [MERFF], NEUROPATHY, ATAXIA, AND RETINITIS PIGMENTOSA [NARP], LEBER HEREDITARY OPTIC NEUROPATHY [LHON]), GENOMIC SEQUENCE, MUST INCLUDE SEQUENCE ANALYSIS OF ENTIRE MITOCHONDRIAL GENOME WITH HETEROPLASMY DETECTION
81465	WHOLE MITOCHONDRIAL GENOME LARGE DELETION ANALYSIS PANEL (EG, KEARNS-SAYRE SYNDROME, CHRONIC PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA), INCLUDING HETEROPLASMY DETECTION, IF PERFORMED
81470	X-LINKED INTELLECTUAL DISABILITY (XLID) (EG, SYNDROMIC AND NON- SYNDROMIC XLID); GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 60 GENES, INCLUDING ARX, ATRX, CDKL5, FGD1, FMR1, HUWE1, IL1RAPL, KDM5C, L1CAM, MECP2, MED12, MID1, OCRL, RPS6KA3, AND SLC16A2
81471	X-LINKED INTELLECTUAL DISABILITY (XLID) (EG, SYNDROMIC AND NON- SYNDROMIC XLID); 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
81493	CORONARY ARTERY DISEASE, MRNA, GENE EXPRESSION PROFILING BY REAL-TIME RT-PCR OF 23 GENES, UTILIZING WHOLE PERIPHERAL BLOOD, ALGORITHM REPORTED AS A RISK SCORE
81554	PULMONARY DISEASE (IDIOPATHIC PULMONARY FIBROSIS [IPF]), MRNA, GENE EXPRESSION ANALYSIS OF 190 GENES, UTILIZING TRANSBRONCHIAL BIOPSIES, DIAGNOSTIC ALGORITHM REPORTED AS CATEGORICAL RESULT (EG, POSITIVE OR NEGATIVE FOR HIGH PROBABILITY OF USUAL INTERSTITIAL PNEUMONIA [UIP])
0004M	SCOLIOSIS, DNA ANALYSIS OF 53 SINGLE NUCLEOTIDE POLYMORPHISMS (SNPS), USING SALIVA, PROGNOSTIC ALGORITHM REPORTED AS A RISK SCORE
0001U	RED BLOOD CELL ANTIGEN TYPING, DNA, HUMAN ERYTHROCYTE ANTIGEN GENE ANALYSIS OF 35 ANTIGENS FROM 11 BLOOD GROUPS, UTILIZING WHOLE BLOOD, COMMON RBC ALLELES REPORTED
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)

CODE	DESCRIPTION
0031U	CYP1A2 (CYTOCHROME P450 FAMILY 1, SUBFAMILY A, MEMBER 2) (EG, DRUG METABOLISM) GENE ANALYSIS, COMMON VARIANTS (IE, *1F, *1K, *6, *7)
0032U	COMT (CATECHOL-O-METHYLTRANSFERASE) (DRUG METABOLISM) GENE ANALYSIS, C.472G>A (RS4680) VARIANT
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])
0034U	TPMT (THIOPURINE S-METHYLTRANSFERASE), NUDT15 (NUDIX HYDROXYLASE 15)(EG, THIOPURINE METABOLISM), GENE ANALYSIS, COMMON VARIANTS (IE, TPMT *2, *3A, *3B, *3C, *4, *5, *6, *8, *12; NUDT15 *3, *4, *5)
0070U	CYP2D6 (CYTOCHROME P450, FAMILY 2, SUBFAMILY D, POLYPEPTIDE 6) (EG, DRUG METABOLISM) GENE ANALYSIS, COMMON AND SELECT RARE VARIANTS (IE, *2, *3, *4, *4N, *5, *6, *7, *8, *9, *10, *11, *12, *13, *14A, *14B, *15, *17, *29, *35, *36, *41, *57, *61, *63, *68, *83, *XN)
0071U	CYP2D6 (CYTOCHROME P450, FAMILY 2, SUBFAMILY D, POLYPEPTIDE 6) (EG, DRUG METABOLISM) GENE ANALYSIS, FULL GENE SEQUENCE (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0072U	CYP2D6 (CYTOCHROME P450, FAMILY 2, SUBFAMILY D, POLYPEPTIDE 6) (EG, DRUG METABOLISM) GENE ANALYSIS, TARGETED SEQUENCE ANALYSIS (IE, CYP2D6-2D7 HYBRID GENE) (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0073U	CYP2D6 (CYTOCHROME P450, FAMILY 2, SUBFAMILY D, POLYPEPTIDE 6) (EG, DRUG METABOLISM) GENE ANALYSIS, TARGETED SEQUENCE ANALYSIS (IE, CYP2D7-2D6 HYBRID GENE) (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0074U	CYP2D6 (CYTOCHROME P450, FAMILY 2, SUBFAMILY D, POLYPEPTIDE 6) (EG, DRUG METABOLISM) GENE ANALYSIS, TARGETED SEQUENCE ANALYSIS (IE, NON- DUPLICATED GENE WHEN DUPLICATION/MULTIPLICATION IS TRANS) (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0075U	CYP2D6 (CYTOCHROME P450, FAMILY 2, SUBFAMILY D, POLYPEPTIDE 6) (EG, DRUG METABOLISM) GENE ANALYSIS, TARGETED SEQUENCE ANALYSIS (IE, 5' GENE DUPLICATION/MULTIPLICATION) (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0076U	CYP2D6 (CYTOCHROME P450, FAMILY 2, SUBFAMILY D, POLYPEPTIDE 6) (EG, DRUG METABOLISM) GENE ANALYSIS, TARGETED SEQUENCE ANALYSIS (IE, 3' GENE DUPLICATION/ MULTIPLICATION) (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0079U	COMPARATIVE DNA ANALYSIS USING MULTIPLE SELECTED SINGLE-NUCLEOTIDE POLYMORPHISMS (SNPS), URINE AND BUCCAL DNA, FOR SPECIMEN IDENTITY

