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Pathology & Laboratory
2021 CPT Updates
Effective 1/1/2021

Each year the CPT code manual is revised to delete codes and/or guidelines, and to add or revise codes to reflect current technologies, techniques, and services. As a service to our clients, APS Medical Billing has summarized those changes to facilitate accurate reporting of the affected services as of January 1, 2021. The American Medical Association has released that there will be 329 code changes in the new CPT edition to capture and describe the latest scientific and technological advances in medical, surgical and diagnostic services.

Below we have also outlined the changes made to these sections. All new codes are highlighted in red and revised/deleted codes are noted:

Therapeutic Drug Assays

New Codes for 2021:

80143 Acetaminophen
80151 Amiodarone
80161 Carbamazepine; 10, 11-epoxide
80167 Felbamate
80179 Salicytate
80181 Flecainide
80189 Itraconazole
80193 Leflumominde
80204 Methortrexate
80210 Rufinamide

Tier I Molecular Pathology Procedures

New Codes for 2021:

81168 CCND1/IGH (t{11;14}) (eg, mantle cell lymphoma) translocation analysis, major breakpoint, qualitative and quantitative, if performed
81191 NTRK1 (neurotrophic receptor tyrosine kinase 1) (eg, solid tumors) translocation analysis
81192 NTRK2 (neurotrophic receptor tyrosine kinase 2) (eg, solid tumors) translocation analysis
81193 NTRK3 (neurotrophic receptor tyrosine kinase 3) (eg, solid tumors) translocation analysis
81194 NTRK (NEUROTROPHIC-TROPOMYOSIN RECEPTOR TYROSINE KINASE 1, 2, and 3) (eg, solid tumors) translocation analysis

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81278 IHG@/BCL2 (t(14;18))) (eg, follicular lymphoma) translocation analysis, major breakpoint region (MBR) and minor cluster region (mcr) breakpoints, qualitative or quantitative

81279 JAK2 (Janus kinase 2) (eg, myeloproliferative disorder) targeted sequence analysis (eg. Exons 12 and 13)

81338 MPL (MPL proto-oncogene, thrombopoietin receptor) (eg myeloproliferative disorder) gene analysis; common variants (eg, W515A, W515K, W515L, W515R)

81339 MPL (MPL proto-oncogene, thrombopoietin receptor) (eg myeloproliferative disorder) gene analysis; sequence analysis, exon 10

81347 SF3B1 (splicing factor [3b] subunit B1) (eg. Myelodysplastic syndrome/acute myeloid leukemia) gene analysis, common variants (eg, A672T, E622D, L833F, R625C, R625L)

81348 SRSF2 (serine and arginine-rich splicing factor 2) (eg, myelodysplastic syndrome, acute myeloid leukemia) gene analysis, common variants (eg, P95H, P95L)

81351 TP53 (tumor protein 53) (eg, Li-Fraumeni syndrome) gene analysis; full gene sequence

81352 TP53 (tumor protein 53) (eg, Li-Fraumeni syndrome) gene analysis; targeted sequence analysis (eg, 4 oncology)

81353 TP53 (tumor protein 53) (eg, Li-Fraumeni syndrome) gene analysis; unknown familial variant

81357 U2AF1 (U2 small nuclear RNA auxiliary factor 1) (eg, myelodysplastic syndrome, acute myeloid leukemia) gene analysis, common variants (eg, S34F, S34Y, Q157R, Q157P)

81360 ZRSR2 (zinc finger CCCH-type, RNA binding motif and serine/arginine-rich 2) (eg, myelodysplastic syndrome, acute myeloid leukemia) gene analysis, common variant(s) (eg, E65fs, E12fs, R448fs)

81419 Epilepsy genomic sequence analysis panel, must include analyses for ALDH7A1, CACNA1A, CHKL5, CHD2, GABRG2, KCNQ2, MECP2, PCDH19, POLG, PRRT2, SCN1A, SCN1B, SCN2A, SCN8A, SLC2A1, SLC9A6, STXBP1, SYNGAP1, TCF4, TPP1, TSC1, TSC2, and ZEB2

81513 Infectious disease, bacterial vaginosis, quantitative real-time amplification of RNA markers for Atopobium vaginae, Gardnerella vaginalis, and Lactobacillus species, utilizing vaginal-fluid specimens, algorithm reported as a positive or negative result for bacterial vaginosis

81514 Infectious disease, bacterial vaginosis and vaginitis, quantitative real-time amplification of DNA markers for Gardnerella vaginalis, Atopobium vaginae, Megasphaera type 1, Bacterial Vaginosis Associated Bacteria-2 (BVAB-2), and Lactobacilus species (L. crispatus and L. jensenii), utilizing vaginal-fluid specimens, algorithm reported as a positive or negative for high likelihood of bacterial vaginosis, includes separate detection of Trichomonas vaginalis and/or Candida species (C. albicans, C tropicalis, C. parapsilosis, C dubliniensis), Candida glabrata, Candida krusei, when reported

81529 Oncology (cutaneous melanoma), mRNA, gene expression profiling by real-time RT-PCR of 31 genes (28 content and 3 housekeeping), utilizing formalin-fixed paraffin-embedded tissue, algorithm reported as recurrence risk, including likelihood of sentinel lymph node metastasis

81546 Oncology (thyroid), mRNA, gene expression analysis of 10,196 genes, utilizing fine needle aspirate, algorithm reported as categorical result (eg, benign or suspicious)

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])

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Revised Code for 2021:

80415 Chorionic gonadotropin stimulation panel; estradiol response this panel must include the following, Estradiol, **total** (82670 x 2 or 3 pooled blood samples)

Tier II Molecular Pathology Procedures

Revised Codes for 2021:

