Clinical Laboratory Fee Schedule CY 2013 Updates

Please note that this listing includes the most recent codes provided by the American Medical Association (AMA), and that it is subject to change. Any changes will be updated as they occur.

New Test Codes

Tier 1 Molecular Pathology Procedures

812XX APC (adenomatous polyposis coli) (eg, familial adenomatosis polyposis [FAP], attenuated FAP) gene analysis; full gene sequence

812XX APC (adenomatous polyposis coli) (eg, familial adenomatosis polyposis [FAP], attenuated FAP) gene analysis; known familial variants

812XX APC (adenomatous polyposis coli) (eg, familial adenomatosis polyposis [FAP], attenuated FAP) gene analysis; duplication/deletion variants

812XX *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)

812XX *GJB2 (gap junction protein, beta 2, 26kDa; connexin 26)* (eg, nonsyndromic hearing loss) gene analysis; full gene sequence

812XX *GJB2 (gap junction protein, beta 2, 26kDa; connexin 26)* (eg, nonsyndromic hearing loss) gene analysis; known familial variants

812XX *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)])

813XX *PTEN (phosphatase and tensin homolog)* (eg, Cowden syndrome, *PTEN* hamartoma tumor syndrome) gene analysis; full sequence analysis

813XX *PTEN (phosphatase and tensin homolog)* (eg, Cowden syndrome, *PTEN* hamartoma tumor syndrome) gene analysis; known familial variant

813XX *PTEN (phosphatase and tensin homolog)* (eg, Cowden syndrome, *PTEN* hamartoma tumor syndrome) gene analysis; duplication/deletion variant

813XX *PMP22 (peripheral myelin protein 22)* (eg, Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; duplication/deletion analysis

813XX *PMP22 (peripheral myelin protein 22)* (eg, Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; full sequence analysis

813XX *PMP22 (peripheral myelin protein 22)* (eg, Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; known familial variant

81200 ASPA (aspartoacylase (eg, Canavan disease) gene analysis, common variants (eg, E285A, Y231X)

81205 BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (eg, Maple syrup urine disease) gene analysis, common variants (eg, R183P, G278S, E422X)

81206 BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; major breakpoint, qualitative or quantitative

81207 BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; minor breakpoint, qualitative or quantitative

81208 BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; other breakpoint, qualitative or quantitative

81209 BLM (Bloom syndrome, RecQ helicase-like) (eg, Bloom syndrome) gene analysis, 2281del6ins7 variant

81210 BRAF (v-raf murine sarcoma viral oncogene humolog B1) (eg, colon cancer), gene analysis, V600E variant

81211 BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; full sequence analysis and common duplication/deletion variants in BRCA1 (ie, exon 13 del 3.835kb, exon 13 dup 6kb, exon 14-20 del 26 kb, exon 22 del 510bp, exon 8-9 del 7.1kb)

81212 BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; 185delAG, 5385insC, 6174delT variants

81213 BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; uncommon duplication/deletion variants

81214 BRCA1 (breast cancer 1) (eg, hereditary breast and ovarian cancer) gene analysis; full sequence analysis and common duplication/deletion variants

(ie, exon 13 del 3.835kb, exon 13 dup 6kb, exon 14-20 del 26kb, exon 22 del 510bp, exon 8-9 del 7.1kb)

81215 BRCA1 (breast cancer 1) (eg, hereditary breast and ovarian cancer) gene analysis; known familial variant

81216 BRCA2 (breast cancer 2) (eg, hereditary breast and ovarian cancer) gene analysis; full sequence analysis

81217 BRCA2 (breast cancer 2) (eg, hereditary breast and ovarian cancer) gene analysis; known familial variant

81220 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)

81221 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; known familial variants

81222 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; duplication/deletion variants

81223 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; full gene sequence

81224 CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; intron 8 poly-T analysis (eg, male infertility)

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)

81228 Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (eg, Bacterial Artificial Chromosome [BAC] or oligo-based comparative genomic hybridization [CGH] microarray analysis)

81229 Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number and single nucleotide

polymorphism (SNP) variants for chromosomal abnormalities

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 1 mental retardation 1) (eg, fragile X mental retardation) gene analysis; evaluation to detect abnormal (eg, expanded) alleles

81244 FMR1 (Fragile 1 mental retardation 1) (eg, fragile X mental retardation) gene analysis; characterization of alleles (eg, expanded size and methylation status)

81245 FLT3 (fms-related tyrosine kinase 3) (eg, acute myeloid leukemia), gene analysis, internal tandem duplication (ITD) variants (ie, exons 14, 15)

81250 G6PC (glucose-6-phosphatase, catalytic subunit) (eg, Glycogen storage disease, Type 1a, von Gierke disease) gene analysis, common variants (eg, R83C, Q347X)

81251 GBA (glucosidase, beta, acid) (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+1G>A)

81255 HEXA (hexosaminidase A (alpha polypeptide) (eg, Tay-Sachs disease) gene analysis, common variants (eg, 1278insTATC, 1421+1G>C, G269S)

81256 HFE (hemochromatosis) (eg, hereditary hemochromatosis) gene analysis, common variants (eg, C282Y, H63D)

81257 HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis, for common deletions or variant (eg, Southeast Asian, Thai, Filipino, Mediterranean, alpha3.7, alpha4.2, alpha20.5, and Constant Spring)

81260 IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (eg, familial dysautonomia) gene analysis,

common variants (eg, 2507+6T>C, R696P)

81261 IGH@ (Immunoglobulin heavy chain locus) (eg, leukemias and lymphomas, B-cell), gene rearrangement analysis to detect abnormal clonal population(s); amplified methodology (eg, polymerase chain reaction)

81262 IGH@ (Immunoglobulin heavy chain locus) (eg, leukemias and lymphomas, B-cell), gene rearrangement analysis to detect abnormal clonal population(s); direct probe methodology (eg, Southern blot)

81263 IGH@ (Immunoglobulin heavy chain locus) (eg, leukemia and lymphoma, B-cell), variable region somatic mutation analysis

81264 IGK@ (Immunoglobulin kappa light chain locus) (eg, leukemia and lymphoma, B-cell), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)

81265 Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing, or maternal cell contamination of fetal cells

81266 Comparative analysis using Short Tandem Repeat (STR) markers; each additional specimen (eg, additional cord blood donor, additional fetal samples from different cultures, or additional zygosity in multiple birth pregnancies) (List separately in addition to code for primary procedure)

81267 Chimerism (engraftment) analysis, post hematopoietic stem cell transplantation specimen, includes comparison to previously performed baseline analyses; without cell selection

81268 Chimerism (engraftment) analysis, post hematopoietic stem cell transplantation specimen, includes comparison to previously performed baseline analyses; with cell selection (eg, CD3, CD33), each cell type

81270 JAK2 (Janus kinase 2) (eg, myeloproliferative disorder) gene analysis, p.Val617Phe (V617F) variant

81275 KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene) (eg, carcinoma)

gene analysis, variants in codons 12 and 13

81280 Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); full sequence analysis