CODE	DESCRIPTION
	VERIFICATION
0084U	RED BLOOD CELL ANTIGEN TYPING, DNA, GENOTYPING OF 10 BLOOD GROUPS WITH PHENOTYPE PREDICTION OF 37 RED BLOOD CELL ANTIGENS
0094U	GENOME (EG, UNEXPLAINED CONSTITUTIONAL OR HERITABLE DISORDER OR SYNDROME), RAPID SEQUENCE ANALYSIS
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 MMRNA 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 MMRNA 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 MMRNA 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)
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)
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)

CODE	DESCRIPTION
0133U	HEREDITARY PROSTATE CANCER-RELATED DISORDERS, TARGETED MRNA SEQUENCE ANALYSIS PANEL (11 GENES) (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
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)
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)
0136U	ATM (ATAXIA TELANGIECTASIA MUTATED) (EG, ATAXIA TELANGIECTASIA) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0137U	PALB2 (PARTNER AND LOCALIZER OF BRCA2) (EG, BREAST AND PANCREATIC CANCER) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0138U	BRCA1 (BRCA1, DNA REPAIR ASSOCIATED), BRCA2 (BRCA2, DNA REPAIR ASSOCIATED) (EG, HEREDITARY BREAST AND OVARIAN CANCER) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0156U	COPY NUMBER (EG, INTELLECTUAL DISABILITY, DYSMORPHOLOGY), SEQUENCE ANALYSIS
0157U	APC (APC REGULATOR OF WNT SIGNALING PATHWAY) (EG, FAMILIAL ADENOMATOSIS POLYPOSIS [FAP]) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0158U	MLH1 (MUTL HOMOLOG 1) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0159U	MSH2 (MUTS HOMOLOG 2) (EG, HEREDITARY COLON CANCER, LYNCH SYNDROME) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0160U	MSH6 (MUTS HOMOLOG 6) (EG, HEREDITARY COLON CANCER, LYNCH SYNDROME) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0161U	PMS2 (PMS1 HOMOLOG 2, MISMATCH REPAIR SYSTEM COMPONENT) (EG, HEREDITARY NONPOLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) MRNA SEQUENCE ANALYSIS (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)
0162U	HEREDITARY COLON CANCER (LYNCH SYNDROME), TARGETED MRNA SEQUENCE ANALYSIS PANEL (MLH1, MSH2, MSH6, PMS2) (LIST SEPARATELY IN ADDITION TO

CODE	DESCRIPTION
	CODE FOR PRIMARY PROCEDURE)
CODE	DESCRIPTION
0169U	NUDT15 (NUDIX HYDROLASE 15) AND TPMT (THIOPURINE S- METHYLTRANSFERASE) (EG, DRUG METABOLISM) GENE ANALYSIS, COMMON VARIANTS
0170U	NEUROLOGY (AUTISM SPECTRUM DISORDER [ASD]), RNA, NEXT-GENERATION SEQUENCING, SALIVA, ALGORITHMIC ANALYSIS, AND RESULTS REPORTED AS PREDICTIVE PROBABILITY OF ASD DIAGNOSIS
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
0180U	RED CELL ANTIGEN (ABO BLOOD GROUP) GENOTYPING (ABO), GENE ANALYSIS SANGER/CHAIN TERMINATION/CONVENTIONAL SEQUENCING, ABO (ABO, ALPHA 1- 3-N-ACETYLGALACTOSAMINYLTRANSFERASE AND ALPHA 1-3- GALACTOSYLTRANSFERASE) GENE, INCLUDING SUBTYPING, 7 EXONS
0181U	RED CELL ANTIGEN (COLTON BLOOD GROUP) GENOTYPING (CO), GENE ANALYSIS, AQP1 (AQUAPORIN 1 [COLTON BLOOD GROUP]) EXON 1
0182U	RED CELL ANTIGEN (CROMER BLOOD GROUP) GENOTYPING (CROM), GENE ANALYSIS, CD55 (CD55 MOLECULE [CROMER BLOOD GROUP]) EXONS 1-10
0183U	RED CELL ANTIGEN (DIEGO BLOOD GROUP) GENOTYPING (DI), GENE ANALYSIS, SLC4A1 (SOLUTE CARRIER FAMILY 4 MEMBER 1 [DIEGO BLOOD GROUP]) EXON 19
0184U	RED CELL ANTIGEN (DOMBROCK BLOOD GROUP) GENOTYPING (DO), GENE ANALYSIS, ART4 (ADP-RIBOSYLTRANSFERASE 4 [DOMBROCK BLOOD GROUP]) EXON 2
0185U	RED CELL ANTIGEN (H BLOOD GROUP) GENOTYPING (FUT1), GENE ANALYSIS, FUT1 (FUCOSYLTRANSFERASE 1 [H BLOOD GROUP]) EXON 4
0186U	RED CELL ANTIGEN (H BLOOD GROUP) GENOTYPING (FUT2), GENE ANALYSIS, FUT2 (FUCOSYLTRANSFERASE 2) EXON 2
0187U	RED CELL ANTIGEN (DUFFY BLOOD GROUP) GENOTYPING (FY), GENE ANALYSIS, ACKR1 (ATYPICAL CHEMOKINE RECEPTOR 1 [DUFFY BLOOD GROUP]) EXONS 1-2
0188U	RED CELL ANTIGEN (GERBICH BLOOD GROUP) GENOTYPING (GE), GENE ANALYSIS, GYPC (GLYCOPHORIN C [GERBICH BLOOD GROUP]) EXONS 1-4
0189U	RED CELL ANTIGEN (MNS BLOOD GROUP) GENOTYPING (GYPA), GENE ANALYSIS, GYPA (GLYCOPHORIN A [MNS BLOOD GROUP]) INTRONS 1, 5, EXON 2
0190U	RED CELL ANTIGEN (MNS BLOOD GROUP) GENOTYPING (GYPB), GENE ANALYSIS, GYPB (GLYCOPHORIN B [MNS BLOOD GROUP]) INTRONS 1, 5, PSEUDOEXON 3