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 dynamic mutation disorder/triplet repeat) ABCC8 (ATP binding cassette, sub-family C [CFTR/MRP], member 8) (eg, familial hyperinsulinism), common variants (eg, c.3898-9G>A], [c3992-9G>A], F1388del) ABL1 (ABL proto-oncogene 1, non-receptor tyrosine kinase) (eg, acquired imatinib resistance), T3151 variant ACADM (acyl-CoA dehydrogenase, C-4 to C-12 straight chain, MCAD) (e., 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) 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 (BCL/IgH, 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]) DEK/NUP214 (t(6;9)) (eg, acute myeloid leukemia), translocation analysis, qualitative, and quantitative, if performed E2A/PBX (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/NTRK3 (t(12;15)) (eg, congenital/infantile fibrosarcoma), translocation analysis, qualitative, and quantitative, if performed~~ ETV6/RUNX1 (t(12;21)) (eg, acute lymphocytic leukemia), translocation analysis qualitative, and quantitative, if performed EWSR1/ATF1 (t(12;22)) (eg, clear cell sarcoma), 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 EWSR//FLI1 (t911;22)) (eg, Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative, and quantitative, if performed EWSR1/WT1 (t(11;22) (eg, Desmoplastic small round cell 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 EVS14+iG>A[Type 1] FGFR3(fibroblast growth factor receptor 3) (eg, achondroplasia, hypochondroplasia, common variants (eg. 1138G>A, 1620C>A, 1620C>G FIP1L1/PDGFRA 9del[4q12]) (eg, imatinib-sensitive chronic eosinophilic leukemia), qualitative, and quantitative, if performed FLG (filaggrin) (eg, ichthyosis vulgaris), common variants (eg. R501X, 2282del4, R2247x, S3247X, 3702delG) FOXO1/PAX3 (t(2;13)) (eg, alveolar rhabdomyosarcoma), translocation analysis, qualitative, and quantitative, if performed FOXO1/PAX3 (t(1:13)) (eg, alveolar

rhabdomyosarcoma), translocation analysis, qualitative, and quantitative, if performed FUS/DDIT3 (t(12;16)) (eg, myxoid liposarcoma), translocation analysis, qualitative , and quantitative, if performed GALC (galactosylceramidase) (eg, Krabbe disease), common variants (eg, c.857G>A, 30-kb deletion) GALT (galactose-1 phosphate uridylytransferase) eg, galactosemia), common variants (eg, Q188R, S135L, K285N, T138M, L195P, Y209C, IVS2-2A>G, P1716, del5kb, N314D, L21863/N314D) H19 (imprinted maternally expressed transcript [non-protein coding]) (eg, Beckwith-Wiedemann syndrome), methylation analysis IGH@BCL2 (t(14;18)) (eg, follicular lymphoma), translocation analysis; single breakpoint (eg, major breakpoint region[MBR] or minor cluster region [mcr]), qualitative or quantitative (When both MBR and mcr breakpoints are performed, use 81278) KCNQ1OT1 (KCNQ1 overlapping transcript 1 [non-protein coding])) (eg, Beckwith-Wiedemann syndrome), methylation analysis LINC00518(long intergenic non-protein coding RNA 518) (eg, melanoma), expression analysis LRRK2 (leucine-rich repeat kinase 2) (eg, Parkinson disease), common variants (eg, R1441G, G2019S, I2020T) MED12 (mediator complex subunit 12) (eg, FG syndrome type 1, Lujan syndrome), common variants (eg, R961W, N1007S) MEG3DLK1 (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 MML/MLLT3 (t(9;11)) (eg, acute myeloid leukemia), translocation analysis, qualitative, and quantitative, if performed 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 dehydrogenase6) (eg, Leber hereditary optic neuropathy[LHON]) common variants (eg, m.11778G>A, m.3460G>A) MT-RNR1 (mitochondrially encoded 12S RNA) (eg, nonsyndromic hearing loss), common variants (eg, m.155A>G, m.1494C>T) MT-TK (mitochondrially encoded tRNA lysine) (eg, myoclonic epilepsy with ragged-red fibers [MERRF]) common variants (eg, 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.14709T>C) MT-TL1 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) MUTYH (muTy homolog [E. coli]) (eg, MYH-associated polyposis), common variants (eg, Y165C, G382D) NOD2 (nucleotide-binding oligomerization domain containing 2) (eg, Crohn's disease, Blau syndrome), common variants (eg, SNP 8, SNP 12, SNP 13) NPM1/ALK (t(2;5)) (eg, anaplastic large cell lymphoma), translocation analysis PAX8/PPARG (t(2;3)) (q.13:p25)) (eg, follicular thyroid carcinoma), translocation analysis PRAME (preferentially expressed antigen in melanoma) (eg. melanoma), expression 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 SS18/SSX1 (t(X;18)) (eg, synovial sarcoma), translocation analysis, qualitative, and quantitative, if performed SS18/SSX2 (t(X;18)) (eg, synovial sarcoma), translocation analysis, qualitative, and quantitative, if performed 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 1p-/19q- (eg, glial tumors), deletion analysis Chromosome 18q- (eg, D18S55, D18S58, D18S61, D18S64, and D18S69) (eg, colon cancer), allelic imbalance assessment (ie, loss of heterozygosity) COL1A1/PDGFB (t(17;22)) (eg, dermatofibrosarcoma protuberans), translocation analysis, multiple breakpoints, qualitative, and quantitative, if performed 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**)** ~~IGH@BCL2 (t(14;18)) (eg, follicular lymphoma), translocation analysis; major breakpoint region (MBR) and minor cluster region (mcr) breakpoints, qualitative or quantitative~~ MEFV (Mediterranean fever) (eg, familial Mediterranean fever), common variants (eg, E148Q, P369S, F479L, M680I, I692del, M694V, M694I, K695R, V726A, A744S, R761H) 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) ANG (angiogenin, ribonuclease, RNase A family, 5) (eg, amyotrophic lateral sclerosis), full gene sequence ARX (aristaless-related homeobox) (eg, X-linked lissencephaly with ambiguous genitalia, X-linked mental retardation), duplication/deletion analysis CEL (carboxyl ester lipase [bile salt-stimulated lipase]) (eg, maturity-onset diabetes of the young [MODY]), targeted sequence analysis of exon 11 (eg, c.1785delC, c.1686delT) CTNNB1 (catenin [cadherin-associated protein], beta 1, 88kDa) (eg, desmoid tumors), targeted sequence analysis (eg, exon 3) DAZ/SRY (deleted in azoospermia and sex determining region Y) (eg, male infertility), common deletions (eg, AZFa, AZFb, AZFc, AZFd) DNMT3A (DNA [cytosine-5-]-methyltransferase 3 alpha) (eg, acute myeloid leukemia), targeted sequence analysis (eg, exon 23) EPCAM (epithelial cell adhesion molecule) (eg, Lynch syndrome), duplication/deletion analysis F8 (coagulation factor VIII) (eg, hemophilia A), inversion analysis, intron 1 and intron 22A F12 (coagulation factor XII [Hageman factor]) (eg, angioedema, hereditary, type III; factor XII deficiency), targeted sequence analysis of exon 9 FGFR3 (fibroblast growth factor receptor 3) (eg, isolated craniosynostosis), targeted sequence analysis (eg, exon 7) (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 GNAQ (guanine nucleotide-binding protein G[q] subunit alpha) (eg, uveal melanoma), common variants (eg, R183, Q209) Human erythrocyte antigen gene analyses (eg, SLC14A1 [Kidd blood group], BCAM [Lutheran blood group], ICAM4 [Landsteiner-Wiener blood group], SLC4A1 [Diego blood group], AQP1 [Colton blood group], ERMAP [Scianna blood group], RHCE [Rh blood group, CcEe antigens}, KEL [Kell blood group] DARC [Duffy blood group] GYPA, GYPB, GYPE [MNS blood group, ART4[Dombrock blood group]) (eg, sickle-cell disease, thalassemia, hemolytic transfusion reactions, hemolytic disease of the fetus or newborn), common variants HRAS (v-Ha-ras-Harvey rat sarcoma viral oncogene homolog) (eg,