81281 Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); known familial sequence variant

81282 Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); duplication/deletion variants

81290 MCOLN1 (mucolipin 1) (eg, Mucolipidosis, type IV) gene alalysis, common variants (eg, IVS3-2A>G, del6.4kb)

81291 MTHFR (5, 10-methylenetetrahydrofolate reductase) (eg, hereditary hypercoagulability) gene alalysis, common variants (eg, 677T, 1298C)

81292 MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

81293 MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

81294 MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

81295 MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

81296 MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

81297 MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

81298 MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

81299 MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

81300 MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

81301 Microsatellite instability analysis (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) of markers for mismatch repair deficiency (eg, BAT25, BAT26), includes comparison of neoplastic and normal tissue, if performed

81302 MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; full sequence analysis

81303 MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; known familial variant

81304 MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; duplication/deletion variants

81310 NPM1 (nucleophosmin) (eg, acute myeloid leukemia) gene analysis; exon 12 variants

81315 PML/RARalpha, (t(15;17)), (PML-RARA regulated adaptor molecule 1) (eg, promyelocytic leukemia) translocation analysis; common breakpoints (eg, intron 3 and intron 6), qualitative or quantitative

81316 PML/RARalpha, (t(15;17)), (PML-RARA regulated adaptor molecule 1) (eg, promyelocytic leukemia) translocation analysis; single breakpoint (eg, intron 3, intron 6, or exon 6), qualitative or quantitative

81317 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary

non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

81318 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

81319 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

81330 SMPD1 (sphingomyelin phosphodiesterase 1, acid sysosomal) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330)

81331 SNRPN/UBE3A, (small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A) (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis

81332 SERPINA1 (serpin peptidase inhibitor, clade A, alpha-1 antiproteinase, antitrypsin, member 1) (eg, alpha-1-antitrypsin deficiency), gene analysis, common variants (eg, *S and *Z)

81340 TCB@ (T cell antigen receptor, beta) (eg, leukemia and lymphoma), gene rearrangement analysis to detect abnormal clonal population(s); using amplification methodology (eg, polymerase chain reaction)

81341 TCB@ (T cell antigen receptor, beta) (eg, leukemia and lymphoma), gene rearrangement analysis to detect abnormal clonal population(s); using direct probe methodology (eg, Southern blot)

81342 TCG@ (T cell receptor, gamma) (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)

81350 UGT1A1 (UDP glucuronosyltransferase 1 family, polypeptide A1) (eg, irinotecan metabolism), gene analysis, common variants (eg, *28, *36, *37)

81355 VKORC1 (vitamin K epoxide reductase complex, subunit 1) (eg, warfarin metabolism), gene analysis, common variants (eg, -1639/3673)

81370 HLA Class I and II typing, low resolution (eg, antigen equivalents); HLA-A, -B, -C, -DRB1/3/4/5, and -DQB1

81371 HLA Class I and II typing, low resolution (eg, antigen equivalents); HLA-A, -B, and -DRB1/3/4/5 (eg, verification typing)

81372 HLA Class I typing, low resolution (eg, antigen equivalents); complete (ie, HLA-A, -B, and -C)

81373 HLA Class I typing, low resolution (eg, antigen equivalents); one locus (eg, HLA-A, -B, or -C), each

81374 HLA Class I typing, low resolution (eg, antigen equivalents); one antigen equivalent (eg, B*27), each

81375 HLA Class II typing, low resolution (eg, antigen equivalents); HLA-DRB1/3/4/5 and -DQB1

81376 HLA Class II typing, low resolution (eg, antigen equivalents); one locus (eg, HLA-DRB1/3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1), each

81377 HLA Class II typing, low resolution (eg, antigen equivalents); one antigen equivalent, each

81378 HLA Class I and II typing, high resolution (ie, alleles or allele groups), HLA-A, -B, -C, and -DRB1

81379 HLA Class I typing, high resolution (ie, alleles or allele groups); complete (ie, HLA-A, -B, and -C)

81380 HLA Class I typing, high resolution (ie, alleles or allele groups); one locus (eg, HLA-A, -B, or -C), each

81381 HLA Class I typing, high resolution (ie, alleles or allele groups); one allele or allele group (eg, B*57:01P), each

81382 HLA Class II typing, high resolution (ie, alleles or allele groups); one locus (eg, HLA-DRB1, -DRB3, -DRB4, -DRB5, -DQB1, -DQA1, -DPB1, or -DPA1), each

81383 HLA Class II typing, high resolution (ie, alleles or allele groups); one allele or allele group (eg, HLA-DQB1*06:02P), each

Tier 2 Molecular Pathology Procedures

81400 Molecular pathology procedure, Level 1 (eg, identification of single germline variant [eg, SNP] by techniques such as restriction enzyme digestion or melt curve analysis)

ABCC8 (ATP-binding cassette, sub-family C [CFTR/MRP], member 8) (eg, familial hyperinsulinism), F1388del variant

ACADM (acyl-CoA dehydrogenase, C-4 to C-12 straight chain, MCAD) (eg, medium chain acyl dehydrogenase deficiency), K304E variant

ACE (angiotensin converting enzyme) (eg, hereditary blood pressure regulation), insertion/deletion variant

AGTR1 (angiotensin II receptor, type 1) (eg, essential hypertension), 1166A>C variant

CCR5 (chemokine C-C motif receptor 5) (eg, HIV resistance), 32 bp deletion mutation/794 825del32 deletion

CLRN1 (clarin 1) (eg, Usher syndrome, type 3), N48K variant

DPYD (dihydropyrimidine dehydrogenase) (eg, 5-fluorouracil/5-FU and capecitabine drug metabolism), IVS14+1G>A variant

F2 (coagulation factor 2) (eg, hereditary hypercoagulability), 1199G>A variant

F5 (coagulation factor V) (eg, hereditary hypercoagulability), HR2 variant

F7 (coagulation factor VII [serum prothrombin conversion accelerator]) (eg, hereditary hypercoagulability), R353Q variant

F13B (coagulation factor XIII, B polypeptide) (eg, hereditary hypercoagulability), V34L variant

FGB (fibrinogen beta chain) (eg, hereditary ischemic heart disease), -455G>A variant

FGFR3 (fibroblast growth factor receptor 3) (eg, Muenke syndrome), P250R variant

Human Platelet Antigen 1 genotyping (HPA-1), ITGB3 (integrin, beta 3 [platelet glycoprotein llla], antigen CD61 [GPllla]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-1a/b (L33P)

Human Platelet Antigen 2 genotyping (HPA-2), GP1BA (glycoprotein lb [platelet], alpha polypeptide [GPlba]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-2a/b (T145M)

Human Platelet Antigen 3 genotyping (HPA-3), ITGA2B (integrin, alpha 2b [platelet glycoprotein llb of llb/llla complex], antigen CD41 [GPllb]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-3a/b (1843S)

Human Platelet Antigen 4 genotyping (HPA-4), ITGB3 (integrin, beta 3 [platelet glycoprotein llla], antigen CD61 [GPllla]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-4a/b (R143Q)