CODE	DESCRIPTION
0191U	RED CELL ANTIGEN (INDIAN BLOOD GROUP) GENOTYPING (IN), GENE ANALYSIS, CD44 (CD44 MOLECULE [INDIAN BLOOD GROUP]) EXONS 2, 3, 6
0192U	RED CELL ANTIGEN (KIDD BLOOD GROUP) GENOTYPING (JK), GENE ANALYSIS, SLC14A1 (SOLUTE CARRIER FAMILY 14 MEMBER 1 [KIDD BLOOD GROUP]) GENE PROMOTER, EXON 9
0193U	RED CELL ANTIGEN (JR BLOOD GROUP) GENOTYPING (JR), GENE ANALYSIS, ABCG2 (ATP BINDING CASSETTE SUBFAMILY G MEMBER 2 [JUNIOR BLOOD GROUP]) EXONS 2-26
0194U	RED CELL ANTIGEN (KELL BLOOD GROUP) GENOTYPING (KEL), GENE ANALYSIS, KEL (KELL METALLO-ENDOPEPTIDASE [KELL BLOOD GROUP]) EXON 8
0195U	KLF1 (KRUPPEL-LIKE FACTOR 1), TARGETED SEQUENCING (IE, EXON 13)
0196U	RED CELL ANTIGEN (LUTHERAN BLOOD GROUP) GENOTYPING (LU), GENE ANALYSIS, BCAM (BASAL CELL ADHESION MOLECULE [LUTHERAN BLOOD GROUP]) EXON 3
0197U	RED CELL ANTIGEN (LANDSTEINER-WIENER BLOOD GROUP) GENOTYPING (LW), GENE ANALYSIS, ICAM4 (INTERCELLULAR ADHESION MOLECULE 4 [LANDSTEINER- WIENER BLOOD GROUP]) EXON 1
0198U	RED CELL ANTIGEN (RH BLOOD GROUP) GENOTYPING (RHD AND RHCE), GENE ANALYSIS SANGER/CHAIN TERMINATION/CONVENTIONAL SEQUENCING, RHD (RH BLOOD GROUP D ANTIGEN) EXONS 1-10 AND RHCE (RH BLOOD GROUP CCEE ANTIGENS) EXON 5
0199U	RED CELL ANTIGEN (SCIANNA BLOOD GROUP) GENOTYPING (SC), GENE ANALYSIS, ERMAP (ERYTHROBLAST MEMBRANE ASSOCIATED PROTEIN [SCIANNA BLOOD GROUP]) EXONS 4, 12
0200U	RED CELL ANTIGEN (KX BLOOD GROUP) GENOTYPING (XK), GENE ANALYSIS, XK (X-LINKED KX BLOOD GROUP) EXONS 1-3
0201U	RED CELL ANTIGEN (YT BLOOD GROUP) GENOTYPING (YT), GENE ANALYSIS, ACHE (ACETYLCHOLINESTERASE [CARTWRIGHT BLOOD GROUP]) EXON 2
0203U	AUTOIMMUNE (INFLAMMATORY BOWEL DISEASE), MRNA, GENE EXPRESSION PROFILING BY QUANTITATIVE RT-PCR, 17 GENES (15 TARGET AND 2 REFERENCE GENES), WHOLE BLOOD, REPORTED AS A CONTINUOUS RISK SCORE AND CLASSIFICATION OF INFLAMMATORY BOWEL DISEASE AGGRESSIVENESS
0205U	OPHTHALMOLOGY (AGE-RELATED MACULAR DEGENERATION), ANALYSIS OF 3 GENE VARIANTS (2 CFH GENE, 1 ARMS2 GENE), USING PCR AND MALDI-TOF, BUCCAL SWAB, REPORTED AS POSITIVE OR NEGATIVE FOR NEOVASCULAR AGE- RELATED MACULAR-DEGENERATION RISK ASSOCIATED WITH ZINC SUPPLEMENTS
0209U	CYTOGENOMIC CONSTITUTIONAL (GENOME-WIDE) ANALYSIS, INTERROGATION OF GENOMIC REGIONS FOR COPY NUMBER, STRUCTURAL CHANGES AND AREAS OF HOMOZYGOSITY FOR CHROMOSOMAL ABNORMALITIES