Costello syndrome), exon 2 sequence ~~JAK2 (Janus kinase 2) (eg, myeloproliferative disorder), exon 12 sequence and exon 13 sequence, if performed~~ KCNC3 (potassium voltage-gated channel, Shaw-related subfamily, member3) (eg, spinocerebellar ataxia), targeted sequence analysis (eg, exon 2) KNCJ2 (potassium inwardly-rectifying channel, subfamily J, member 2) (eg, Andersen-Tawil syndrome), full gene sequence KCNJ11 (potassium inwardly-rectifying cannel, subfamily J, member 11) (eg, familial hyperinsulinism), full gene sequence Killer cell immunoglobulin-like receptor (KIR) gene family (eg, hematopoietic stem cell transplantation), genotyping of KIR family genes Known familial variant not otherwise specified, for gene listed in Tier 1 or Tier 2, or identified during a genomic sequencing procedure, DNA sequence analysis, each variant exon (For known familial variant that is considered a common variant, use specific common variant Tier 1 or tier 2 code) MC4R (melanocortin 4 receptor) (eg, obesity), full gene sequence MICA (MHC class 1 polypeptide-related sequence A) eg, solid organ transplantation), common variants (eg, *001, *002) ~~MPL (myeloproliferative leukemia virus oncogene, thromboprotein 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 NDP (Norrie disease [pseudoglioma]) (eg, Norrie disease duplication/deletion analysis NHLRC1 (NHL repeat containing 1) (eg, progressive myoclonus epilepsy), full gene sequence PHOX2B (paired-like homeobox 2b) (eg, congenital central hypoventilation syndrome), duplication/deletion analysis PLN (phospholamban) (eg, dilated cardiomyopathy, hypertrophic cardiomyopathy), full gene sequence RHD (Rh blood group, D antigen) (eg, hemolytic disease of the fetus and newborn, Rh maternal/fetal compatibility), deletion analysis (eg, exons 4, 5, and 7, pseudogene) RHD (Rh blood group, D antigen) (eg, hemolytic disease of the fetus and newborn, Rh maternal/fetal compatibility), deletion analysis (eg, exons 4, 5, and 7, pseudogene) performed on cell-free fetal DNA in maternal blood (For human erythrocyte gene analysis of RHD, use a separate unit of 81403) SH2D1A (SH2 domain containing 1A(eg, X-linked lymphoproliferative syndrome), duplication/deletion analysis TWIST1 (twist homolog 1 [Drosophila]) (eg, Saethre-Chotzen syndrome), duplication/deletion analysis UBA1 (ubiquitin-like modifier activating enzyme 1) eg, spinal muscular atrophy, X-linked, targeted sequence analysis (eg, exon 15) VHL (con Hippel-Lindau tumor suppressor) (eg, con 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 AVPR2 (arginine vasopressin receptor 2) (eg, nephrogenic diabetes insipidus), full gene sequence BBS10 (Bardet-Biedl syndrome 10) (eg, Bardet-Biedl syndrome), full gene sequence BTD (biotinidase) (eg, biotinidase deficiency), full gene sequence C10orf2 (chromosome 10 open reading frame 2) (eg, mitochondrial DNA depletion syndrome), full gene sequence CAV3 (caveolin 3) (eg, CAV3-related distal myopathy, limb-girdle muscular dystrophy type 1C), full gene sequence CD40LG (CD40 ligand) (eg, X-linked hyper IgM syndrome), 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 COX6B1 (cytochrome c oxidase subunit VIb polypeptide 1) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence CPT2 (carnitine palmitoyltransferase 2) (eg, carnitine palmitoyltransferase II deficiency), full gene sequence CRX (cone-rod homeobox) (eg, cone-rod dystrophy 2, Leber congenital amaurosis), full gene sequence CYP1B1 (cytochrome P450, family 1, subfamily B, polypeptide 1) (eg, primary congenital glaucoma), full gene sequence EGR2 (early growth response 2) (eg, Charcot-Marie-Tooth), full gene sequence EMD (emerin) (eg, Emery-Dreifuss muscular dystrophy), duplication/deletion analysis EPM2A (epilepsy, progressive myoclonus epilepsy), full gene sequence FGF23 (fibroblast growth factor 23) (eg, hypophosphatemic rickets), full gene sequence FGFR2 (fibroblast growth factor receptor 2) (eg, craniosynostosis, Apert syndrome, Crouzon syndrome). Targeted sequence analysis (eg, exon 8, 10) FGFR3 (fibroblast growth factor receptor 3 (eg, achondroplasia, hypochondroplasia), targeted sequence analysis (eg, exons 8,11,12,13) FHL (four and a half LIM domains 1) (eg, Emery-Dreifuss muscular dystrophy), full gene sequence FKRP (fukutin related protein) (eg, congenital muscular dystrophy type 1C [MDC1C], limb-girdle muscular dystrophy [LGMD] type 21), 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) GH1 (growth hormone 1) (eg, growth hormone deficiency) full gene sequence) GP1BB (glycoprotein 1b [platelet], beta polypeptide) (Bernard-Soulier syndrome type B), full gene sequence (For common deletion variants of alpha globin 1 and alpha globin 2 genes, use 81257) 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 HSD3B2 (hydroxyl-delta-isomerase 2) (eg, 3 beta-hydroxysteroid dehydrogenase type II deficiency), full gene sequence HSD11B2 (hydroxysteroid [11-beta] dehydrogenase 2) (eg, mineralocorticoid excess syndrome), full gene sequence HSPB1 (heat shock 27kDa protein 1) (eg, Charcot-Marie-Tooth disease), full gene sequence INS (insulin) (eg, diabetes mellitus), full gene sequence KCNJ1 (potassium inwardly-rectifying channel subfamily J, member 1) (eg, Bartter syndrome). Full gene sequence KCNJ10 (potassium inwardly-rectifying channel, subfamily J, member 10), (eg, SeSAME syndrome, EAST syndrome, sensorineural hearing loss), full gene sequence 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 1) (eg, multiple endocrine neoplasia type 1, Wermer syndrome) MMACHC (methylmalonic aciduria [cobalamin deficiency] cb1C type, with homocystinuria) (eg, methylmalonic academia and homocystinuria), full gene sequence MPV17(MpV17 mitochondrial inner membrane protein) (eg, mitochondrial DNA depletion syndrome), duplication/deletion analysis NDP (Norrie disease [pseudoglioma]) (eg, Norrie disease), full gene sequence NDUFA1 (FADH dehydrogenase [ubiquinone] 41 alpha subcomplex, 1, 7.5kDa) eg, Leigh syndrome, mitochondrial complex 1 deficiency), full gene sequence NDUFAF2 (NADH dehydrogenase [ubiquinone] 1 alpha subcomplex, assembly factor 2) (eg, Leigh syndrome mitochondrial complex 1 deficiency), full gene sequence NDUFS4 (NADH dehydrogenase [ubiquinone] Fe-S protein4, 18kDa [NADH-coenzyme Q reductase] (eg, Leigh syndrome, mitochondrial complex 1