Human Platelet Antigen 5 genotyping (HPA-5), ITGA2 (integrin, alpha 2 [CD49B, alpha 2 subunit of VLA-2 receptor] [GPla]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-5a/b (K505E)

Human Platelet Antigen 6 genotyping (HPA-6w), ITGB3 (integrin, beta 3 [platelet glycoprotein llla, antigen CD61] [GPllla]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-6a/b (R489Q)

Human Platelet Antigen 9 genotyping (HPA-9w), ITGA2B (integrin, alpha 2b [platelet glycoprotein llb of llb/llla complex, antigen CD41] [GPllb]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-9a/b (V837M)

Human Platelet Antigen 15 genotyping (HPA-15), CD109 (CD109 molecule) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-15a/b(S682Y)

IVD (isovaleryl-CoA dehydrogenase) (eg, isovaleric academia), A282V variant

SERPINE1 (serpine peptidase inhibitor clade E, member 1, plasminogen activator inhibitor -1, PAI-1) (eg, thrombophilia), 4G variant

-SHOC2 (soc-2 suppressor of clear homolog) (eg, Noonan-like syndrome with loose anagen hair), S2G variant

-SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy), exon 7 deletion

-SRY (sex determining region Y) (eg, 46, XX testicular disorder of sex development, gonadal dysgenesis), gene analysis

-TOR1A (torsin family 1, member A [torsin A])(eg, early-onset primary dystonia [DYT1]), 907_909delGAG (904_906delGAG) variant

81401 Molecular pathology procedure, Level 2 (eg, 2-10 SNPs, 1 methylated variant, or 1 somatic variant [typically using nonsequencing target variant analysis], or detection of a dynamic mutation disorder/triplet repeat)

ABL (c-abl oncogene 1, receptor tyrosine kinase) (eg, acquired imatinib resistance), T315I variant

ACADM (acyl-CoA deydrogenase, C-4 to C-12 straight chain, MCAD) (eg, medium chain acyl dehydrogenase deficiency), commons variants (eg, K304E, Y42H)

ADRB2 (adrenergic beta-2 receptor surface) (eg, drug metabolism), common variants (eg, G16R, Q27E) APOB (apolipoprotein B) (eg, familial hypercholesterolemia type B), common variants (eg, R3500Q, R3500W)

APOE (apolipoprotein E)-_(eg, hyperlipoproteinemia type III, cardiovascular disease, Alzheimer disease), common variants (eg, *2, *3, *4)

AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation), characterization of alleles (eg, expanded size or methylation status)

ATN1 (atrophin 1) (eg, dentatorubral-pallidoluysian atrophy), evaluation to detect abnormal (eg, expanded) alleles

*CBFB/MYH11 (inv(16)-*_(eg, acute myeloid leukemia), qualitative, and quantitative, if performed

CBS (cystathionine-beta-synthase) (eg, homocystinuria, cystathionine beta-synthase deficiency), common variants (eg, I278T, G307S)

CCND1/IGH (BCL1/lgH, t(11;14)) (eg, mantle cell lymphoma) translocation analysis, major breakpoint, qualitative, and quantitative, if performed

*CFH/ARMS2 (complement factor H/age-related maculopathy susceptibility 2)-*_(eg, macular degeneration), common variants (eg, Y402H [CFH], A69S [ARMS2])

*CYP3A4 (cytochrome P450, family 3, subfamily A, polypeptide 4)-*_(eg, drug metabolism), common variants (eg, *2, *3, *4, *5, *6)

CYP3A5 (cytochrome P450, family 3, subfamily A, polypeptide 5) (eg, drug metabolism), common variants (eg, *2, *3, *4, *5, *6)

DMPK (dystrophia myotonica-protein kinase) (eg, myotonic dystrophy, type 1), evaluation to detect abnormal (eg, expanded) alleles

E2A/PBX1 (t(1;19)) (eg, acute lymphocytic leukemia), translocation analysis, qualitative, and quantitative, if performed

EML4/ALK (inv(2)) (eg, non-small cell lung cancer), translocation or inversion analysis

ETV6/RUNX1 (t(12;21)) (eg, acute lymphocytic leukemia), translocation analysis, qualitative, and quantitative, if performed

EWSR1/ERG (t(21;22)) (eg, Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative, and quantitative, if performed

EWSR1/FLI1 (t(11;22)) (eg, Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative, and quantitative, if performed

EWSR1/WT1 (t(11;22)) (eg, Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative, and quantitative, if performed

F11 (coagulation factor XI) (eg, coagulation disorder), common variants (eg, E117X [Type II], F283L [Type III], IVS14del14, and IVS14+1G>A [Type I])

FGFR3 (fibroblast growth factor receptor 3) (eg, achondroplasia), common variants (eg, 1138G>A, 1138G>C), 1620C>A, 1620C>G

FIP1L1/PDGFRA (del[4q12])-_(eg, imatinib-sensitive chronic eosinophilic leukemia), qualitative, and quantitative, if performed

FOXO1/PAX3 (t(1;13)) (eg, Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative, and quantitative, if performed

FOXO1/PAX7 (t(2;13)) (eg, Ewing sarcoma/peripheral neuroectodermal tumor), translocation anlaysis, qualitative, and quantitative, if performed

FXN (frataxin) (eg, Friedreich ataxia), evaluation to detect abnormal (expanded) alleles

GALT (galactose-1-phosphate uridylyltransferase)-_(eg, galactosemia), common variants (eg, Q188R, S135L, K285N, T138M, L195P, Y209C, IVS2-2A>G, P171S, del5kb, N314D, L218L/N314D

H19 (imprinted maternally expressed transcript [non-protein coding]) (eg, Beckwith-Wiedemann syndrome), methylation analysis

HBB (hemoglobin, beta) (eg, sickle cell anemia, hemoglobin C, hemoglobin E), common variants (eg, HbS, HbC, HbE)

HTT (huntingtin) (eg, Huntington disease), evaluation to detect abnormal alleles)expanded , eg(

KCNQ1OT1 (KCNQ1 overlapping transcript 1 [non-protein coding]) (eg, Beckwith-Wiedemann syndrome), methylation analysis

MEG3/DLK1 (maternally expressed 3 [non-protein coding]/delta-like 1 homolog [Drosophila]) (eg, intrauterine growth retardation), methylation analysis

MLL/AFF1 (t(4;11)) (eg, acute lymphoblastic leukemia), translocation analysis, qualitative, and quantitative, if performed

MLL/MLLT3 (t(9;11)) (eg, acute myeloid leukemia), translocation analysis, qualitative, and quantitative, if performed

MT-RNR1 (mitochondrially encoded 12S RNA) (eg, nonsyndromic hearing loss), common variants (eg, m, 1555A>G, m. 1494C>T)

MUTYH (mutY homolog [E. coli]) (eg, MYH-associated polyposis), common variants (eg, Y165C, G382D