CODE	DESCRIPTION
0212U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), WHOLE GENOME AND MITOCHONDRIAL DNA SEQUENCE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT GENE EXPANSIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS, BLOOD OR SALIVA, IDENTIFICATION AND CATEGORIZATION OF GENETIC VARIANTS, PROBAND
0213U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), WHOLE GENOME AND MITOCHONDRIAL DNA SEQUENCE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT GENE EXPANSIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS, BLOOD OR SALIVA, IDENTIFICATION AND CATEGORIZATION OF GENETIC VARIANTS, EACH COMPARATOR GENOME (EG, PARENT, SIBLING)
0214U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), WHOLE EXOME AND MITOCHONDRIAL DNA SEQUENCE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT GENE EXPANSIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS, BLOOD OR SALIVA, IDENTIFICATION AND CATEGORIZATION OF GENETIC VARIANTS, PROBAND
0215U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), WHOLE EXOME AND MITOCHONDRIAL DNA SEQUENCE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT GENE EXPANSIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS, BLOOD OR SALIVA, IDENTIFICATION AND CATEGORIZATION OF GENETIC VARIANTS, EACH COMPARATOR EXOME (EG, PARENT, SIBLING)
0216U	NEUROLOGY (INHERITED ATAXIAS), GENOMIC DNA SEQUENCE ANALYSIS OF 12 COMMON GENES INCLUDING SMALL SEQUENCE CHANGES, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT GENE EXPANSIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS, BLOOD OR SALIVA, IDENTIFICATION AND CATEGORIZATION OF GENETIC VARIANTS
0217U	NEUROLOGY (INHERITED ATAXIAS), GENOMIC DNA SEQUENCE ANALYSIS OF 51 GENES INCLUDING SMALL SEQUENCE CHANGES, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT GENE EXPANSIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS, BLOOD OR SALIVA, IDENTIFICATION AND CATEGORIZATION OF GENETIC VARIANTS
0218U	NEUROLOGY (MUSCULAR DYSTROPHY), DMD GENE SEQUENCE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES, DELETIONS, DUPLICATIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS, BLOOD OR SALIVA, IDENTIFICATION AND CHARACTERIZATION OF GENETIC VARIANTS
0221U	RED CELL ANTIGEN (ABO BLOOD GROUP) GENOTYPING (ABO), GENE ANALYSIS, NEXT-GENERATION SEQUENCING, ABO (ABO, ALPHA 1-3-N- ACETYLGALACTOSAMINYLTRANSFERASE AND ALPHA 1-3- GALACTOSYLTRANSFERASE) GENE

CODE	DESCRIPTION
0222U	RED CELL ANTIGEN (RH BLOOD GROUP) GENOTYPING (RHD AND RHCE), GENE ANALYSIS, NEXT-GENERATION SEQUENCING, RH PROXIMAL PROMOTER, EXONS 1- 10, PORTIONS OF INTRONS 2-3
0230U	AR (ANDROGEN RECEPTOR) (EG, SPINAL AND BULBAR MUSCULAR ATROPHY, KENNEDY DISEASE, X CHROMOSOME INACTIVATION), FULL SEQUENCE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT (STR) EXPANSIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS
0231U	CACNA1A (CALCIUM VOLTAGE-GATED CHANNEL SUBUNIT ALPHA 1A) (EG, SPINOCEREBELLAR ATAXIA), FULL GENE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT (STR) GENE EXPANSIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS
0232U	CSTB (CYSTATIN B) (EG, PROGRESSIVE MYOCLONIC EPILEPSY TYPE 1A, UNVERRICHT-LUNDBORG DISEASE), FULL GENE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT (STR) EXPANSIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS
0233U	FXN (FRATAXIN) (EG, FRIEDREICH ATAXIA), GENE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, SHORT TANDEM REPEAT (STR) EXPANSIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS
0234U	MECP2 (METHYL CPG BINDING PROTEIN 2) (EG, RETT SYNDROME), FULL GENE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS
0235U	PTEN (PHOSPHATASE AND TENSIN HOMOLOG) (EG, COWDEN SYNDROME, PTEN HAMARTOMA TUMOR SYNDROME), FULL GENE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS
0236U	SMN1 (SURVIVAL OF MOTOR NEURON 1, TELOMERIC) AND SMN2 (SURVIVAL OF MOTOR NEURON 2, CENTROMERIC) (EG, SPINAL MUSCULAR ATROPHY) FULL GENE ANALYSIS, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DUPLICATIONS AND DELETIONS, AND MOBILE ELEMENT INSERTIONS
0237U	CARDIAC ION CHANNELOPATHIES (EG, BRUGADA SYNDROME, LONG QT SYNDROME, SHORT QT SYNDROME, CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA), GENOMIC SEQUENCE ANALYSIS PANEL INCLUDING ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, AND SCN5A, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN

CODE	DESCRIPTION
	NON-UNIQUELY MAPPABLE REGIONS
0238U	ONCOLOGY (LYNCH SYNDROME), GENOMIC DNA SEQUENCE ANALYSIS OF MLH1, MSH2, MSH6, PMS2, AND EPCAM, INCLUDING SMALL SEQUENCE CHANGES IN EXONIC AND INTRONIC REGIONS, DELETIONS, DUPLICATIONS, MOBILE ELEMENT INSERTIONS, AND VARIANTS IN NON-UNIQUELY MAPPABLE REGIONS
0246U	RED BLOOD CELL ANTIGEN TYPING, DNA, GENOTYPING OF AT LEAST 16 BLOOD GROUPS WITH PHENOTYPE PREDICTION OF AT LEAST 51 RED BLOOD CELL ANTIGENS
0258U	AUTOIMMUNE (PSORIASIS), MRNA, NEXT-GENERATION SEQUENCING, GENE EXPRESSION PROFILING OF 50-100 GENES, SKIN-SURFACE COLLECTION USING ADHESIVE PATCH, ALGORITHM REPORTED AS LIKELIHOOD OF RESPONSE TO PSORIASIS BIOLOGICS
0260U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), IDENTIFICATION OF COPY NUMBER VARIATIONS, INVERSIONS, INSERTIONS, TRANSLOCATIONS, AND OTHER STRUCTURAL VARIANTS BY OPTICAL GENOME MAPPING
0264U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), IDENTIFICATION OF COPY NUMBER VARIATIONS, INVERSIONS, INSERTIONS, TRANSLOCATIONS, AND OTHER STRUCTURAL VARIANTS BY OPTICAL GENOME MAPPING
0265U	RARE CONSTITUTIONAL AND OTHER HERITABLE DISORDERS, WHOLE GENOME AND MITOCHONDRIAL DNA SEQUENCE ANALYSIS, BLOOD, FROZEN AND FORMALIN-FIXED PARAFFINEMBEDDED (FFPE) TISSUE, SALIVA, BUCCAL SWABS OR CELL LINES, IDENTIFICATION OF SINGLE NUCLEOTIDE AND COPY NUMBER VARIANTS
0266U	UNEXPLAINED CONSTITUTIONAL OR OTHER HERITABLE DISORDERS OR SYNDROMES, TISSUESPECIFIC GENE EXPRESSION BY WHOLETRANSCRIPTOME AND NEXT-GENERATION SEQUENCING, BLOOD, FORMALIN-FIXED PARAFFINEMBEDDED (FFPE) TISSUE OR FRESH FROZEN TISSUE, REPORTED AS PRESENCE OR ABSENCE OF SPLICING OR EXPRESSION CHANGES
0267U	RARE CONSTITUTIONAL AND OTHER HERITABLE DISORDERS, IDENTIFICATION OF COPY NUMBER VARIATIONS, INVERSIONS, INSERTIONS, TRANSLOCATIONS, AND OTHER STRUCTURAL VARIANTS BY OPTICAL GENOME MAPPING AND WHOLE GENOME SEQUENCING
0268U	HEMATOLOGY (ATYPICAL HEMOLYTIC UREMIC SYNDROME [AHUS]), GENOMIC SEQUENCE ANALYSIS OF 15 GENES, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0269U	HEMATOLOGY (AUTOSOMAL DOMINANT CONGENITAL THROMBOCYTOPENIA), GENOMIC SEQUENCE ANALYSIS OF 22 GENES, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0270U	HEMATOLOGY (CONGENITAL COAGULATION DISORDERS), GENOMIC SEQUENCE ANALYSIS OF 20 GENES, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0271U	HEMATOLOGY (CONGENITAL NEUTROPENIA), GENOMIC SEQUENCE ANALYSIS OF

