Appendix B

Summary of Additions, Deletions, and Revisions

Appendix B shows the actual changes that were made to the code descriptors. New codes appear with a bullet (●) and are indicated as "Code Added." Revised codes are preceded with a triangle (▲). Within revised codes, the deleted language appears with a ~~strikethrough~~, while new text appears underlined. Codes with which moderate sedation would not be separately reported when performed at the same session by the same individual are denoted with the bullseye (⊙). The symbol ⌁ is used to identify codes for vaccines that are pending FDA approval (see **Appendix K**).The symbol # is used to identify codes that have been resequenced (see **Appendix N**). CPT add-on codes are annotated by the symbol + (see **Appendix D**). The symbol ⊘ is used to identify codes that are exempt from the use of modifier 51 (see **Appendix E**).

Evaluation and Management (E/M) Services Guidelines

Evaluation and Management

●	**99446**	Code added
●	**99447**	Code added
●	**99448**	Code added
●	**99449**	Code added
#+●	**99481**	Code added
#+●	**99482**	Code added

Surgery

⊙●	**10030**	Code added
	~~13150~~	~~Repair, complex, eyelids, nose, ears and/or lips; 1.0 cm or less~~
▲	**13151**	Repair, complex, eyelids, nose, ears and/or lips; 1.1 cm to 2.5 cm
▲	**13152**	2.6 cm to 7.5 cm
+▲	**13153**	each additional 5 cm or less (List separately in addition to code for primary procedure)
+▲	**15777**	Implantation of biologic implant (eg, acellular dermal matrix) for soft tissue reinforcement (~~egie~~, breast, trunk) (List separately in addition to code for primary procedure);
●	**19081**	Code added
+●	**19082**	Code added
●	**19083**	Code added
+●	**19084**	Code added
●	**19085**	Code added
+●	**19086**	Code added
	~~19102~~	~~percutaneous, needle core, using imaging guidance~~
	~~19103~~	~~percutaneous, automated vacuum assisted or rotating biopsy device, using imaging guidance~~
●	**19281**	Code added
+●	**19282**	Code added
●	**19283**	Code added
+●	**19284**	Code added
●	**19285**	Code added
+●	**19286**	Code added
●	**19287**	Code added
+●	**19288**	Code added
	~~19290~~	~~Preoperative placement of needle localization wire, breast;~~
	~~19291~~	~~each additional lesion (List separately in addition to code for primary procedure)~~
	~~19295~~	~~Image guided placement, metallic localization clip, percutaneous, during breast biopsy/aspiration (List separately in addition to code for primary procedure);~~
▲	**21015**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of face or scalp; less than 2 cm
▲	**21016**	2 cm or greater
▲	**21557**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of neck or anterior thorax; less than 5 cm
▲	**21558**	5 cm or greater
▲	**21935**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of back or flank; less than 5 cm
▲	**21936**	5 cm or greater
▲	**22904**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of abdominal wall; less than 5 cm
▲	**22905**	5 cm or greater
▲	**23077**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of shoulder area; less than 5 cm
▲	**23078**	5 cm or greater
	~~23331~~	~~deep (eg, Neer hemiarthroplasty removal)~~
	~~23332~~	~~complicated (eg, total shoulder)~~
●	**23333**	Code added
●	**23334**	Code added
●	**23335**	Code added
▲	**24077**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of upper arm or elbow area; less than 5 cm
▲	**24079**	5 cm or greater
▲	**24160**	~~Implant removal~~Removal of prosthesis, includes debridement and synovectomy when performed; ~~elbow joint~~humeral **and** ulnar components
▲	**24164**	radial head
▲	**25077**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of forearm and/or wrist area; less than 3 cm
▲	**25078**	3 cm or greater
▲	**26117**	Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of hand or finger; less than 3 cm

▲ **26118** 3 cm or greater

▲ **27049** Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of pelvis and hip area; less than 5 cm

#▲ **27059** 5 cm or greater

#▲ **27329** Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of thigh or knee area; less than 5 cm

▲ **27364** 5 cm or greater

▲ **27615** Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of leg or ankle area; less than 5 cm

▲ **27616** 5 cm or greater

▲ **28046** Radical resection of tumor (eg, ~~malignant neoplasm~~sarcoma), soft tissue of foot or toe; less than 3 cm

▲ **28047** 3 cm or greater

~~32201~~ ~~with percutaneous drainage of abscess or cyst~~

⊙▲ **33222** ~~Revision or relocation~~Relocation of skin pocket for pacemaker;

⊙▲ **33223** ~~Revision~~Relocation of skin pocket for cardioverter-defibrillator;

⊙▲ **33282** Implantation of patient-activated cardiac event recorder

⊙▲ **33284** Removal of an implantable, patient-activated cardiac event recorder

● **33366** Code added

● **34841** Code added

● **34842** Code added

● **34843** Code added

● **34844** Code added

● **34845** Code added

● **34846** Code added

● **34847** Code added

● **34848** Code added

~~37204~~ ~~Transcatheter occlusion or embolization (eg, for tumor destruction, to achieve hemostasis, to occlude a vascular malformation), percutaneous, any method, non-central nervous system, non-head or neck;~~