MT-ATP6 (mitochondrially encoded ATP synthase 6) (eg, neuropathy with ataxia and retinitis pigmentosa [NARP], Leigh syndrome), common variants (eg, m.8993T>G, m.8993T>C

MT-ND4, MT-ND6 (mitochondrially encoded NADH dehydrogenase 4, mitochondrially encoded NADH dehydrogenase 6) (eg, Leber hereditary optic neuropathy [LHON]), common variants (eg, m. 11778G>A, m. 3460G>A, m. 14484T>C)

MT-TK (mitochondrially encoded tRNA lysine) (eg, myoclonic epilepsy with ragged-red fibers [MERRF]), common variants (eg, m.8344A>G, m.8356T>C)

MT-TL1 (mitochondrially encoded tRNA leucine 1 [UUA/G]) (eg, diabetes and hearing loss), common variants (eg, m.3243A>G, m.14709 T>C) MT-TL1,

MT-ND5 (mitochondrially encoded tRNA leucine 1 [UUA/G], mitochondrially encoded NADH dehydrogenase 5) (eg, mitochondrial encephalopathy with lactic acidosis and stroke-like episodes [MELAS]), common variants (eg, m.3243A>G, m.3271T>C, m.3252A>G, m.13513G>A)

MT-TS1, MT-RNR1 (mitochondrially encoded tRNA serine 1 [UCN], mitochondrially encoded 12S RNA) (eg, nonsyndromic sensorineural deafness [including aminoglycoside-induced nonsyndromic deafness]), common variants (eg, m.7445A>G, m.1555A>G)

NPM1/ALK (t(2;5)) (eg, anaplastic large cell lymphoma), translocation analysis PAX8/PPARG (t(2;3) (q13;p25)) (eg, follicular thyroid carcinoma), translocation analysis PRSS1 (protease, serine, 1 [trypsin 1]) (eg, hereditary pancreatitis), common variants (eg, N29I, A16V, R122H)

PYGM (phosphorylase, glycogen, muscle) (eg, glycogen storage disease type V, McArdle disease), common variants (eg, R50X, G205S)

RUNX1/RUNX1T1 (t(8;21)) (eg, acute myeloid leukemia) translocation analysis, qualitative, and quantitative, if performed

SEPT9 (Septin 9) (eg, colon cancer), methylation analysis

SMN1/SMN2 (survival of motor neuron 1, telomeric/survival of motor neuron 2, centromeric) (eg, spinal muscular atrophy), dosage analysis (eg, carrier testing)

TPMT (thiopurine S-methyltransferase) (eg, drug metabolism), common variants (eg, *2, *3) TYMS (thymidylate synthetase) (eg, 5-fluorouracil/5-FU drug metabolism), tandem repeat variant

VWF (von Willebrand factor)-_(eg, von Willebrand disease type 2N), common variants (eg, T791M, R816W, R854Q)

81402 Molecular pathology procedure, Level 3 (eg, >10 SNPs, 2-10 methylated variants, or 2-10 somatic variants [typically using non-sequencing target variant analysis], immunoglobulin and T-cell receptor gene rearrangements, duplication/deletion variants of 1 exon, loss of heterozygosity [LOH], uniparental disomy [UPD])

Chromosome 18q- (eg, D18S55, D18S58, D18S61, D18S64, and D18S69) (eg, colon cancer), allelic imbalance assessment (ie, loss of heterozygosity)

CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide 2) (eg, congenital adrenal hyperplasia, 21-hydroxylase deficiency), common variants (eg, IVS2-13G, P30L, I172N, exon 6 mutation cluster [I235N, V236E, M238K], V281L, L307FfsX6, Q318X, R356W, P453S, G110VfsX21, 30-kb deletion variant

ESR1/PGR (receptor 1/progesterone receptor) ratio (eg, breast cancer)

KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog)

(eg, mastocytosis), common variants (eg, D816V, D816Y, D816F)

MEFV (Mediterranean fever) (eg, familial Mediterranean fever), common variants (eg, E148Q, P369S, F479L, M680I, I692del, M694V, M694I, K695R, V726A, A744S, R761H)

MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (eg, myeloproliferative disorder), common variants (eg, W515A, W515K, W515L, W515R)
TRD@ (T cell antigen receptor, delta) (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population

Uniparental disomy (UPD) (eg, Russell-Silver syndrome, Prader-Willi/Angelman syndrome), short tandem repeat (STR) analysis

81403 Molecular pathology procedure, Level 4 (eg, analysis of single exon by DNA sequence analysis, analysis of >10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)

ABL1 (c-abl oncogene1, receptor tyrosine kinase) (eg, acquired imatinib tyrosine kinase inhibitor resistance), variants in the kinase domain

ANG (angiogenin, ribonuclease, RNase A family, 5) (eg, amyotrophic lateral sclerosis), full gene sequence

CEBPA (CCAAT/enhancer binding protein [C/EBP], alpha) (eg, acute myeloid leukemia), full gene sequence

CEL (carboxyl ester lipase [bile salt-stimulated lipase]) (eg, maturity-onset diabetes of the young [MODY]), targeted sequence analysis of exon 11 (eg, c. 1686delT)

DAZ/SRY (deleted in azoospermia and sex determining region Y) (eg, male infertility), common deletions (eg, AZFa, AZFb, AZFc, AZFd)

F8 (coagulation factor VIII) (eg, hemophilia A), inversion analysis, intron 1 and intron 22A

(For targeted sequence analysis of multiple FGFR3 exons, use 81404)

GJB1 (gap junction protein, beta 1) (eg, Charcot-Marie-Tooth X-linked), full gene sequence

HBB (hemoglobin, beta, beta-globin) (eg, beta thalassemia), duplication/deletion analysis

HRAS (v-Ha-ras Harvey rat sarcoma viral oncogene homolog) (eg, Costello syndrome), exon 2 sequence
IDH1 (isocitrate dehydrogenase 1 [NADP+], soluble) (eg, glioma), common exon 4 variants (eg, R132H, R132C)

IDH2 (isocitrate dehydrogenase 2 [NADP+], mitochondrial) (eg, glioma), common exon 4 variants (eg, R140W, R172M)

JAK2 (Janus kinase 2) (eg, myeloproliferative disorder), exon 12 sequence and exon 13 sequence, if performed

Known familial variant not otherwise specified, for gene listed in Tier 1 or Tier 2, DNA sequence analysis, each variant exon

(For a known familial variant that is considered a common variant, use specific common variant Tier 1 or Tier 2 code)

KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene) (eg, carcinoma), gene analysis, variant(s) in exon 3 (eg, codon 61)

MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (eg, myeloproliferative disorder), exon 10 sequence

MT-RNR1 (mitochondrially encoded 12S RNA) (eg, nonsyndromic hearing loss), full gene sequence

MT-TS1 (mitochondrially encoded tRNA serine 1) (eg, nonsyndromic hearing loss), full gene sequence

SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy), known familial sequence variant(s)

VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), deletion/duplication analysis

VWF (von Willebrand factor) (eg, von Willebrand disease types 2A, 2B, 2M), targeted sequence analysis (eg, exon 28)