deficiency). full gene sequence NIPA1 (non-imprinted in Prader-Willi/Angelman syndrome 1) (eg, spastic paraplegia), full gene sequence NLGN4X (neuroligin 4, X-linked) (eg, autism spectrum disorders), duplication/deletion analysis NPC2 (Niemann-Pick disease, type C2 [epididymal secretory protein E1]) (eg, Niemann-Pick disease type C2), full gene sequence NROB1 (nuclear receptor subfamily O, group Bmember1 (eg, congenital adrenal hypoplasia), full gene sequence PDX1 (pancreatic and duodenal homeobox 1) (eg, maturity-onset diabetes of the young [MODY], full gene sequence PHOX2B (paired-like homeobox 2b) (eg, congenital central hypoventilation syndrome), full gene sequence PLP1 (proteolipid protein 1) (eg, Pelizaeus-Merzbacher disease, spastic paraplegia), duplication/deletion analysis PQBP1 (polyglutamine binding protein1) (eg, Renpenning syndrome), duplication/deletion analysis PRNP (prion protein) (eg, genetic prion disease), full gene sequence PROP1 (PROP paired-like homeobox 1) (eg, combined pituitary hormone deficiency), full gene sequence PRPH2 (perpipherin 2 [retinal degeneration, slow]) (eg, retinitis pigmentosa), full gene sequence PRSS1 (protease, serine, 1 [trypsin1]) 9eg, 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) RHO (rhodopsin) (eg, retinitis pigmentosa), full gene sequence RP1 (retinitis pigmentosa 1) (eg, retinitis pigmentosa), full gene sequence SCN1B (sodium channel, voltage-gated type 1, beta) (eg, Brugada syndrome), full gene sequence SCO2 (SCO cytochrome oxidase deficient homolog 2 [SCO1L]) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence SDHC (succinate dehydrogenase complex, subunit C, integral membrane protein, 15kDa) (eg, hereditary paraganglioma-pheochromocytoma syndrome), duplication/deletion analysis SDHD (succinate dehydrogenase complex, subunit D, integral membrane protein) (eg, hereditary paraganglioma), full gene SGDG(sarcoglycan, gamma [25kDa dystrophin-associated glycoprotein]) (eg, limb-girdle muscular dystrophy), duplication/deletion analysis SH2D1A (SH2domain containing 1A) (eg, X-linked lymphoproliferative syndrome), full gene sequence SLC16A2 (solute carrier family 16, member 2[thyroid hormone transporter]) (eg, specific thyroid hormone cell transporter deficiency, Allan-Herndon-Dudley syndrome), duplication/deletion analysis SLC25A20 9solute carrier family 25[carnitine/acylcarnitine translocase], memver20) (eg, carnitine-acylcarnitine translocase deficiency), duplication deletion analysis SLC25A4 (solute carrier family 25[mitochondrial carrier; adenine nucleotide translocator), member 4) (eg, progressive external ophthalmoplegia), full gene sequence SOD1 (superoxide dismutase 1, soluble), (eg, amyotrophic lateral sclerosis), full gene sequence SPINK1 (serine peptidase inhibitor Kazal type 1) (eg, hereditary pancreatitis), full gene sequence STK11 (serine/threonine kinase 11) (eg, Peutz-0Jeghers syndrome), duplication/deletion analysis TACO1 (translational activator of mitochondrial encoded cytochrome c oxidase 1) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence THAP1 (THAP domain containing, apoptosis associated protein 1) (eg, torsion dystonia), full gene sequenceTOR1A (torsin family 1, member a [torsin A]) (eg, torsion dystonia), full gene sequence ~~TP53 (tumor protein 56) (eg, targeted sequence analysis of 2-5 exons~~ TOR1A (torsin family 1, member A [torsion A](eg, torsion dystonia), full gene sequence TTPA (tocopherol [alpha] transfer protein) (eg, ataxia) full gene sequence TTR (transthyretin) (eg, familial transthyretin amyloidosis), full gene sequence TWIST1 (twist homolog 1 [Drosophila]) (eg, Saethre-Chotzen syndrome), full gene sequence TYR (tyrosinase [oculocutaneous albinism 1A]) (eg, oculocutaneous albinism 1A), full gene sequence UGT1A1 (UDP glucuronosyltransferase 1 family,

polypeptide A1) (eg, hereditary unconjugated hyperbilirubinemia [Crigler-Najjar syndrome]) full gene sequence USH1G (Usher syndrome 1G [autosomal recessive]) (eg, Usher syndrome, type1), gull gene sequence VHL (von Willebrand factor) (eg, von Willebrand disease type 1C), targeted sequence analysis (eg, exons 26, 27, 37) ZEB2 (zinc finger E-box binding homeobox 2) (eg, Mowat-Wilson syndrome), duplication/deletion analysis ZNF41 (zinc finger protein 41) (eg, X-linked mental retardation 89), full gene sequence