~~37205~~ ~~Transcatheter placement of an intravascular stent(s) (except coronary, carotid, vertebral, iliac, and lower extremity arteries), percutaneous; initial vessel~~

~~37206~~ ~~each additional vessel (List separately in addition to code for primary procedure)~~

~~37207~~ ~~Transcatheter placement of an intravascular stent(s) (except coronary, carotid, vertebral, iliac and lower extremity arteries), open; initial vessel~~

~~37208~~ ~~Transcatheter placement of an intravascular stent(s) (except coronary, carotid, vertebral, iliac and lower extremity arteries), open; each additional vessel (List separately in addition to code for primary procedure)~~

~~37210~~ ~~Uterine fibroid embolization (UFE, embolization of the uterine arteries to treat uterine fibroids, leiomyomata), percutaneous approach inclusive of vascular access, vessel selection, embolization, and all radiological supervision and interpretation, intraprocedural roadmapping, and imaging guidance necessary to complete the procedure;~~

● **37217** Code added

⊙● **37236** Code added

⊙✚● **37237** Code added

⊙● **37238** Code added

⊙✚● **37239** Code added

⊙● **37241** Code added

⊙● **37242** Code added

⊙● **37243** Code added

⊙● **37244** Code added

~~42802~~ ~~hypopharynx~~

● **43191** Code added

● **43192** Code added

● **43193** Code added

● **43194** Code added

● **43195** Code added

● **43196** Code added

● **43197** Code added

● **43198** Code added

⊙▲ **43200** Esophagoscopy, ~~rigid or~~ flexible, transoral; diagnostic, ~~with or without~~including collection of specimen(s) by brushing or washing, when performed (separate procedure)

⊙▲ **43201** with directed submucosal injection(s), any substance

⊙▲ **43202** with biopsy, single or multiple

⊙▲ **43204** with injection sclerosis of esophageal varices

⊙▲ **43205** with band ligation of esophageal varices

⊙▲ **43206** with optical endomicroscopy

⊙▲ **43215** with removal of foreign body

⊙▲ **43216** with removal of tumor(s), polyp(s), or other lesion(s) by hot biopsy forceps or bipolar cautery

⊙▲ **43217** with removal of tumor(s), polyp(s), or other lesion(s) by snare technique

#⊙● **43211** Code added

~~43219~~ ~~with insertion of plastic tube or stent~~

#⊙● **43212** Code added

⊙▲ **43220** with transendoscopic balloon dilation (less than 30 mm diameter)

#⊙● **43213** Code added

#⊙● **43214** Code added

⊙▲ **43226** with insertion of guide wire followed by ~~dilation~~passage of dilator(s) over guide wire

⊙▲ **43227** with control of bleeding ~~(eg, injection, bipolar cautery, unipolar cautery, laser, heater probe, stapler, plasma coagulator)~~ any method

~~43228~~ ~~with ablation of tumor(s), polyp(s), or other lesion(s), not amenable to removal by hot biopsy forceps, bipolar cautery or snare technique~~

⊙● **43229** Code added

⊙▲ **43231** with endoscopic ultrasound examination

⊙▲ **43232** with transendoscopic ultrasound-guided intramural or transmural fine needle aspiration/biopsy(s)

⊙▲ **43235** ~~Upper gastrointestinal endoscopy including esophagus~~Esophagogastroduodenoscopy, ~~stomach~~flexible, ~~and either the duodenum and/or jejunum as appropriate~~transoral; diagnostic, ~~with or without~~including collection of specimen(s) by brushing or washing, when performed (separate procedure)

⊙▲ **43236** with directed submucosal injection(s), any substance

⊙▲ **43237** with endoscopic ultrasound examination limited to the esophagus, stomach or duodenum, and adjacent structures

⊙▲ **43238** with transendoscopic ultrasound-guided intramural or transmural fine needle aspiration/biopsy(s), esophagus (includes endoscopic ultrasound examination limited to the esophagus, stomach or duodenum, and adjacent structures)

⊙▲ **43239** with biopsy, single or multiple

⊙▲ **43240** with transmural drainage of pseudocyst (includes placement of transmural drainage catheter[s]/stent[s], when performed, and endoscopic ultrasound, when performed)

⊙▲ **43241** with ~~transendoscopic~~insertion of intraluminal tube or catheter ~~placement~~

⊙▲ **43242** with transendoscopic ultrasound-guided intramural or transmural fine needle aspiration/biopsy(s) (includes endoscopic ultrasound examination of the esophagus, stomach, and either the duodenum ~~and/~~or a surgically altered stomach where the jejunum ~~as appropriate~~is examined distal to the anastomosis)

⊙▲ **43243** with injection sclerosis of esophageal ~~and/or~~ gastric varices

⊙▲ **43244** with band ligation of esophageal ~~and/or~~ gastric varices

⊙▲ **43245** with dilation of gastric ~~outlet for obstruction~~/duodenal stricture(s) (eg, balloon~~, guide wire~~, bougie)