81404 Molecular pathology procedure, Level 5 (eg, analysis of 2-5 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 6-10 exons, or characterization of a dynamic mutation disorder/triplet repeat by Southern blot analysis)
ACADS (acyl-CoA dehydrogenase, C-2 to C-3 short chain) (eg, short chain acyl-CoA dehydrogenase deficiency), targeted sequence analysis (eg, exons 5 and 6)

AQP2 (aquaporin 2 [collecting duct]) (eg, nephrogenic diabetes insipidus), full gene sequence

ARX (aristaless related homeobox) (eg, X-linked lissencephaly with ambiguous genitalia, X-linked mental retardation), full gene sequence

BTD (biotinidase) (eg, biotinidase deficiency sequence gene full ,)

CAV3 (caveolin 3) (eg, CAV3-related distal myopathy, limb-girdle muscular dystrophy type 1C), full gene sequence

CDKN2A (cyclin-dependent kinase inhibitor 2A) (eg, CDKN2A-related cutaneous malignant melanoma, familial atypical mole-malignant melanoma syndrome), full gene sequence

CLRN1 (clarin 1) (eg, Usher syndrome, type 3), full gene sequence

CPT2 (carnitine palmitoyltransferase 2) (eg, carnitine palmitoyltransferase II deficiency), full gene sequence

CYP1B1 (cytochrome P450, family 1, subfamily B, polypeptide 1) (eg, primary congenital glaucoma), full gene sequence

DMPK (dystrophia myotonica-protein kinase) (eg, myotonic dystrophy type 1), characterization of abnormal (eg, expanded) alleles

EGR2 (early growth response 2) (eg, Charcot-Marie-Tooth), full gene sequence

FGFR2 (fibroblast growth factor receptor 2) (eg, craniosynostosis, Apert syndrome, Crouzon syndrome), targeted sequence analysis (eg, exons 8, 10)

FGFR3 (fibroblast growth factor receptor 3) (eg, achondroplasia, hypochondroplasia), targeted sequence analysis (eg, exons 8, 11, 12, 13)

FKRP (Fukutin related protein) (eg, congenital muscular dystrophy type 1C [MDC1C], limb-girdle muscular dystrophy [LGMD] type 2I), full gene sequence

FOXG1 (forkhead box G1) (eg, Rett syndrome), full gene sequence

FSHMD1A (facioscapulohumeral muscular dystrophy 1A) (eg, facioscapulohumeral muscular dystrophy), evaluation to detect abnormal (eg, deleted) alleles

FSHMD1A (facioscapulohumeral muscular dystrophy 1A) (eg, facioscapulohumeral muscular dystrophy), characterization of haplotype(s) (ie, chromosome 4A and 4B haplotypes)

FXN (frataxin) (eg, Friedreich ataxia), full gene sequence

HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia), duplication/deletion analysis

(For common deletion variants of alpha globin 1 and alpha globin 2 genes, use 81257

HBB (hemoglobin, beta, Beta-Globin) (eg, thalassemia), full gene Sequence

HNF1B (HNF1 homeobox B) (eg, maturity-onset diabetes of the young [MODY]), duplication/deletion analysis

HRAS (v-HA-ras Harvey rat sarcoma viral oncogene homolog) (eg, Costello syndrome), full gene sequence

KCNJ10 (potassium inwardly-rectifying channel, subfamily J, member 10) (eg, SeSAME syndrome, EAST syndrome, sensorineural hearing loss), full gene sequence

KIT (C-kit) (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (eg, GIST, acute myeloid leukemia, melanoma), targeted gene analysis (eg, exons 8, 11, 13, 17, 18)

LITAF (lipopolysaccharide-induced TNF factor) (eg, Charcot-Marie-Tooth), full gene sequence

MEFV (Mediterranean fever) (eg, familial Mediterranean fever), full gene Sequence

MEN1 (multiple endocrine neoplasia I) (eg, multiple endocrine neoplasia type 1, Wermer syndrome), duplication/deletion analysis

NRAS (neuroblastoma RAS viral oncogene homolog) (eg, colorectal carcinoma), exon 1 and exon 2 sequences

PDGFRA (platelet-derived growth factor receptor alph polypeptide) (eg, gastrointestinal stromal tumor), targeted sequence analysis (eg, exons 12, 18)

PDX1 (pancreatic and duodenal homeobox 1) (eg, maturity-onset diabetes of the young [MODY]), full gene sequence

PRNP (prion protein) (eg, genetic prion disease), full gene sequence

PRSS1 (protease, serine, 1 [trypsin 1]) (eg, hereditary pancreatitis), full gene sequence

RAF1 (v-raf-1 murine leukemia viral oncogene homolog 1) (eg, LEOPARD syndrome), targeted sequence analysis (eg, exons 7, 12, 14, 17)

RET (ret proto-oncogene) (eg, multiple endocrine neoplasia, type 2B and familial medullary thyroid carcinoma), common variants (eg, M918T, 2647_2648delinsTT, A883F)

SDHD (succinate dehydrogenase complex, subunit D, integral membrane protein) (eg, hereditary paraganglioma), full gene sequence

SLC25A4 (solute carrier family 25 [mitochondrial carrier; adenine nucleotide translocator], member 4) (eg, progressive external opthalmoplegia), full gene sequence

TP53 (tumor protein 53) (eg, tumor samples), targeted sequence analysis of 2-5 exons

TTR (transthyretin) (eg, familial transthyretin amloidosis), full gene sequence

TYR (tyrosinase [oculocutaneous albinism IA]) (eg, oculocutaneous albinism IA), full gene sequence

USH1G (Usher syndrome 1G [autosomal recessive]) (eg, Usher syndrome, type 1), full gene sequence

VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), full gene sequence

VWF (von Willebrand factor) (eg, von Willebrand disease type 1C), targeted sequence analysis (eg, exons 26, 27, 37)

81405 Molecular pathology procedure, Level 6 (eg, analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons)

ABCD1 (ATP-binding cassette, sub-family D [ALD], member 1) (eg, adrenoleukodystrophy), full gene sequence

ACADS (acyl-CoA deydrogenase, C-2 to C-3 short chain) (eg, short chain acyl-CoA deydrogenase deficiency), full gene sequence

ACTC1 (actin, alpha, cardiac muscle 1) (eg, familial hypertrophic cardiomyopathy), full gene sequence

APTX (aprataxin) (eg, ataxia with oculomotor apraxia 1), full gene sequence

AR (androgen receptor) (eg, androgen insensitivity syndrome), full gene sequence

CHRNB2 (cholinergic receptor, nicotinic, beta 2 [neuronal]) (eg, nocturnal frontal lobe epilepsy), full gene sequence

CHRNA4 (cholinergic receptor, nicotinic, alpha 4) (eg, nocturnal frontal lobe epilepsy), full gene sequence

CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide2) (eg, steroid 21-hydroxylase isoform, congenital adrenal hyperplasia), full gene sequence