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, regionally targeted cytogenomic array analysis) ABCD1 (ATP-binding cassette, sub-family D [ALD], member 1) (eg, adrenoleukodystrophy), full gene sequence ACADS (acyl-CoA dehydrogenase, C-2 to C-3 short chain) (eg, short chain acyl-CoA dehydrogenase deficiency), full gene sequence ACTA2 (actin, alpha 2, smooth muscle, aorta) (eg, thoracic aortic aneurysms and aortic dissections), full gene sequence ACTC1 (actin, alpha, cardiac muscle 1) (eg, familial hypertrophic cardiomyopathy), full gene sequence ANKRD1 (ankyrin repeat domain 1) (eg, dilated cardiomyopathy), full gene sequence APTX (aprataxin) (eg, ataxia with oculomotor apraxia 1), full gene sequence ARSA (arylsulfatase A) (eg, arylsulfatase A deficiency), full gene sequence BCKDHA (branched chain keto acid dehydrogenase E1, alpha polypeptide) (eg, maple syrup urine disease, type 1A), full gene sequence BCS1L (BCS1-like [S. cerevisiae]) (eg, Leigh syndrome, mitochondrial complex III deficiency, GRACILE syndrome), full gene sequence BMPR2 (bone morphogenetic protein receptor, type II [serine/threonine kinase]) (eg, heritable pulmonary arterial hypertension), duplication/deletion analysis CASQ2 (calsequestrin 2 [cardiac muscle]) (eg, catecholaminergic polymorphic ventricular tachycardia), full gene sequence CASR (calcium-sensing receptor) (eg, hypocalcemia), full gene sequence CDKL5 (cyclin-dependent kinase-like 5) (eg, early infantile epileptic encephalopathy), duplication/deletion analysis CHRNA4 (cholinergic receptor, nicotinic, alpha 4) (eg, nocturnal frontal lobe epilepsy), full gene sequence CHRNB2 (cholinergic receptor, nicotinic, beta 2 [neuronal]) (eg, nocturnal frontal lobe epilepsy), full gene sequence COX10 (COX10 homolog, cytochrome c oxidase assembly protein) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence COX15 (COX15homolog, cytochrome c oxidase assembly protein) (eg, mitochondrial respiratory chain complex, IV deficiency), full gene sequence CPOX (coproporphyrinogen oxidase) (eg, hereditary coproporphyria), full gene sequence CTRC (chymotrypsin C) (eg, hereditary pancreatitis), full gene sequence CYP11B1 (cytochrome P450, family 17, subfamily A, polypeptide 1) (eg, congenital adrenal hyperplasia), full gene sequence CYP17A1 (cytochrome P450, family 17, subfamily A, polypeptide 1) (eg, congenital adrenal hyperplasia), full gene sequence CYP2A2 (cytochrome P450, family 21, subfamily A1, polypeptide2) (eg, steroid 21-dydroxlase isoform, congenital adrenal hyperplasia), full gene sequence Cytogenomic constitutional targeted microarray analysis of chromosome 22q13 by interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants for chromosomal abnormalities (When performing genome-wide cytogenomic constitutional microarray analysis, see 81228-81229) (Do not report analyte-specific molecular pathology procedures separately when the specific analytes are included as part of the microarray analysis of chromosome 22q13) DBT (dihydrolipoamide branched chain transacylase E2) (eg, maple syrup urine disease, type 2), duplication/deletion analysis DCX (doublecortin) (eg, X-linked lissenecephaly), full gene sequence DES (desmin) (eg, myofibrillar myopathy), full gene sequence DRNB59 (deafness, autosomal recessive 59) (eg, autosomal recessive

nonsyndromic hearing impairment), full gene sequence DGUOK (deoxyguanosine kinase) (eg, hepatocerebral mitochondrial DNA depletion syndrome), full gene sequence DHCR7 (7 dehydrocholesterol reductase) (eg, Smith-Lemli-Opitz syndrome), full gene sequence EIF2B2 (eukaryotic translation initiation factor 2B, subunit 2 beta, 39kDa) (eg, leukoencephalopathy with vanishing white matter), full gene sequence EMD (emerin) (eg, Emery-Dreifuss muscular dystrophy), full gene sequence ENG (endoglin) (eg, hereditary hemorrhagic telangiectasia, type 1) (duplication/deletion analysis EYA1 (eyes absent homolog 1 [drosophila]) (eg, branchio-oto-renal [BOR] spectrum disorders), duplication/deletion analysis FGFR1 (fibroblast growth factor receptor 1) (eg, Kallmann syndrome 2), 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 FTSJ1 (FtsJ RNA methyltransferase homolog 1 [E. coli]) (eg, X-lined mental retardation 9), duplication/deletion analysis GABRG2 (gamma-aminobutyric acid [GABA] A receptor gamma 2 (eg, generalized epilepsy with febrile seizures), full gene sequence GCH1 (GTP cyclohydrolase 1) (eg, autosomal dominant dopa-responsive dystonia), full gene sequence GDAP1 (ganglioside-induced differentiation-associated protein 1) (eg, Charcot-Marie-Tooth disease), full gene sequence GFAP (glial fibrillary acidic protein) (eg, Alexander disease), full gene sequence GHR (growth hormone receptor) (eg, Laron syndrome), full gene sequence GHRHR (growth hormone releasing hormone receptor) (eg, growth hormone deficiency), full gene sequence GLA (galactosidase, alpha) (eg, Fabry disease), full gene sequence HNF1A (HNF1 homeobox A (eg, maturity-onset diabetes of the young [MODY]), full gene sequence HNF1B (HNG1 homeobox B) (eg, maturity-onset diabetes of the young [MODY]) full gene sequence HTRA1 (HtrA serine peptidase 1) (eg, macular degeneration), full gene sequence IDS (iduronate 2 sulfatase) (eg, mucopolysacchridosis, type II), full gene sequence IL2RG (interleukin 2 receptor, gamma) (eg, X-linked severe combined immunodeficiency), full gene sequence ISPD (isoprenoid synthase domain containing) (eg, Noonan syndrome), full gene sequence LAMP2 (lysosomal-associated membrane protein2) (eg, Danon disease), full gene sequence LDLR (low density lipoprotein receptor) (eg, familial hypercholesterolemia), duplication/deletion analysis MEN1 (multiple endocrine neoplasia 1) (eg, multiple endocrine neoplasia type 1, Wermer syndrome), full gene sequence MMAA (methylmalonic aciduria[cobalamine deficiency] type A) (eg, MMAA-related methylmalonic academia), full gene sequence MMAB (methylmalonic aciduria[cobalamine deficiency] type B) (eg, MMAA-related methylmalonic academia), full gene MPI (mannose phosphate isomerase) (eg, congenital disorder of glycosylation 1b), full gene sequence MPV17 (MpV17 mitochondrial inner membrane protein) (eg, mitochondrial DNA depletion syndrome), full gene sequence MPZ (myelin protein zero) (eg, Charcot-Marie-Tooth), full gene sequence MTM1 (myotubularin 1) (eg, X-linked centronuclear myopathy), duplication/deletion analysis 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 NDUFS7 (NADH dehydrogenase [ubiquinone] Fe-S protein 7, 20kDa [NADH- coenzyme Q reductase]) (eg, Leigh syndrome, mitochondrial complex 1 deficiency), full gene sequence NDUFS8 (NADH dehydrogenase [ubiquinone] Fe-S protein 8, 23kDa [NADH-coenzyme Q reductase]) (eg, Leigh syndrome, mitochondrial complex 1 deficiency), full gene sequence NDUFV1 (NADH dehydrogenase [ubiquinone] flavoprotein 1,51kDa) (eg, Leigh syndrome, mitochondrial complex 1 deficiency), full gene sequence NEFL (neruofilament, light polypeptide) (eg,