⊙▲ **43246** with directed placement of percutaneous gastrostomy tube

⊙▲ **43247** with removal of foreign body

⊙▲ **43248** with insertion of guide wire followed by ~~dilation~~passage of dilator(s) through esophagus over guide wire

⊙▲ **43249** with transendoscopic balloon dilation of esophagus (less than 30 mm diameter)

#⊙● **43233** Code added

⊙▲ **43250** with removal of tumor(s), polyp(s), or other lesion(s) by hot biopsy forceps or bipolar cautery

⊙▲ **43251** with removal of tumor(s), polyp(s), or other lesion(s) by snare technique

⊙▲ **43252** with optical endomicroscopy

⊙● **43253** Code added

⊙● **43254** Code added

⊙▲ **43255** with control of bleeding, any method

~~43256~~ ~~with transendoscopic stent placement (includes predilation)~~

#⊙● **43266** Code added

⊙▲ **43257** with delivery of thermal energy to the muscle of lower esophageal sphincter and/or gastric cardia, for treatment of gastroesophageal reflux disease

~~43258~~ ~~with ablation of tumor(s), polyp(s), or other lesion(s) not amenable to removal by hot biopsy forceps, bipolar cautery or snare technique~~

#⊙● **43270** Code added

⊙▲ **43259** with endoscopic ultrasound examination, including the esophagus, stomach, and either the duodenum ~~and/~~or a surgically altered stomach where the jejunum ~~as appropriate~~is examined distal to the anastomosis

⊙▲ **43260** Endoscopic retrograde cholangiopancreatography (ERCP); diagnostic, ~~with or without~~including collection of specimen(s) by brushing or washing, when performed (separate procedure)

⊙▲ **43263** with pressure measurement of sphincter of Oddi ~~(pancreatic duct or common bile duct)~~

⊙▲ **43264** with ~~endoscopic retrograde~~ removal of ~~calculus~~calculi/~~calculi~~debris from biliary ~~and/or~~ pancreatic ducts~~(s)~~

⊙▲ **43265** with ~~endoscopic retrograde~~ destruction~~, lithotripsy~~ of ~~calculus/~~ calculi, any method (eg, mechanical, electrohydraulic, lithotripsy)

~~43267~~ ~~with endoscopic retrograde insertion of nasobiliary or nasopancreatic drainage tube~~

~~43268~~ ~~with endoscopic retrograde insertion of tube or stent into bile or pancreatic duct~~

#⊙● **43274** Code added

~~43269~~ ~~with endoscopic retrograde removal of foreign body and/or change of tube or stent~~

~~43271~~ ~~with endoscopic retrograde balloon dilation of ampulla, biliary and/or pancreatic duct(s)~~

~~43272~~ ~~with ablation of tumor(s), polyp(s), or other lesion(s) not amenable to removal by hot biopsy forceps, bipolar cautery or snare technique~~

#⊙● **43275** Code added

#⊙● **43276** Code added

#⊙● **43277** Code added

#⊙● **43278** Code added

⊙✚▲ **43273** Endoscopic cannulation of papilla with direct visualization of pancreatic/common bile ~~duct(s) and/or pancreatic~~ duct(s) (List separately in addition to code(s) for primary procedure)

~~43456~~ ~~Dilation of esophagus, by balloon or dilator, retrograde;~~

~~43458~~ ~~Dilation of esophagus with balloon (30 mm diameter or larger) for achalasia;~~

~~44901~~ ~~percutaneous~~

~~47011~~ ~~for percutaneous drainage of abscess or cyst, 1 or 2 stages~~

▲ **47552** Biliary endoscopy, percutaneous via T-tube or other tract; diagnostic, with ~~or without~~ collection of specimen(s) by brushing and/or washing, when performed (separate procedure)

~~48511~~ ~~percutaneous~~

~~49021~~ ~~percutaneous~~

~~49041~~ ~~percutaneous~~

~~49061~~ ~~percutaneous~~

⊙● **49405** Code added

⊙● **49406** Code added

⊙● **49407** Code added

~~50021~~ ~~percutaneous~~

#● **52356** Code added

~~58823~~ ~~Drainage of pelvic abscess, transvaginal or transrectal approach, percutaneous (eg, ovarian, pericolic);~~

~~64613~~ ~~neck muscle(s) (eg, for spasmodic torticollis, spasmodic dysphonia)~~

~~64614~~ ~~extremity and/or trunk muscle(s) (eg, for dystonia, cerebral palsy, multiple sclerosis)~~

● **64616** Code added

● **64617** Code added

● **64642** Code added

+● **64643** Code added

● **64644** Code added

+● **64645** Code added

● **64646** Code added

● **64647** Code added

▲ **65778** Placement of amniotic membrane on the ocular surface ~~for wound healing~~; ~~self-retaining~~without sutures

▲ **65779** single layer, sutured

● **66183** Code added

▲ **69210** Removal impacted cerumen ~~(separate procedure)~~requiring instrumentation, ~~1 or both ears~~unilateral