CODE	DESCRIPTION
	24 GENES, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0272U	HEMATOLOGY (GENETIC BLEEDING DISORDERS), GENOMIC SEQUENCE ANALYSIS OF 60 GENES AND DUPLICATION/DELETION OF PLAU, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID, COMPREHENSIVE
0273U	HEMATOLOGY (GENETIC HYPERFIBRINOLYSIS, DELAYED BLEEDING), GENOMIC SEQUENCE ANALYSIS OF 8 GENES (F13A1, F13B, FGA, FGB, FGG, SERPINA1, SERPINE1, SERPINF2, PLAU), BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0274U	HEMATOLOGY (GENETIC PLATELET DISORDERS), GENOMIC SEQUENCE ANALYSIS OF 62 GENES AND DUPLICATION/DELETION OF PLAU, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0276U	HEMATOLOGY (INHERITED THROMBOCYTOPENIA), GENOMIC SEQUENCE ANALYSIS OF 42 GENES, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0277U	HEMATOLOGY (GENETIC PLATELET FUNCTION DISORDER), GENOMIC SEQUENCE ANALYSIS OF 40 GENES AND DUPLICATION/DELETION OF PLAU, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0278U	HEMATOLOGY (GENETIC THROMBOSIS), GENOMIC SEQUENCE ANALYSIS OF 14 GENES, BLOOD, BUCCAL SWAB, OR AMNIOTIC FLUID
0282U	RED BLOOD CELL ANTIGEN TYPING, DNA, GENOTYPING OF 12 BLOOD GROUP SYSTEM GENES TO PREDICT 44 RED BLOOD CELL ANTIGEN PHENOTYPES
0286U	CEP72 (CENTROSOMAL PROTEIN, 72-KDA), NUDT15 (NUDIX HYDROLASE 15) AND TPMT (THIOPURINE S-METHYLTRANSFERASE) (EG, DRUG METABOLISM) GENE ANALYSIS, COMMON VARIANTS
0289U	NEUROLOGY (ALZHEIMER DISEASE), MRNA, GENE EXPRESSION PROFILING BY RNA SEQUENCING OF 24 GENES, WHOLE BLOOD, ALGORITHM REPORTED AS PREDICTIVE RISK SCORE
0290U	PAIN MANAGEMENT, MRNA, GENE EXPRESSION PROFILING BY RNA SEQUENCING OF 36 GENES, WHOLE BLOOD, ALGORITHM REPORTED AS PREDICTIVE RISK SCORE
0291U	PSYCHIATRY (MOOD DISORDERS), MRNA, GENE EXPRESSION PROFILING BY RNA SEQUENCING 144 GENES, WHOLE BLOOD, ALGORITHM REPORTED AS PREDICTIVE RISK SCORE
0292U	PSYCHIATRY (STRESS DISORDERS), MRNA, GENE EXPRESSION PROFILING BY RNA SEQUENCING OF 72 GENES, WHOLE BLOOD, ALGORITHM REPORTED AS PREDICTIVE RISK SCORE
0293U	PSYCHIATRY (SUICIDAL IDEATION), MRNA, GENE EXPRESSION PROFILING BY RNA SEQUENCING OF 54 GENES, WHOLE BLOOD, ALGORITHM REPORTED AS PREDICTIVE RISK SCORE
0294U	LONGEVITY AND MORTALITY RISK, MRNA, GENE EXPRESSION PROFILING BY RNA SEQUENCING OF 18 GENES, WHOLE BLOOD, ALGORITHM REPORTED AS PREDICTIVE RISK SCORE