Charcot-Marie-Tooth), full gene sequence NF2 (neurofibromin 2 [merlin]) (eg, neurofibromatosis, type2), duplication/deletion analysis NLGN3 (neuroligin 3) (eg, autism spectrum disorders), full gene sequence NLGN4X (neuroligin 4, X-linked) (eg, autism spectrum disorders), full gene sequence NPHP1 (nephronophthisis 1 [juvenile]) (eg, Joubert syndrome), deletion analysis, and duplication analysis, if performed NPHS2 (nephrosis 2, idiopathic, steroid-resistant [podocin]) (eg, steroid-resistant nephrotic syndrome), full gene sequence NSD1 (nuclear receptor binding SET domain protein 1) (eg, Sotos syndrome), duplication/deletion analysis OTC (ornithine carbamoyltransferase) (eg, ornithine transcarbamylase deficiency), full gene sequence PAFAH1B1 (platelet-activating factor acetylhydrolase 1b, regulatory subunit 1 [45kDa]) (eg, lissenecephaly, Miller-Dieker syndrome), duplication/deletion analysis PARK2 (Parkinson protein 2, E3 ubiquitin protein ligase [parkin]) (eg, Parkinson disease), duplication/deletion analysis PCCA (propionyl) CoA carboxylase, alpha polypeptide) (eg, propionic academia, type 1), duplication/deletion analysis PCDH19 (protocadherin 19) (eg, epileptic encephalopathy), full gene sequence PDHA1 (pyruvate dehydrogenase [lipoamide] beta) (eg, lactic acidosis), full gene sequence PKLR (pyruvate kinase, liver and RBC) (eg, pyruvate kinase deficiency), full gene sequence PLP1 proteolipid protein 1) (eg, Pelizaeus-Merzbacher disease, spastic paraplegia), full gene sequence POU1F1 POU class 1 homeobox 1) (eg, combined pituitary hormone deficiency), full gene sequence PRX (periaxin) (eg, Charcot-Marie-Tooth disease), full gene sequence PQBP1 (polyglutamine binding protein 1) (eg, Renpenning syndrome), full gene sequence PSEN1 (presenilin 1) (eg, Alzheimer disease), full gene sequence RAB7A (RAB7A, member RAS oncogene family) (eg, Charcot-Marie-Tooth disease), full gene sequence RAI1 (ret proto-oncogene) (eg, multiple endocrine neoplasia, type 1A and familial medullary thyroid carcinoma), targeted sequence analysis (eg, exons 10, 11, 13-16) RPS19 (ribosomal protein S19) (eg, Diamond-Blackfan anemia), full gene sequence RRM2B (ribonucleotide reductase M 2 B [TP53 inducible]) (eg, mitochondrial DNA depletion), full gene sequence SCO1 (SCO cytochrome oxidase deficient homolog 1) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence SDHB (succinate dehydrogenase complex, subunit B, iron sulfur), eg, hereditary paraganglioma),full gene sequence SDHC (succinate dehydrogenase complex, subunit C, intergral 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, gamma [35kDa dystrophin-associated glycoprotein]) (eg, limb-girdle muscular dystrophy), full gene sequence SGCE (sarcoglycan, epsilon) (eg, myoclonic dystonia), duplication/deletion analysis 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 SHOX (short stature homeobox) (eg, Langer mesomelic dysplasia), full gene sequence SIL1 (SIL1 homolog, endoplasmic reticulum chaperone [S. cerevisiae]) (eg, ataxia), full gene sequence SCLC2A (solute carrier family 2 [facilitated glucose transporter]. Member 1) (eg, glucose transporter type 1 [GLUT 1) deficiency syndrome), full gene sequence SLC16A2, (solute carrier family 16, member 2 [thyroid hormone transporter]) (eg, specific thyroid hormone cell transporter deficiency, Allan-Herndon-Dudley syndrome), full gene sequence SLC22A5 (solute carrier family 22 [organic cation/carnitine transporter], member 5) (eg, systemic primary carnitine deficiency), full gene sequence SLC25A20 (solute carrier family 25 [carnitine/acylcarnitine translocase], member 20) (eg, carnitine-acylcarnitine translocase deficiency), full gene sequence