Radiology

▲ **72040** Radiologic examination, spine, cervical; 2 or 3 views ~~or less~~

~~75960~~ ~~Transcatheter introduction of intravascular stent(s) (except coronary, carotid, vertebral, iliac, and lower extremity artery), percutaneous and/or open, radiological supervision and interpretation, each vessel;~~

~~77031~~ ~~Stereotactic localization guidance for breast biopsy or needle placement (eg, for wire localization or for injection), each lesion, radiological supervision and interpretation;~~

~~77032~~ ~~Mammographic guidance for needle placement, breast (eg, for wire localization or for injection), each lesion, radiological supervision and interpretation;~~

+● **77293** Code added

#▲ **77295** ~~Therapeutic radiology simulation~~3-dimensional radiotherapy plan, including dose ~~aided field setting~~volume histograms; ~~3-dimensional~~

Pathology and Laboratory

● **80155** Code added

● **80159** Code added

● **80169** Code added

● **80171** Code added

● **80175** Code added

● **80177** Code added

● **80180** Code added

● **80183** Code added

● **80199** Code added

● **80203** Code added

#● **81287** Code added

▲ **81371** *HLA-A, -B, and -DRB1~~/3/4/5~~* (eg, verification typing)

▲ **81376** one locus ~~(eg, *HLA-DRB1/3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1*);~~ *(eg, HLA-DRB1, -DRB3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1),* each

▲ **81382** HLA Class II typing, high resolution (ie, alleles or allele groups); one locus (eg, *HLA-DRB1, -DRB3, -DRB4, -DRB5/4/5, -DQB1, -DQA1, -DPB1,* or *-DPA1*), each

▲ **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

BCKDHA (branched chain keto acid dehydrogenase E1, alpha polypeptide) (eg, maple syrup urine disease, type 1A), Y438N 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

FGFR1 (fibroblast growth factor receptor 1) (eg, Pfeiffer syndrome type 1, craniosynostosis), P252R variant

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

FKTN (fukutin) (eg, Fukuyama congenital muscular dystrophy), retrotransposon insertion variant

GNE (glucosamine [UDP-N-acetyl]-2-epimerase/N-acetylmannosamine kinase) (eg, inclusion body myopathy 2 [IBM2], Nonaka myopathy), M712T variant

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

Human Platelet Antigen 2 genotyping (HPA-2), GP1BA (glycoprotein Ib [platelet], alpha polypeptide [GPIba]) (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 IIb of IIb/IIIa complex], antigen CD41 [GPIIb]) (eg, neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-3a/b (I843S)

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

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

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

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

IL28B (interleukin 28B [interferon, lambda 3]) (eg, drug response), rs12979860 variant

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

LCT (lactase-phlorizin hydrolase) (eg, lactose intolerance), 13910 C>T variant

NEB (nebulin) (eg, nemaline myopathy 2), exon 55 deletion variant

PCDH15 (protocadherin-related 15) (eg, Usher syndrome type 1F), R245X 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

SLCO1B1 (solute carrier organic anion transporter family, member 1B1) (eg, adverse drug reaction), V174A 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)

ABCC8 (ATP-binding cassette, sub-family C [CFTR/MRP], member 8) (eg, familial hyperinsulinism), common variants (eg, c.3898-9G>A [c.3992-9G>A], F1388del)

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

ACADM (acyl-CoA dehydrogenase, 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)

AFF2 (AF4/FMR2 family, member 2 [FMR2]) (eg, fragile X mental retardation 2 [FRAXE]), evaluation to detect abnormal (eg, expanded) alleles

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

ATXN1 (ataxin 1) (eg, spinocerebellar ataxia), evaluation to detect abnormal (eg, expanded) alleles

ATXN2 (ataxin 2) (eg, spinocerebellar ataxia), evaluation to detect abnormal (eg, expanded) alleles

ATXN3 (ataxin 3) (eg, spinocerebellar ataxia, Machado-Joseph disease), evaluation to detect abnormal (eg, expanded) alleles

ATXN7 (ataxin 7) (eg, spinocerebellar ataxia), evaluation to detect abnormal (eg, expanded) alleles

ATXN8OS (ATXN8 opposite strand [non-protein coding]) (eg, spinocerebellar ataxia), evaluation to detect abnormal (eg, expanded) alleles

ATXN10 (ataxin 10) (eg, spinocerebellar ataxia), evaluation to detect abnormal (eg, expanded) alleles

CACNA1A (calcium channel, voltage-dependent, P/Q type, alpha 1A subunit) (eg, spinocerebellar ataxia), 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/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])

CNBP (CCHC-type zinc finger, nucleic acid binding protein) (eg, myotonic dystrophy type 2), evaluation to detect abnormal (eg, expanded) alleles

CSTB (cystatin B [stefin B]) (eg, Unverricht-Lundborg disease), evaluation to detect abnormal (eg, expanded) alleles

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/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

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~~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 IVS14+1G>A [Type I])