DFNB59 (deafness, autosomal recessive 59) (eg, autosomal recessive nonsyndromic hearing impairment), full gene sequence

DHCR7 (7-dehydrocholesterol reductase) (eg, Smith-Lemli-Opitz syndrome), full gene sequence

EYA1 (eyes absent homolog 1 [Drosophila]) (eg, branchio-oto-renal [BOR] spectrum disorders), duplication/deletion analysis

F9 (coagulation factor IX) (eg, hemophilia B), full gene sequence

FH (fumarate hydratase) (eg, fumarate hydratase deficiency, hereditary leiomyomatosis with renal cell cancer), full gene sequence

FKTN (Fukutin) (eg, limb-girdle muscular dystrophy [LGMD] type 2M or 2L), full gene sequence

GFAP (glial fibrillary acidic protein) (eg, Alexander disease), full gene sequence

GLA (galactosidase, alpha) (eg, Fabry disease), full gene sequence

HBA1/HBA2 (alpha globin 1 and alph globin 2) (eg, thalassemia), full gene sequence

HNF1A (HNF1 homeobox A) (eg, maturity-onset diabetes of the young [MODY]), full gene sequence

HNF1B (HNF1 homeobox B) (eg, maturity-onset diabetes of the young [MODY]), full gene sequence

KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog) (eg, Noonan syndrome), full gene sequence

LAMP2 (lysosomal-associated membrane protein 2) (eg, Danon disease), full gene sequence

MEN1 (multiple endocrine neoplasia I) (eg, multiple endocrine neoplasia type 1, Wermer syndrome), full gene sequence

MPZ (myelin protein zero) (eg, Charcot-Marie-Tooth), full gene sequence

MYL2 (myosin, light chain 2, regulatory, cardiac, slow) (eg, familial hypertrophic cardiomyopathy), full gene sequence

MYL3 (myosin, light chain 3, alkali, ventricular, skeletal, slow) (eg, familial hypertrophic cardiomyopathy), full gene sequence

MYOT (myotilin) (eg, limb-girdle muscular dystrophy), full gene sequence

NEFL (neurofilament, light polypeptide) (eg, Charcot-Marie-Tooth), full gene sequence

NF2 (neurofibromin 2 [merlin]) (eg., neurofibromatosis, type 2), duplication/deletion analysis

NSD1 (nuclear receptor binding SET domain protein 1) (eg, Sotos syndrome), duplication/deletion analysis

OTC (ornithine carbamoyltransferase) (eg, ornithine transcarbamylase deficiency), full gene sequence

PDHB (pyruvate dehydrogenase [liboamide] beta) (eg, lactic acidosis), full gene sequence

PSEN1 (presenilin 1) (eg, Alzheimer disease), full gene sequence

RET (ret proto-oncogene) (eg, multiple endocrine neoplasia, type 2A and familial medullary thyroid carcinoma), targeted sequence analysis (eg, exons 10, 11, 13-16)

SDHB (succinate dehydrogenase complex, subunit B, iron sulfur) (eg, hereditary paraganglioma), full gene sequence

SDHC (succinate dehydrogenase complex, subunit C, integral membrane protein, 15kDa) (eg, hereditary paraganglioma-pheochromocytoma syndrome), full gene sequence

SGCA (sarcoglycan, alpha [50kDa dystrophin-associated glycoprotein]) (eg, limb-girdle muscular dystrophy), full gene sequence

SGCB (sarcoglycan, beta [43kDa dystrophin-associated glycoprotein]) (eg, limb-girdle muscular dystrophy), full gene sequence

SGCD (sarcoglycan, delta [35kDa dystrophin-associated glycoprotein]) (eg, limb-girdle muscular dystrophy), full gene sequence

SGCG (sarcoglycan, gamma [35kDa dystrophin-associated glycoprotein]) (eg, limb-girdle muscular dystrophy), full gene sequence

SHOC2 (soc-2 suppressor of clear homolog) (eg, Noonan-like syndrome with loose anagen hair), full gene sequence

SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy), full gene sequence

SPRED1 (sprout-related, EVH1 domain containing 1) (eg, Legius syndrome), full gene sequence

TGFBR1 (transforming growth factor, beta receptor 1) (eg, Marfan syndrome), full gene sequence

TGFBR2 (transforming growth factor, beta receptor 2) (eg, Marfan syndrome), full gene sequence

THRB (thyroid hormone receptor, beta) (eg, thyroid hormone resistance, thyroid hormone beta receptor deficiency), full gene sequence or targeted sequence analysis of >5 exons

TNNI3 (troponin I, type 3 [cardiac]) (eg, familial hypertrophic cardiomyopathy, full gene sequence

TP53 (tumor protein 53) (eg, Li-Fraumeni syndrome, tumor samples), full gene sequence or targeted sequence analysis of >5 exons

TPM1 (tropomyosin 1 [alpha]) (eg, familial hypertrophic cardiomyopathy), full gene sequence

TSC1 (tuberous sclerosis 1) (eg, tuberous sclerosis), duplication/deletion analysis

VWF (von Willebrand factor) (eg, von Willebrand disease type 2N),

targeted sequence analysis (eg, exons 18-20, 23-25)

81406 Molecular pathology procedure, Level 7 (eg, analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons, cytogenomic array analysis for neoplasia)

ACADVL (acyl-CoA dehydrogenase, very long chain) (eg, very long chain acyl-coenzyme A dehydrogenase deficiency), full gene sequence

ACTN4 (actinin, alpha 4) (eg, focal segmental glomerulosclerosis), full gene sequence

ANO5 (anoctamin 5) (eg, limb-girdle muscular dystrophy), full gene sequence

APP (amyloid beta [A4] precursor protein) (eg, Alzheimer disease), full gene sequence

ATP7B (ATPase, Cu++ transporting, beta polypeptide) (eg, Wilson disease), full gene sequence

BRAF (v-raf murine sarcoma viral oncogene homolog B1) (eg, Noonan syndrome), full gene sequence

CAPN3 (Calpain 3) (eg, limb-girdle muscular dystrophy [LGMD] type 2A, alpainopathy), full gene sequence

CBS (cystathionine-beta-synthase) (eg, homocystinuria, cystathionine beta-synthase deficiency), full gene sequence

CDH1 (cadherin 1, type 1, E-cadherin [epithelial]) (eg, hereditary diffuse gastric cancer), full gene sequence

CDKL5 (cyclin-dependent kinase-like 5) (eg, early infantile epileptic encephalopathy), full gene sequence

Cytogenomic microarray analysis, neoplasia (eg, interrogation of copy number, and loss-of-heterozygosity via single nucleotide polymorphism [SNP]-based comparative genomic hybridization [CGH] microarray analysis)

DLAT (dihydrolipoamide S-acetyltransferase) (eg, pyruvate dehydrogenase E2 deficiency), full gene sequence

DLD (dihydrolipoamide dehydrogenase) (eg, maple syrup urine disease, type III), full gene sequence