CODE	DESCRIPTION
0318U	PEDIATRICS (CONGENITAL EPIGENETIC DISORDERS), WHOLE GENOME METHYLATION ANALYSIS BY MICROARRAY FOR 50 OR MORE GENES, BLOOD
0345U	PSYCHIATRY (EG, DEPRESSION, ANXIETY, ATTENTION DEFICIT HYPERACTIVITY DISORDER [ADHD]), GENOMIC ANALYSIS PANEL, VARIANT ANALYSIS OF 15 GENES, INCLUDING DELETION/DUPLICATION ANALYSIS OF CYP2D6
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
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
0355U	APOL1 (APOLIPOPROTEIN L1) (EG, CHRONIC KIDNEY DISEASE), RISK VARIANTS (G1, G2)
0378U	RFC1 (REPLICATION FACTOR C SUBUNIT 1), REPEAT EXPANSION VARIANT ANALYSIS BY TRADITIONAL AND REPEAT-PRIMED PCR, BLOOD, SALIVA, OR BUCCAL SWAB
0380U	DRUG METABOLISM (ADVERSE DRUG REACTIONS AND DRUG RESPONSE), TARGETED SEQUENCE ANALYSIS, 20 GENE VARIANTS AND CYP2D6 DELETION OR DUPLICATION ANALYSIS WITH REPORTED GENOTYPE AND PHENOTYPE
0389U	PEDIATRIC FEBRILE ILLNESS (KAWASAKI DISEASE [KD]), INTERFERON ALPHA- INDUCIBLE PROTEIN 27 (IFI27) AND MAST CELL-EXPRESSED MEMBRANE PROTEIN 1 (MCEMP1), RNA, USING REVERSE TRANSCRIPTION POLYMERASE CHAIN REACTION (RT-QPCR), BLOOD, REPORTED AS A RISK SCORE FOR KD
0392U	DRUG METABOLISM (DEPRESSION, ANXIETY, ATTENTION DEFICIT HYPERACTIVITY DISORDER [ADHD]), GENE-DRUG INTERACTIONS, VARIANT ANALYSIS OF 16 GENES, INCLUDING DELETION/DUPLICATION ANALYSIS OF CYP2D6, REPORTED AS IMPACT OF GENE-DRUG INTERACTION FOR EACH DRUG
0400U	OBSTETRICS (EXPANDED CARRIER SCREENING), 145 GENES BY NEXT- GENERATION SEQUENCING, FRAGMENT ANALYSIS AND MULTIPLEX LIGATIONDEPENDENT PROBE AMPLIFICATION, DNA, REPORTED AS CARRIER POSITIVE OR NEGATIVE
0401U	CARDIOLOGY (CORONARY HEART DISEASE [CAD]), 9 GENES (12 VARIANTS), TARGETED VARIANT GENOTYPING, BLOOD, SALIVA, OR BUCCAL SWAB, ALGORITHM REPORTED AS A GENETIC RISK SCORE FOR A CORONARY EVENT

CODE	DESCRIPTION
0411U	PSYCHIATRY (EG, DEPRESSION, ANXIETY, ATTENTION DEFICIT HYPERACTIVITY DISORDER [ADHD]), GENOMIC ANALYSIS PANEL, VARIANT ANALYSIS OF 15 GENES, INCLUDING DELETION/DUPLICATION ANALYSIS OF CYP2D6
0417U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), WHOLE MITOCHONDRIAL GENOME SEQUENCE WITH HETEROPLASMY DETECTION AND DELETION ANALYSIS, NUCLEAR-ENCODED MITOCHONDRIAL GENE ANALYSIS OF 335 NUCLEAR GENES, INCLUDING SEQUENCE CHANGES, DELETIONS, INSERTIONS, AND COPY NUMBER VARIANTS ANALYSIS, BLOOD OR SALIVA, IDENTIFICATION AND CATEGORIZATION OF MITOCHONDRIAL DISORDER-ASSOCIATED GENETIC VARIANTS
0419U	NEUROPSYCHIATRY (EG, DEPRESSION, ANXIETY), GENOMIC SEQUENCE ANALYSIS PANEL, VARIANT ANALYSIS OF 13 GENES, SALIVA OR BUCCAL SWAB, REPORT OF EACH GENE PHENOTYPE
0425U	GENOME (EG, UNEXPLAINED CONSTITUTIONAL OR HERITABLE DISORDER OR SYNDROME), RAPID SEQUENCE ANALYSIS, EACH COMPARATOR GENOME (EG, PARENTS, SIBLINGS)
0426U	GENOME (EG, UNEXPLAINED CONSTITUTIONAL OR HERITABLE DISORDER OR SYNDROME), ULTRA-RAPID SEQUENCE ANALYSIS
0439U	CARDIOLOGY (CORONARY HEART DISEASE [CHD]), DNA, ANALYSIS OF 5 SINGLE- NUCLEOTIDE POLYMORPHISMS (SNPS) (RS11716050 [LOC105376934], RS6560711 [WDR37], RS3735222 [SCIN/LOC107986769], RS6820447 [INTERGENIC], AND RS9638144 [ESYT2]) AND 3 DNA METHYLATION MARKERS (CG00300879 [TRANSCRIPTION START SITE {TSS200} OF CNKSR1], CG09552548 [INTERGENIC], AND CG14789911 [BODY OF SPATC1L]), QPCR AND DIGITAL PCR, WHOLE BLOOD, ALGORITHM REPORTED AS A 4-TIERED RISK SCORE FOR A 3-YEAR RISK OF SYMPTOMATIC CHD
0440U	CARDIOLOGY (CORONARY HEART DISEASE [CHD]), DNA, ANALYSIS OF 10 SINGLE- NUCLEOTIDE POLYMORPHISMS (SNPS) (RS710987 [LINC010019], RS1333048 [CDKN2B-AS1], RS12129789 [KCND3], RS942317 [KTN1-AS1], RS1441433 [PPP3CA], RS2869675 [PREX1], RS4639796 [ZBTB41], RS4376434 [LINC00972], RS12714414 [TMEM18], AND RS7585056 [TMEM18]) AND 6 DNA METHYLATION MARKERS (CG03725309 [SARS1], CG12586707 [CXCL1, CG04988978 [MPO], CG17901584 [DHCR24-DT], CG21161138 [AHRR], AND CG12655112 [EHD4]), QPCR AND DIGITAL PCR, WHOLE BLOOD, ALGORITHM REPORTED AS DETECTED OR NOT DETECTED FOR CHD
0449U	CARRIER SCREENING FOR SEVERE INHERITED CONDITIONS (EG, CYSTIC FIBROSIS, SPINAL MUSCULAR ATROPHY, BETA HEMOGLOBINOPATHIES [INCLUDING SICKLE CELL DISEASE], ALPHA THALASSEMIA), REGARDLESS OF RACE OR SELF-IDENTIFIED ANCESTRY, GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE ANALYSIS OF 5 GENES (CFTR, SMN1, HBB, HBA1, HBA2)
0454U	RARE DISEASES (CONSTITUTIONAL/HERITABLE DISORDERS), IDENTIFICATION OF