FGFR3 (fibroblast growth factor receptor 3) (eg, achondroplasia, hypochondroplasia), 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

FLG (filaggrin) (eg, icthyosis vulgaris), common variants (eg, R501X, 2282del4, R2447X, S3247X, 3702delG)

FOXO1/PAX3 ~~(t(1;13))~~(t(2;13)) (eg, ~~Ewing sarcoma/peripheral neuroectodermal tumor~~alveolar rhabdomyosarcoma), translocation analysis, qualitative, and quantitative, if performed

FOXO1/PAX7 ~~(t(2;13))~~(t(1;13)) (eg, alveolar rhabdomyosarcoma), translocation analysis, qualitative, and quantitative, ~~Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative, and quantitative, if performed~~

FUS/DDIT3 (t(12;16)) (eg, myxoid liposarcoma), translocation analysis, qualitative, and quantitative, if performed

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

GALC (galactosylceramidase) (eg, Krabbe disease), common variants (eg, c.857G>A, 30-kb deletion)

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 (eg, expanded) alleles

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

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)

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

PABPN1 (poly[A] binding protein, nuclear 1) (eg, oculopharyngeal muscular dystrophy), evaluation to detect abnormal (eg, expanded) alleles

PAX8/PPARG (t(2;3) (q13;p25)) (eg, follicular thyroid carcinoma), translocation analysis

PPP2R2B (protein phosphatase 2, regulatory subunit B, beta) (eg, spinocerebellar ataxia), evaluation to detect abnormal (eg, expanded) alleles

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)

(For duplication/deletion analysis of SMN1/SMN2, use 81401)

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

TBP (TATA box binding protein) (eg, spinocerebellar ataxia), evaluation to detect abnormal (eg, expanded) alleles

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)

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

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 oncogene 1, 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

ARX (aristaless-related homeobox) (eg, X-linked lissencephaly with ambiguous genitalia, X-linked mental retardation), duplication/deletion analysis

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.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)

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

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, 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)

KCNC3 (potassium voltage-gated channel, Shaw-related subfamily, member 3) (eg, spinocerebellar ataxia), targeted sequence analysis (eg, exon 2)

KCNJ2 (potassium inwardly-rectifying channel, subfamily J, member 2) (eg, Andersen-Tawil syndrome), full gene sequence

KCNJ11 (potassium inwardly-rectifying channel, subfamily J, member 11) (eg, familial hyperinsulinism), full gene sequence

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

MC4R (melanocortin 4 receptor) (eg, obesity), full gene sequence

MICA (MHC class I polypeptide-related sequence A) (eg, solid organ transplantation), common variants (eg, *001, *002)

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

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

SH2D1A (SH2 domain containing 1A) (eg, X-linked lymphoproliferative syndrome), duplication/deletion analysis

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

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 (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)

AFF2 (AF4/FMR2 family, member 2 [FMR2]) (eg, fragile X mental retardation 2 [FRAXE]), characterization of alleles (eg, expanded size and methylation status)

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

CSTB (cystatin B [stefin B]) (eg, Unverricht-Lundborg disease), 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

EMD (emerin) (eg, Emery-Dreifuss muscular dystrophy), duplication/deletion analysis

EPM2A (epilepsy, progressive myoclonus type 2A, Lafora disease [laforin]) (eg, 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, exons 8, 10)

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

FHL1 (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 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

GH1 (growth hormone 1) (eg, growth hormone deficiency), full gene sequence

GP1BB (glycoprotein Ib [platelet], beta polypeptide) (eg, Bernard-Soulier syndrome type B), 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

HSD3B2 (hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid 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

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 syndromeanalysis deletion/duplication ,)

MMACHC (methylmalonic aciduria [cobalamin deficiency] cblC type, with homocystinuria) (eg, methylmalonic acidemia and homocystinuria), full gene sequence

NDP (Norrie disease [pseudoglioma]) (eg, Norrie disease), full gene sequence

NDUFA1 (NADH dehydrogenase [ubiquinone] 1 alpha subcomplex, 1, 7.5kDa) (eg, Leigh syndrome, mitochondrial complex I deficiency), full gene sequence

NDUFAF2 (NADH dehydrogenase [ubiquinone] 1 alpha subcomplex, assembly factor 2) (eg, Leigh syndrome, mitochondrial complex I deficiency), full gene sequence

NDUFS4 (NADH dehydrogenase [ubiquinone] Fe-S protein 4, 18kDa [NADH-coenzyme Q reductase]) (eg, Leigh syndrome, mitochondrial complex I 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

NR0B1 (nuclear receptor subfamily 0, group B, member 1) (eg, congenital adrenal hypoplasia), full gene sequence

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

PDGFRA (platelet-derived growth factor receptor alpha 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

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 protein 1) (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 (peripherin 2 [retinal degeneration, slow]) (eg, retinitis pigmentosa), 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)

RHO (rhodopsin) (eg, retinitis pigmentosa), full gene sequence

RP1 (retinitis pigmentosa 1) (eg, retinitis pigmentosa), full gene sequence

SCN1B (sodium channel, voltage-gated, type I, 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 sequence