EYA1 (eyes absent homolog 1 [Drosophila]) (eg, branchio-oto-renal [BOR] spectrum disorders), full gene sequence

F8 (coagulation factor VIII) (eg, hemophilia A), duplication/deletion analysis

GAA (glucosidase, alpha; acid) (eg, glycogen storage disease type II [Pompe disease]), full gene sequence

GALT (galactose-1-phosphate uridylyltransferase (eg, galactosemia), full gene sequence

GCDH (glutaryl-CoA dehydrogenase) (eg, glutaricacidemia type 1), full gene sequence

GCK (glucokinase [hexokinase 4]) (eg, maturity-onset diabetes of the young [MODY]), full gene sequence

HADHA (hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase [trifunctional protein] alpha subunit) (eg, long chain acyl-coenzyme A dehydrogenase deficiency), full gene sequence

HEXA (hexosaminidase A, alpha polypeptide) (eg, Tay-Sachs disease), full gene sequence

HNF4A (hepatocyte nuclear factor 4, alpha) (eg, maturity-onset diabetes of the young [MODY]), full gene sequence

IVD (isovaleryl-CoA dehydrogenase) (eg, isovaleric acidemia), full gene sequence

JAG1 (jagged 1) (eg, Alagille syndrome), duplication/deletion analysis

LBD3 (LIM domain binding 3) (eg, familial dilated cardiomyopathy, myofibrillar myopathy), full gene sequence

LMNA (lamin A/C) (eg, Emery-Dreifuss muscular dystrophy [EDMD1, 2 and 3] limb-girdle muscular dystrophy [LGMD] type 1B, dilated cardiomyopathy [CMD1A], familial partial lipodystrophy [FPLD2]), full gene sequence
MAP2K1 (mitogen-activated protein kinase 1) (eg, cardiofaciocutaneous syndrome), full gene sequence

MAP2K2 (mitogen-activated protein kinase 2) (eg, cardiofaciocutaneous syndrome), full gene sequence

MCCC2 (methylcrotonoyl-CoA carboxylase 2 [beta]) (eg, 3-methylcrotonyl carboxylase deficiency), full gene sequence

MUTYH (mutY homolog [E. coli]) (eg, MYH-associated polyposis), full gene sequence

NF2 (neurofibromin 2 [merlin]) (eg, neurofibromatosis, type 2), full gene sequence

NOTCH3 (notch 3) (eg, cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy [CADASIL]), targeted sequence analysis (eg, exons 1-23)

NSD1 (nuclear receptor binding SET domain protein 1) (eg, Sotos syndrome), full gene sequence

OPA1 (optic atrophy 1) (eg, optic atrophy), duplication/deletion analysis

PAH (phenylalanine hydroxylase) (eg, phenylketonuria), full gene sequence

PALB2 (partner and localizer of BRCA2) (eg, breast and pancreatic cancer), full gene sequence

PAX2 (paired box 2) (eg, renal coloboma syndrome), full gene sequence

PC (pyruvate carboxylase) (eg, pyruvate carboxylase deficiency), full gene sequence

PCCB (propionyl CoA carboxylase, beta polypeptide) (eg, propionic acidemia), full gene sequence

PDHA1 (pyruvate dehydrogenase [lipoamide] alpha 1) (eg, lactic acidosis), full gene sequence

PDHX (pyruvate dehydrogenase complex, component X) (eg, lactic acidosis), full gene sequence

POLG (polymerase [DNA directed], gamma) (eg, Alpers-Huttenlocher syndrome, autosomal dominant progressive external opthalmoplegia), full gene sequence

POMGNT1 (protein O-linked mannose beta 1,2-N acetylglucosaminyltransferase) (eg, muscle-eye-brain disease, Walker-Warburg syndrome), full gene sequence

POMT1 (protein-O-mannosyltransferase 1) (eg, limb-girdle muscular dystrophy [LGMD] type 2K, Walker-Warburg syndrome), full gene sequence

POMT2 (protein-O-mannosyltransferase 2) (eg, limb-girdle muscular dystrophy [LGMD] type 2N, Walker-Warburg syndrome), full gene sequence

PRKAG2 (protein kinase, AMP-activated, gamma 2 non-catalytic subunit) (eg, familial hypertrophic cardiomyopathy with Wolff-Parkinson-White syndrome, lethal congenital glycogen storage disease of heart), full gene sequence

PSEN2 (presenilin 2 [Alzheimer disease 4]) (eg, Alzheimer disease), full gene sequence

PTPN11 (protein tyrosine phosphatase, non-receptor type 11) (eg, Noonan syndrome, LEOPARD syndrome), full gene sequence

PYGM (phosphorylase, glycogen, muscle) (eg, glycogen storage disease type V, McArdle disease), full gene sequence

RAF1 (v-raf-1 murine leukemia viral oncogene homolog 1) (eg, LEOPARD syndrome), full gene sequence

RET (ret proto-oncogene) (eg, Hirschsprung disease), full gene sequence

RYR1 (ryanodine receptor 1, skeletal) (eg, malignant hyperthermia), targeted sequence analysis of exons with functionally-confirmed mutations

SLC9A6 (solute carrier family 9 [sodium/hydrogen exchanger], member 6) (eg, Christianson syndrome), full gene sequence

SLC26A4 (solute carrier family 26, member 4) (eg, Pendred syndrome), full gene sequence

SOS1 (son of sevenless homolog 1) (eg, Noonan syndrome, gingival fibromatosis), full gene sequence

TAZ (tafazzin) (eg, methylglutaconic aciduria type 2, Barth syndrome), full gene sequence

TNNT2 (troponin T, type 2 [cardiac]) (eg, familial hypertrophic cardiomyopathy), full gene sequence

TSC1 (tuberous sclerosis 1) (eg, tuberous sclerosis), full gene sequence

TSC2 (tuberous sclerosis 2) (eg, tuberous sclerosis), duplication/deletion analysis

UBE3A (ubiquitin protein ligase E3A) (eg, Angelman syndrome), full gene sequence

VWF (von Willebrand factor) (von Willebrand disease type 2A), extended targeted sequence analysis (eg, exons 11-16, 24-26, 51, 52)

81407 Molecular pathology procedure, Level 8 (eg, analysis of 26-50 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of >50 exons, sequence analysis of multiple genes on one platform)

ABCC8 (ATP-binding cassette, sub-family C [CFTR/MRP], member 8) (eg, familial hyperinsulinism), full gene sequence

CHD7 (chromodomain helicase DNA binding protein 7) (eg, CHARGE syndrome), full gene sequence

F8 (coagulation factor VIII) (eg, hemophilia A), full gene sequence

JAG1 (jagged 1) (eg, Alagille syndrome), full gene sequence

MYBPC3 (myosin binding protein C, cardiac) (eg, familial hypertrophic cardiomyopathy), full gene sequence

MYH6 (myosin, heavy chain 6, cardiac muscle, alpha) (eg, familial dilated cardiomyopathy), full gene sequence

MYH7 (myosin, heavy chain 7, cardiac muscle, beta) (eg, familial hypertrophic cardiomyopathy, Liang distal myopathy), full gene sequence