DESCRIPTION
COPY NUMBER VARIATIONS, INVERSIONS, INSERTIONS, TRANSLOCATIONS, AND OTHER STRUCTURAL VARIANTS BY OPTICAL GENOME MAPPING
ONCOLOGY, WHOLE BLOOD OR BUCCAL, DNA SINGLE-NUCLEOTIDE POLYMORPHISM (SNP) GENOTYPING BY REAL-TIME PCR OF 24 GENES, WITH VARIANT ANALYSIS AND REPORTED PHENOTYPES
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
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
HEREDITARY PROSTATE CANCERRELATED 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
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
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
DESCRIPTION
AUTOINFLAMMATORY DISEASE (VEXAS SYNDROME), DNA, UBA1 GENE MUTATIONS, TARGETED VARIANT ANALYSIS (M41T, M41V, M41L, C.118-2A>C, C.118-1G>C, C.118-9_118-2DEL, S56F, S621C)
DRUG METABOLISM, WHOLE BLOOD, PHARMACOGENOMIC GENOTYPING OF 40 GENES AND CYP2D6 COPY NUMBER VARIANT ANALYSIS, REPORTED AS

For tests billed under the Not Otherwise Classified (NOC) CPT® code 81479:

Tests billed under the NOC $CPT^{(R)}$ code 81479 that are used for germline testing and are further defined by a unique identifier (such as a Z-Identifier code) are limited to once per lifetime, as are such tests that are duplicative.

If non-duplicative testing of such a test is considered medically reasonable and necessary, please use a -77 modifier to trigger a medical review.

Group 2 Codes: (1 Code)

CODE	DESCRIPTION
81479	UNLISTED MOLECULAR PATHOLOGY PROCEDURE

CPT/HCPCS Modifiers

Group 1 Paragraph:

N/A

Group 1 Codes: (1 Code)

CODE	DESCRIPTION
77	REPEAT PROCEDURE BY ANOTHER PHYSICIAN: THE PHYSICIAN MAY NEED TO
	INDICATE THAT A BASIC PROCEDURE OR SERVICE PERFORMED BY ANOTHER
	PHYSICIAN HAD TO BE REPEATED. THIS SITUATION MAY BE REPORTED BY ADDING
	MODIFIER -77 TO THE REPEATED PROCEDURE/SERVICE OR THE SEPARATE FIVE
	DIGIT MODIFIER CODE 09977 MAY BE USED.

ICD-10-CM Codes that Support Medical Necessity

Group 1 Paragraph:

N/A

Group 1 Codes: (1 Code)

CODE	DESCRIPTION
XX000	Not Applicable

ICD-10-CM Codes that DO NOT Support Medical Necessity		
Group 1 Paragraph:		
N/A		
Group 1 Codes:		

N/A

ICD-10-PCS Codes

Group 1 Paragraph:

N/A

Group 1 Codes:

N/A

Additional ICD-10 Information

N/A

Bill Type Codes

Contractors may specify Bill Types to help providers identify those Bill Types typically used to report this service. Absence of a Bill Type does not guarantee that the article does not apply to that Bill Type. Complete absence of all Bill Types indicates that coverage is not influenced by Bill Type and the article should be assumed to apply equally to all claims.

CODE	DESCRIPTION
999x	Not Applicable

Revenue Codes

Contractors may specify Revenue Codes to help providers identify those Revenue Codes typically used to report this service. In most instances Revenue Codes are purely advisory. Unless specified in the article, services reported under other Revenue Codes are equally subject to this coverage determination. Complete absence of all Revenue Codes indicates that coverage is not influenced by Revenue Code and the article should be assumed to apply equally to all Revenue Codes.

N/A

Other Coding Information		
Group 1 Paragraph:		
N/A		
Group 1 Codes:		
CODE		
N/A		

Revision History Information

REVISION HISTORY DATE	REVISION HISTORY NUMBER	REVISION HISTORY EXPLANATION
10/01/2024	R14	Under CPT/HCPCS Codes Group 1: Codes added 0476U, 0477U, 0500U, and 0516U. Deleted 0078U. This revision is due to the 2024 Q4 CPT/HCPCS Code Update and is effective 10/1/2024.
09/05/2024	R13	Under CPT/HCPCS Codes Group 1: Codes deleted 81288, 81345, 81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408, 81416, 81417, 81420, 81422, 81426, and 81427. This revision is effective 9/5/2024.
07/01/2024	R12	Under CPT/HCPCS Codes Group 1: Codes added 0454U, 0460U, 0461U, 0474U, and 0475U. This revision is due to the 2024 Q3 CPT/HCPCS Code Update and is effective 7/1/2024.
04/01/2024	R11	Under CPT/HCPCS Codes Group 1: Codes added 0439U, 0440U, and 0449U. This revision is due to the 2024 Q2 CPT/HCPCS Code Update and is effective 4/1/2024.
01/01/2024	R10	Under CPT/HCPCS Codes Group 1: Codes the description was revised for 81171, 81172, 81242, and 81243. Added 0425U and 0426U. This revision is due to the 2024 Annual/Q1 CPT/HCPCS Code Update and is effective 1/1/2024.
10/01/2023	R9	Updated to indicate this article is an LCD Reference Article.
10/01/2023	R8	Under CPT/HCPCS Codes Group 1: Codes the description was revised for 0258U, 0269U, 0271U, 0272U, 0274U, 0277U, and 0278U. Added 0411U, 0417U, and 0419U. This revision is due to the 2023 Q4 CPT/HCPCS Code Update and is effective on 10/1/2023.
07/01/2023	R7	Under CMS National Coverage Policy updated section heading. This revision is effective on 7/1/2023.
		Under CPT/HCPCS Codes Group 1: Codes added 0389U, 0392U, 0400U, and 0401U. This revision is due to the 2023 Q3 CPT/HCPCS Code Update and is effective on 7/1/2023.