SGCG (sarcoglycan, gamma [35kDa dystrophin-associated glycoprotein]) (eg, limb-girdle muscular dystrophy), duplication/deletion analysis

SH2D1A (SH2 domain 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 (solute carrier family 25 [carnitine/acylcarnitine translocase], member 20) (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-Jeghers syndrome), duplication/deletion analysis

TACO1 (translational activator of mitochondrial encoded cytochrome c oxidase I) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence

THAP1 (THAP domain containing, apoptosis associated protein 1) (eg, torsion dystonia), full gene sequence

TOR1A (torsin family 1, member A [torsin A]) (eg, torsion dystonia), full gene sequence

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

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 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)

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*~~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

AR (androgen receptor) (eg, androgen insensitivity syndrome), 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 (COX15 homolog, cytochrome c oxidase assembly protein) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence

CYP11B1 (cytochrome P450, family 11, subfamily B, 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

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

Cytogenomic constitutional targeted microarray analysis of the X chromosome 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 the X chromosome)

(Do not report 88271 when performing cytogenomic microarray analysis)

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)

(Do not report 88271 when performing cytogenomic microarray analysis)

DBT (dihydrolipoamide branched chain transacylase E2) (eg, maple syrup urine disease, type 2), duplication/deletion analysis

DCX (doublecortin) (eg, X-linked lissencephaly), full gene sequence

DES (desmin) (eg, myofibrillar myopathy), full gene sequence

DFNB59 (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

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

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-linked 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

HBA1/HBA2 (alpha globin 1 and alpha 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

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, muscle-eye-brain disease, Walker-Warburg syndrome), 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

LDLR (low density lipoprotein receptor) (eg, familial hypercholesterolemia), duplication/deletion analysis

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

Mitochondrial genome deletions (eg, Kearns-Sayre syndrome [KSS], chronic progressive external ophthalmoplegia [CPEO], Pearson syndrome), deletion analysis, and duplication analysis, if performed

MMAA (methylmalonic aciduria [cobalamine deficiency] type A) (eg, MMAA-related methylmalonic acidemia), full gene sequence

MMAB (methylmalonic aciduria [cobalamine deficiency] type B) (eg, MMAA-related methylmalonic acidemia), full gene sequence

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 I deficiency), full gene sequence

NDUFS8 (NADH dehydrogenase [ubiquinone] Fe-S protein 8, 23kDa [NADH-coenzyme Q reductase]) (eg, Leigh syndrome, mitochondrial complex I deficiency), full gene sequence

NDUFV1 (NADH dehydrogenase [ubiquinone] flavoprotein 1, 51kDa) (eg, Leigh syndrome, mitochondrial complex I deficiency), full gene sequence

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

NF2 (neurofibromin 2 [merlin]) (eg, neurofibromatosis, type 2), duplication/deletionanalysis

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, lissencephaly, 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 acidemia, type 1), duplication/deletion analysis

PCDH19 (protocadherin 19) (eg, epileptic encephalopathy), full gene sequence

PDHA1 (pyruvate dehydrogenase [lipoamide] alpha 1) (eg, lactic acidosis), duplication/deletion analysis

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

PINK1 (PTEN induced putative kinase 1) (eg, Parkinson disease), 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 (retinoic acid induced 1) (eg, Smith-Magenis syndrome), full gene sequence

REEP1 (receptor accessory protein 1) (eg, spastic paraplegia), 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)

RPS19 (ribosomal protein S19) (eg, Diamond-Blackfan anemia), full gene sequence

RRM2B (ribonucleotide reductase M2 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, 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

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

SLC2A1 (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

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

SPAST (spastin) (eg, spastic paraplegia), duplication/deletion analysis

SPG7 (spastic paraplegia 7 [pure and complicated autosomal recessive]) (eg, spastic paraplegia), duplication/deletion analysis

SPRED1 (sprouty-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 11) (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-Drash syndrome, familial Wilms tumor), full gene sequence

ZEB2 (zinc finger E-box binding homeobox 2) (eg, Mowat-Wilson syndrome), full gene sequence

▲ **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

AFG3L2 (AFG3 ATPase family gene 3-like 2 [S. cerevisiae]) (eg, spinocerebellar ataxia), full gene sequence

AIRE (autoimmune regulator) (eg, autoimmune polyendocrinopathy syndrome type 1), full gene sequence

ALDH7A1 (aldehyde dehydrogenase 7 family, member A1) (eg, pyridoxine-dependent epilepsy), 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

ASS1 (argininosuccinate synthase 1) (eg, citrullinemia type I), full gene sequence

ATL1 (atlastin GTPase 1) (eg, spastic paraplegia), full gene sequence

ATP1A2 (ATPase, Na+/K+ transporting, alpha 2 polypeptide) (eg, familial hemiplegic migraine), full gene sequence