SMAD4 (SMAD family member 4) (eg, hemorrhagic telangiectasia syndrome, juvenile polyposis), duplication/deletion analysis SPAST (spastin) (eg, spastic paraplegia), duplication/deletion analysis SPG7 (spastic paraplegia 7 [pure and complicated autosomal recessive]) (eg, spastic paraplegia duplication/deletion analysis SPRED1 (sprout-related, EVH1 domain containing 1) (eg, Legius syndrome), full gene sequence STAT3 (signal transducer and activator of transcription 3 [acute-phase response factor]) (eg, autosomal dominant hyper-IgE syndrome), targeted sequence analysis)eg, exons 12, 13, 14, 16, 17, 20, 21 STK11 (serine/threonine kinase 1) (eg, Peutz-Jeghers syndrome), full gene sequence SURF1 (surfeit 1) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence TARDBP (TAR DNA binding protein) (eg, amyotrophic lateral sclerosis), full gene sequence TBX5 (T-box 5) (eg, Holt-Oram syndrome), full gene sequence TCF4 (transcription factor 4) (eg, Pitt-Hopkins syndrome), duplication/deletion analysis 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 TK2 (thymidine kinase 2, mitochondrial) (eg, mitochondrial DNA depletion syndrome), full gene sequence TNNC1 (troponin C type 1 [slow]) (eg, hypertrophic cardiomyopathy or dilated cardiomyopathy), full gene sequence 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 TYMP (thymidine phosphorylase) (eg, mitochondrial DNA depletion syndrome), full gene sequence VWF (von Willebrand factor) eg, von Willebrand disease type 2N), targeted sequence analysis (eg, exons 18-20, 23-25) WT1 (Wilms tumor 1) (eg, Denys-Drahs syndrome, familial Wilms tumor), full gene sequence AEB2 (zinc finger E-box binding homeobox 2) (eg, Mowat-Wilson syndrome), full gene sequence

Multianalyte Assays with Algorithmic Analyses

Deleted Code for 2021:

81545 Oncology (thyroid), gene expression analysis of 142 genes, utilizing fine needle aspirate, algorithm reported as a categorical result (eg, benign or suspicious)

Chemistry

New codes for 2021:

82077 Alcohol (ethanol); any specimen except urine and breath immunoassay (eg, IA, EIA, ELISA, RIA, EMIT, FPIA) and enzymatic methods (eg, alcohol dehydrogenase)
82681 Estradiol; free, direct measurement (eg, equilibrium dialysis)

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Revised Codes for 2021:

82075 Alcohol (ethanol); breath
82670 Estradiol; total

Proprietary Laboratory Analyses

New Codes for 2021:

0068U Candida species panel (C. albicans, C. glabrata, C. parapsilosis, C. kruseii, C. tropicalis, and C. auris), amplified probe technique with qualitative report of the presence or absence of each species

0139U Neurology (autism spectrum disorder [ASD]), quantitative measurements of 6 central carbon metabolites (ie, αketoglutarate, alanine, lactate, phenylalanine, pyruvate, and succinate), LC-MS/MS, plasma, algorithmic analysis with result reported as negative or positive (with metabolic subtypes of ASD)

0140U Infectious disease (fungi), fungal pathogen identification, DNA (15 fungal targets), blood culture, amplified probe technique, each target reported as detected or not detected

0141U Infectious disease (bacteria and fungi), gram-positive organism identification and drug resistance element detection, DNA (20 gram-positive bacterial targets, 4 resistance genes, 1 pan gram-negative bacterial target,1 pan Candida target), blood culture, amplified probe technique, each target reported as detected or not detected.

0142U Infectious disease (bacteria and fungi), gram-negative bacterial identification and drug resistance element detection, DNA (21 gram-negative bacterial targets, 6 resistance genes, 1 pan gram-positive bacterial target, 1 pan Candida target), amplified probe technique, each target reported as detected or not detected

0143U Drug assay, definitive, 120 or more drugs or metabolites, urine, quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service (For additional PLA code with identical clinical descriptor, see 0150U. See Appendix O to determine appropriate code assignment)

0144U Drug assay, definitive, 160 or more drugs or metabolites, urine, quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service

0145U Drug assay, definitive, 65 or more drugs or metabolites, urine, quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service

0146U Drug assay, definitive, 80 or more drugs or metabolites, urine, by quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service

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0147U Drug assay, definitive, 85 or more drugs or metabolites, urine, quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service

0148U Drug assay, definitive, 100 or more drugs or metabolites, urine, quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service

0149U Drug assay, definitive, 60 or more drugs or metabolites, urine, quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service

0150U Drug assay, definitive, 120 or more drugs or metabolites, urine, quantitative liquid chromatography with tandem mass spectrometry (LC-MS/MS) using multiple reaction monitoring (MRM), with drug or metabolite description, comments including sample validation, per date of service (For additional PLA code with identical clinical descriptor, see 0143U. See Appendix O to determine appropriate code assignment)

0151U Infectious disease (bacterial or viral respiratory tract infection), pathogen specific nucleic acid (DNA or RNA), 33 targets, real-time semi-quantitative PCR, bronchoalveolar lavage, sputum, or endotracheal aspirate, detection of 33 organismal and antibiotic resistance genes with limited semi-quantitative results

0152U Infectious disease (bacteria, fungi, parasites, and DNA viruses), DNA, PCR and next generation sequencing, plasma, detection of >1,000 potential microbial organisms for significant positive pathogens

0153U Oncology (breast), mRNA, gene expression profiling by next-generation sequencing of 101 genes, utilizing formalin-fixed paraffin-embedded tissue, algorithm reported as a triple negative breast cancer clinical subtype(s) with information on immune cell involvement

0156U Copy number (eg, intellectual disability, dysmorphology), sequence analysis

0163U Oncology (colorectal) screening, biochemical enzyme-linked immunosorbent assay (ELISA) of 3 plasma or serum proteins (teratocarcinoma derived growth factor-1 [TDGF-1, Cripto1], carcinoembryonic antigen [CEA], extracellular matrix protein [ECM]), with demographic data (age, gender, CRC screening compliance) using a proprietary algorithm and reported as likelihood of CRC or advanced adenomas

0164U Gastroenterology (irritable bowel syndrome [IBS]), immunoassay for antiCdtB and anti-vinculin antibodies, utilizing plasma, algorithm for elevated or not elevated qualitative results

0166U Liver disease, 10 biochemical assays (α2- macroglobulin, haptoglobin, apolipoprotein A1, bilirubin, GGT, ALT, AST, triglycerides, cholesterol, fasting glucose) and biometric and demographic data, utilizing serum, algorithm reported as scores for fibrosis, necroinflammatory activity, and steatosis with a summary interpretation

0167U Gonadotropin, chorionic (hCG), immunoassay with direct optical observation, blood