REVISION HISTORY DATE	REVISION HISTORY NUMBER	REVISION HISTORY EXPLANATION
04/01/2023	R6	Under CPT/HCPCS Codes Group 1: Codes added 0378U and 0380U. This revision is due to the 2023 Q2 CPT/HCPCS Code Update and is effective on 4/1/2023.
01/01/2023	R5	Under CPT/HCPCS Codes Group 1: Codes the description was revised for 81330. Added 81418, 81441, and 0355U. This revision is due to the 2023 Annual/Q1 CPT/HCPCS Code Update and is effective on January 1, 2023. Under CPT/HCPCS Codes Group 1: Codes the description was revised for 0276U. This revision is due to the 2022 Q4 CPT/HCPCS Code Update and is effective for dates of
		service on or after 10/1/2022.
10/01/2022	R4	Under CPT/HCPCS Codes Group 1: Codes added 81307 and 81309. This revision is effective for dates of service on or after 10/1/2022.
		Under CPT/HCPCS Codes Group 1: Codes deleted 0012U. Added 0345U, 0347U, 0348U, 0349U, and 0350U. This revision is due to the Q4 CPT/HCPCS Code Update and is effective for dates of service on or after 10/1/2022.
05/12/2022	R3	Under CPT/HCPCS Codes Group 1: Codes added 0318U. This revision is due to the Q2 CPT/HCPCS Code Update and is effective for dates of service on or after 4/1/2022.
		Under CMS National Coverage Policy added regulation CMS Internet-Only Manual, Pub. 100-04, Medicare Claims Processing Manual, Chapter 16, §50.5 Jurisdiction of Laboratory Claims, §60.1.2 Independent Laboratory Specimen Drawing, §60.2 Travel Allowance. Under CPT/HCPCS Codes Group 1: Codes deleted 81340, 81341, and 81342. Added 81220, 81221, 81222, 81223, 81224, 81419, 81493, 81554, 0001U, 0004M, 0012U, 0029U, 0030U, 0031U, 0032U, 0033U, 0034U, 0070U, 0071U, 0072U, 0073U, 0074U, 0075U, 0076U, 0078U, 0079U, 0084U, 0094U, 0101U, 0102U, 0103U, 0129U, 0130U, 0131U, 0132U, 0133U, 0134U, 0135U, 0136U, 0137U, 0138U, 0156U, 0157U, 0158U, 0159U, 0160U, 0161U, 0162U, 0169U, 0170U, 0173U, 0175U, 0180U, 0181U, 0182U, 0183U, 0184U, 0185U, 0186U, 0187U, 0188U, 0189U, 0190U, 0191U, 0192U, 0193U, 0194U, 0195U, 0196U, 0197U, 0198U, 0199U, 0200U, 0201U, 0203U, 0205U, 0209U, 0212U, 0213U, 0214U, 0215U, 0216U, 0217U, 0218U, 0221U, 0222U, 0230U, 0231U, 0232U, 0233U, 0234U, 0235U, 0236U, 0237U, 0238U, 0246U, 0258U, 0260U, 0264U, 0265U, 0266U, 0267U, 0268U, 0269U, 0270U, 0271U, 0272U, 0273U, 0274U, 0276U, 0277U, 0278U, 0282U, 0286U, 0289U, 0290U , 0291U, 0292U, 0293U, and 0294U. This revision is effective 5/12/2022.
01/01/2022	R2	Under CPT/HCPCS Code Group 1: Codes the description was revised for 81228 and 81229. The revision is due to the 2022 Annual CPT/HCPCS Code Update and is effective on January 1, 2022.

REVISION HISTORY DATE	REVISION HISTORY NUMBER	REVISION HISTORY EXPLANATION
01/01/2021	R1	Under CPT/HCPCS Codes Group 1: Added 81168, 81191, 81192, 81193, 81194, 81278, 81279, 81338, 81339, 81347, 81348, 81351, 81352, 81353, 81357, 81360, 81419.
		This revision is due to the Q1 2021 CPT/HCPCS Code Update and is effective for dates of service on or after 01/01/2021.

Associated Documents

Related Local Coverage Documents

LCDs

L38353 - MoIDX: Repeat Germline Testing

Related National Coverage Documents

N/A

Statutory Requirements URLs

N/A

Rules and Regulations URLs

N/A

CMS Manual Explanations URLs

N/A

Other URLs

N/A

Public Versions

UPDATED ON	EFFECTIVE DATES	STATUS
Some older versions have been archived. Please visit the MCD Archive Site to retrieve them.		
09/30/2024	10/01/2024 - N/A	Currently in Effect (This Version)
08/29/2024	09/05/2024 - 09/30/2024	Superseded
07/01/2024	07/01/2024 - 09/04/2024	Superseded
04/01/2024	04/01/2024 - 06/30/2024	Superseded
01/05/2024	01/01/2024 - 03/31/2024	Superseded
11/22/2023	10/01/2023 - 12/31/2023	Superseded
10/16/2023	10/01/2023 - N/A	Superseded

Keywords

N/A