MY07A (myosin VIIA) (eg, Usher syndrome, type 1), full gene sequence

NOTCH1 (notch 1) (eg, aortic valve disease), full gene sequence

OPA1 (optic atrophy 1) (eg, optic atrophy), full gene sequence

PCDH15 (protocadherin-related 15) (eg, Usher syndrome, type 1), full gene sequence

SCN1A (sodium channel, voltage-gated, type 1, alpha subunit) (eg, generalized epilepsy with febrile seizures), full gene sequence

SCN5A (sodium channel, voltage-gated, type V, alpha subunit) (eg, familial dilated cardiomyopathy), full gene sequence

TSC2 (tuberous sclerosis 2) (eg, tuberous sclerosis), full gene sequence

USH1C (Usher syndrome 1C [autosomal recessive, severe]) (eg, Usher syndrome, type 1), full gene sequence

81408 Molecular pathology procedure, Level 9 (eg, analysis of >50 exons in a single gene by DNA sequence analysis)

ATM (ataxia telangiectasia mutated) (eg, ataxia telangiectasia), full gene sequence

COL1A1 (collagen, type I, alpha 1) (eg, osteogenesis imperfect, type I), full gene sequence

COL1A2 (collagen, type I, alpha 2) (eg, osteogenesis imprefecta, type I), full gene sequence

CDH23 (cadherin-related 23) (eg, Usher syndrome, type 1), full gene sequence

DYSF (dysferlin, limb girdle muscular dystrophy 2B [autosomal recessive]) (eg, limb-girdle muscular dystrophy), full gene sequence

FBN1 (fibrillin 1) (eg, Marfan syndrome), full gene sequence

NF1 (neurofibromin 1) (eg, neurofibromatosis, type 1), full gene sequence

RYR1 (ryanodine receptor 1, skeletal) (eg, malignant hyperthermia), full gene sequence

USH2A (Usher syndrome 2A [autosomal recessive, mild]) (eg, Usher syndrome, type 2), full gene sequence

VWF (von Willebrand factor) (eg, von Willebrand disease types 1 and 3), full gene sequence

Multianalyte Assays with Algorithmic Analyses (MAAA)

815XX Oncology (ovarian), biochemical assays of two proteins (CA-125 and HE4), utilizing serum, with menopausal status, algorithm reported as a risk score

815XX Oncology (ovarian), biochemical assays of five proteins (CA-125, apoliproprotein A1, beta-2 microglobulin, transferrin and pre-albumin), utilizing serum, algorithm reported as a risk score

815XX Endocrinology (type 2 diabetes), biochemical assays of seven analytes (glucose, HbA1c, insulin, hs-CRP, adoponectin, ferritin, interleukin 2-receptor alpha), utilizing serum or plasma, algorithm reporting a risk score

815XX Fetal chromosomal abnormalities, biochemical assays of three proteins (PAPP-A, hCG (any form), DIA), utilizing maternal serum, algorithm reported as a risk score

815XX Fetal chromosomal abnormalities, biochemical assays of three analytes (AFP, uE3, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score

815XX Fetal chromosomal abnormalities, biochemical assays of four analytes (AFP, uE3, hCG [any form], DIA) utilizing maternal serum, algorithm reported as a risk score (may include additional results from previous biochemical testing)

815XX Fetal chromosomal abnormalities, biochemical assays of five analytes (AFP, uE3, total hCG, hyperglycosylated hCG, DIA) utilizing maternal serum, algorithm reported as a risk score

XXXX1M Infectious disease, HCV, six biochemical assays (ALT, A2-macroglobulin, apolipoprotein A-1, total bilirubin, GGT, and haptoglobin) utilizing serum, prognostic algorithm reported as scores for fibrosis and necroinflammatory activity in liver

XXXX2M Liver disease, ten biochemical assays (ALT, A2-macroglobulin, apolipoprotein A-1, total bilirubin, GGT, haptoglobin, AST, glucose, total cholesterol and triglycerides) utilizing serum, prognostic algorithm reported as quantitative scores for fibrosis, steatosis and alcoholic steatohepatitis (ASH)

XXXX3M Liver disease, ten biochemical assays (ALT, A2-macroglobulin, apolipoprotein A-1, total bilirubin, GGT, haptoglobin, AST, glucose, total cholesterol and triglycerides) utilizing serum, prognostic algorithm reported as quantitative scores for fibrosis, steatosis and nonalcoholic steatohepatitis (NASH)

Chemistry

827XX Galectin-3

Immunology

861XX Cell enumeration using immunologic selection and identification in fluid specimen (eg, circulating tumor cells in blood);

867XX JC (John Cunningham) virus

Tissue Typing

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); qualitative assessment of the presence or absence of antibody(ies) to HLA Class I and Class II HLA antigens

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); qualitative assessment of the presence or absence of antibody(ies) to HLA Class I or Class II HLA antigens

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); antibody identification by qualitative panel using complete HLA phenotypes, HLA Class I

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); antibody identification by qualitative panel using complete HLA phenotypes, HLA Class II

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); high definition qualitative panel for identification of antibody specificities (eg, individual antigen per bead methodology), HLA Class I

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); high definition qualitative panel for identification of antibody specificities (eg, individual antigen per bead methodology), HLA Class II

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); semi-quantitative panel (eg, titer), HLA Class I

868XX Antibody to human leukocyte antigens (HLA), solid phase assays (eg, microspheres or beads, ELISA, flow cytometry); semi-quantitative panel (eg, titer), HLA Class II

Microbiology

876XX *Infectious agent detection by nucleic acid (DNA or RNA); Bartonella henselae and Bartonella quintana, direct probe technique*; respiratory virus (eg, adenovirus, influenza virus, coronavirus, metapneumovirus, parainfluenza virus, respiratory syncytial virus, rhinovirus), multiplex reverse transcription and amplified probe technique, multiple types or subtypes, 3-5 targets

876XX *Infectious agent detection by nucleic acid (DNA or RNA); Bartonella henselae and Bartonella quintana, direct probe technique*; respiratory virus (eg, adenovirus, influenza virus, coronavirus, metapneumovirus, parainfluenza virus, respiratory syncytial virus, rhinovirus), multiplex reverse transcription and amplified probe technique, multiple types or subtypes, 6-11 targets

876XX *Infectious agent detection by nucleic acid (DNA or RNA); Bartonella henselae and Bartonella quintana, direct probe technique*; respiratory virus (eg, adenovirus, influenza virus, coronavirus, metapneumovirus, parainfluenza virus, respiratory syncytial virus, rhinovirus), multiplex reverse transcription and amplified probe technique, multiple types or subtypes, 12-25 targets

879XX Infectious agent genotype analysis by nucleic acid (DNA or RNA); cytomegalovirus

879XX Infectious agent genotype analysis by nucleic acid (DNA or RNA); Hepatitis B virus

Reconsideration Requests

86386 Nuclear Matrix Protein 22 (NMP22), qualitative