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

BBS1 (Bardet-Biedl syndrome 1) (eg, Bardet-Biedl syndrome), full gene sequence

BBS2 (Bardet-Biedl syndrome 2) (eg, Bardet-Biedl syndrome), full gene sequence

BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (eg, maple syrup urine disease, type 1B), full gene sequence

BEST1 (bestrophin 1) (eg, vitelliform macular dystrophy), full gene sequence

BMPR2 (bone morphogenetic protein receptor, type II [serine/threonine kinase]) (eg, heritable pulmonary arterial hypertension), full gene sequence

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

BSCL2 (Berardinelli-Seip congenital lipodystrophy 2 [seipin]) (eg, Berardinelli-Seip congenital lipodystrophy), full gene sequence

BTK (Bruton agammaglobulinemia tyrosine kinase) (eg, X-linked agammaglobulinemia), full gene sequence

CACNB2 (calcium channel, voltage-dependent, beta 2 subunit) (eg, Brugada syndrome), full gene sequence

CAPN3 (~~Calpain~~calpain 3) (eg, limb-girdle muscular dystrophy [LGMD] type 2A, calpainopathy), 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

CLCN1 (chloride channel 1, skeletal muscle) (eg, myotonia congenita), full gene sequence

CLCNKB (chloride channel, voltage-sensitive Kb) (eg, Bartter syndrome 3 and 4b), full gene sequence

CNTNAP2 (contactin-associated protein-like 2) (eg, Pitt-Hopkins-like syndrome 1), full gene sequence

COL6A2 (collagen, type VI, alpha 2) (eg, collagen type VI-related disorders), duplication/deletion analysis

CPT1A (carnitine palmitoyltransferase 1A [liver]) (eg, carnitine palmitoyltransferase 1A [CPT1A] deficiency), full gene sequence

CRB1 (crumbs homolog 1 [Drosophila]) (eg, Leber congenital amaurosis), full gene sequence

CREBBP (CREB binding protein) (eg, Rubinstein-Taybi syndrome), duplication/deletion analysis

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

(Do not report analyte-specific molecular pathology procedures separately when the specific analytes are included as part of the cytogenomic microarray analysis for neoplasia)

(Do not report 88271 when performing cytogenomic microarray analysis)

DBT (dihydrolipoamide branched chain transacylase E2) (eg, maple syrup urine disease, type 2), full gene sequence

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

DSC2 (desmocollin) (eg, arrhythmogenic right ventricular dysplasia/cardiomyopathy 11), full gene sequence

DSG2 (desmoglein 2) (eg, arrhythmogenic right ventricular dysplasia/cardiomyopathy 10), full gene sequence

DSP (desmoplakin) (eg, arrhythmogenic right ventricular dysplasia/cardiomyopathy 8), full gene sequence

EFHC1 (EF-hand domain [C-terminal] containing 1) (eg, juvenile myoclonic epilepsy), full gene sequence

EIF2B3 (eukaryotic translation initiation factor 2B, subunit 3 gamma, 58kDa) (eg, leukoencephalopathy with vanishing white matter), full gene sequence

EIF2B4 (eukaryotic translation initiation factor 2B, subunit 4 delta, 67kDa) (eg, leukoencephalopathy with vanishing white matter), full gene sequence

EIF2B5 (eukaryotic translation initiation factor 2B, subunit 5 epsilon, 82kDa) (eg, childhood ataxia with central nervous system hypomyelination/vanishing white matter), full gene sequence

ENG (endoglin) (eg, hereditary hemorrhagic telangiectasia, type 1), 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

FAH (fumarylacetoacetate hydrolase [fumarylacetoacetase]) (eg, tyrosinemia, type 1), full gene sequence

FASTKD2 (FAST kinase domains 2) (eg, mitochondrial respiratory chain complex IV deficiency), full gene sequence

FIG4 (FIG4 homolog, SAC1 lipid phosphatase domain containing [S. cerevisiae]) (eg, Charcot-Marie-Tooth disease), full gene sequence

FTSJ1 (FtsJ RNA methyltransferase homolog 1 [E. coli]) (eg, X-linked mental retardation 9), full gene sequence

FUS (fused in sarcoma) (eg, amyotrophic lateral sclerosis), full gene sequence

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

GALC (galactosylceramidase) (eg, Krabbe disease), full gene sequence

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

GARS (glycyl-tRNA synthetase) (eg, Charcot-Marie-Tooth disease), 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

GLUD1 (glutamate dehydrogenase 1) (eg, familial hyperinsulinism), full gene sequence

GNE (glucosamine [UDP-N-acetyl]-2-epimerase/N-acetylmannosamine kinase) (eg, inclusion body myopathy 2 [IBM2], Nonaka myopathy), full gene sequence

GRN (granulin) (eg, frontotemporal dementia), 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

HADHB (hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase [trifunctional protein], beta subunit) (eg, trifunctional protein deficiency), full gene sequence

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

HLCS (HLCS holocarboxylase synthetase) (eg, holocarboxylase synthetase deficiency), full gene sequence

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

IDUA (iduronidase, alpha-L-) (eg, mucopolysaccharidosis type I), full gene sequence