0168U Fetal aneuploidy (trisomy 21, 18, and 13) DNA sequence analysis of selected regions using maternal plasma without fetal fraction cutoff, algorithm reported as a risk score for each trisomy

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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

0171U Targeted genomic sequence analysis panel, acute myeloid leukemia, myelodysplastic syndrome, and myeloproliferative neoplasms, DNA analysis, 23 genes, interrogation for sequence variants, rearrangements and minimal residual disease, reported as presence/absence

0172U Oncology (solid tumor as indicated by the label), somatic mutation analysis of BRCA1 (BRCA1, DNA repair associated), BRCA2 (BRCA2, DNA repair associated) and analysis of homologous recombination deficiency pathways, DNA, formalin-fixed paraffin-embedded tissue, algorithm quantifying tumor genomic instability score

0173U Psychiatry (ie, depression, anxiety), genomic analysis panel, includes variant analysis of 14 genes

0174U Oncology (solid tumor), mass spectrometric 30 protein targets, formalin-fixed paraffin-embedded tissue, prognostic and predictive algorithm reported as likely, unlikely, or uncertain benefit of 39 chemotherapy and targeted therapeutic oncology agents

0175U Psychiatry (eg, depression, anxiety), genomic analysis panel, variant analysis of 15 genes

0176U Cytolethal distending toxin B (CdtB) and vinculin IgG antibodies by immunoassay (ie, ELISA)

0177U Oncology (breast cancer), DNA, PIK3CA (phosphatidylinositol-4, 5-bisphosphate 3- kinase catalytic subunit alpha) gene analysis of 11 gene variants utilizing plasma, reported as PIK3CA gene mutation status

0178U Peanut allergen-specific quantitative assessment of multiple epitopes using enzyme-linked immunosorbent assay (ELISA), blood, report of minimum eliciting exposure for a clinical reaction

0179U Oncology (non-small cell lung cancer), cell-free DNA, targeted sequence analysis of 23 genes (single nucleotide variations, insertions and deletions, fusions without prior knowledge of partner/breakpoint, copy number variations), with report of significant mutation(s)

0180U Red cell antigen (ABO blood group) genotyping (ABO), gene analysis Sanger/chain termination/conventional sequencing, ABO (ABO, alpha 1-3-Nacetylgalactosaminyltransferase 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

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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
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
0202U Infectious disease (bacterial or viral respiratory tract infection), pathogen-specific nucleic acid (DNA or RNA), 22 targets including severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2), qualitative RT-PCR, nasopharyngeal swab, each pathogen reported as detected or not detected
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
0204U Oncology (thyroid), mRNA, gene expression analysis of 593 genes (including BRAF, RAS, RET, PAX8, and NTRK) for sequence variants and rearrangements, utilizing fine needle aspirate, reported as detected or not detected
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

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0206U Neurology (Alzheimer disease); cell aggregation using morphometric imaging and protein kinase C-epsilon (PKCe) concentration in response to amylospheroid treatment by ELISA, cultured skin fibroblasts, each reported as positive or negative for Alzheimer disease

0208U Oncology (medullary thyroid carcinoma), mRNA, gene expression analysis of 108 genes, utilizing fine needle aspirate, algorithm reported as positive or negative for medullary thyroid carcinoma

0209U Cytogenomic constitutional (genome-wide) analysis, interrogation of genomic regions for copy number, structural changes and areas of homozygosity for chromosomal abnormalities

0210U Syphilis test, non-treponemal antibody, immunoassay, quantitative (RPR)

0211U Oncology (pan-tumor), DNA and RNA by next-generation sequencing, utilizing formalin-fixed paraffin-embedded tissue, interpretative report for single nucleotide variants, copy number alterations, tumor mutational burden, and microsatellite instability, with therapy association

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
(Do not report 0212U in conjunction with 81425)

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)
(Do not report 0213U in conjunction with 81426)

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
(Do not report 0214U in conjunction with 81415)

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)
(Do not report 0215U in conjunction with 81416)

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

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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

0219U Infectious agent (human immunodeficiency virus), targeted viral next-generation sequence analysis (ie, protease [PR], reverse transcriptase [RT], integrase [INT]), algorithm reported as prediction of antiviral drug susceptibility

0220U Oncology (breast cancer), image analysis with artificial intelligence assessment of 12 histologic and immunohistochemical features, reported as a recurrence score

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

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

Add on codes for 2021:

+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) (Use 0157U in conjunction with 81201)

+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)
(Use 0158U in conjunction with 81292)

+0159U MSH2 (mutS homolog 2) (eg, hereditary colon cancer, Lynch syndrome) mRNA sequence analysis (List separately in addition to code for primary procedure)
(Use 0159U in conjunction with 81295)

+0160U MSH6 (mutS homolog 6) (eg, hereditary colon cancer, Lynch syndrome) mRNA sequence analysis (List separately in addition to code for primary procedure)
(Use 0160U in conjunction with 81298)

+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)
(Use 0161U in conjunction with 81317)

+0162U Hereditary colon cancer (Lynch syndrome), targeted mRNA sequence analysis panel (MLH1, MSH2, MSH6, PMS2) (List separately in addition to code for primary procedure)
(Use 0162U in conjunction with 81292, 81295, 81298, 81317, 81435)

+0207U Quantitative imaging of phosphorylated ERK1 and ERK2 in response to bradykinin treatment by in situ immunofluorescence, using cultured skin fibroblasts, reported as a probability index for Alzheimer disease (List separately in addition to code for primary procedure)
(Use 0207U in conjunction with 0206U)

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Deleted Codes for 2021:

0081U Oncology (uveal melanoma) mRNA, gene-expression profiling by real-time RT-PCR of 15 genes (12, content and 3 housekeeping genes), utilizing fine needle aspirate or formalin-fixed paraffin-embedded tissue, algorithm reported as a risk of metastasis. Deleted crosswalk CPT 81552

0104U Hereditary pan cancer (eg, hereditary breast and ovarian cancer, hereditary endometrial cancer, hereditary colorectal cancer, genomic sequence analysis panel utilizing a combination of NGS, Sanger, MLPA and array CGH, with mRNA analytics to resolve variants of unknown significance when indicated (32 genes [sequencing and deletion/duplication] EPCAM and GREM1 [deletion/duplication only]).

The following resources were used in the preparation of this document: the AMA's *Current Procedural Terminology (CPT) 2021, and CPT 2021 Changes-An Insider's View.*