INF2 (inverted formin, FH2 and WH2 domain containing) (eg, focal segmental glomerulosclerosis), full gene sequence

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

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

JUP (junction plakoglobin) (eg, arrhythmogenic right ventricular dysplasia/cardiomyopathy 11), full gene sequence

KAL1 (Kallmann syndrome 1 sequence) (eg, Kallmann syndrome), full gene sequence

KCNH2 (potassium voltage-gated channel, subfamily H [eag-related], member 2) (eg, short QT syndrome, long QT syndrome), full gene sequence

(Do not report 81406 for KCNH2 full gene sequence in conjunction with 81280)

KCNQ1 (potassium voltage-gated channel, KQT-like subfamily, member 1) (eg, short QT syndrome, long QT syndrome), full gene sequence

(Do not report 81406 for KCNQ1 full gene sequence with 81280)

KCNQ2 (potassium voltage-gated channel, KQT-like subfamily, member 2) (eg, epileptic encephalopathy), full gene sequence

LDB3 (LIM domain binding 3) (eg, familial dilated cardiomyopathy, myofibrillar myopathy), full gene sequence

LDLR (low density lipoprotein receptor) (eg, familial hypercholesterolemia), full gene sequence

LEPR (leptin receptor) (eg, obesity with hypogonadism), full gene sequence

LHCGR (luteinizing hormone/choriogonadotropin receptor) (eg, precocious male puberty), 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

LRP5 (low density lipoprotein receptor-related protein 5) (eg, osteopetrosis), 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

MAPT (microtubule-associated protein tau) (eg, frontotemporal dementia), full gene sequence

MCCC1 (methylcrotonoyl-CoA carboxylase 1 [alpha]) (eg, 3-methylcrotonyl-CoA carboxylase deficiency), full gene sequence

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

MFN2 (mitofusin 2) (eg, Charcot-Marie-Tooth disease), full gene sequence

MTM1 (myotubularin 1) (eg, X-linked centronuclear myopathy), full gene sequence

MUT (methylmalonyl CoA mutase) (eg, methylmalonic acidemia), full gene sequence

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

NDUFS1 (NADH dehydrogenase [ubiquinone] Fe-S protein 1, 75kDa [NADH-coenzyme Q reductase]) (eg, Leigh syndrome, mitochondrial complex I deficiency), 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)

NPC1 (Niemann-Pick disease, type C1) (eg, Niemann-Pick disease), full gene sequence

NPHP1 (nephronophthisis 1 [juvenile]) (eg, Joubert syndrome), full gene sequence

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

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

OPTN (optineurin) (eg, amyotrophic lateral sclerosis), full gene sequence

PAFAH1B1 (platelet-activating factor acetylhydrolase 1b, regulatory subunit 1 [45kDa]) (eg, lissencephaly, Miller-Dieker syndrome), full gene sequence

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

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

PARK2 (Parkinson protein 2, E3 ubiquitin protein ligase [parkin]) (eg, Parkinson disease), full gene sequence

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

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

PCCA (propionyl CoA carboxylase, alpha polypeptide) (eg, propionic acidemia, type 1), full gene sequence

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

PCDH15 (protocadherin-related 15) (eg, Usher syndrome type 1F), duplication/deletion analysis

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

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

PHEX (phosphate-regulating endopeptidase homolog, X-linked) (eg, hypophosphatemic rickets), full gene sequence

PKD2 (polycystic kidney disease 2 [autosomal dominant]) (eg, polycystic kidney disease), full gene sequence

PKP2 (plakophilin 2) (eg, arrhythmogenic right ventricular dysplasia/cardiomyopathy 9), full gene sequence

PNKD (eg, paroxysmal nonkinesigenic dyskinesia) (eg, paroxysmal nonkinesigenic dyskinesia), full gene sequence

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

POMGNT1 (protein O-linked mannose beta1,2-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

PRKCG (protein kinase C, gamma) (eg, spinocerebellar ataxia), 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

RPE65 (retinal pigment epithelium-specific protein 65kDa) (eg, retinitis pigmentosa, Leber congenital amaurosis), full gene sequence

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

SCN4A (sodium channel, voltage-gated, type IV, alpha subunit) (eg, hyperkalemic periodic paralysis), full gene sequence

SCNN1A (sodium channel, nonvoltage-gated 1 alpha) (eg, pseudohypoaldosteronism), full gene sequence

SCNN1B (sodium channel, nonvoltage-gated 1, beta) (eg, Liddle syndrome, pseudohypoaldosteronism), full gene sequence

SCNN1G (sodium channel, nonvoltage-gated 1, gamma) (eg, Liddle syndrome, pseudohypoaldosteronism), full gene sequence

SDHA (succinate dehydrogenase complex, subunit A, flavoprotein [Fp]) (eg, Leigh syndrome, mitochondrial complex II deficiency), full gene sequence

SETX (senataxin) (eg, ataxia), full gene sequence

SGCE (sarcoglycan, epsilon) (eg, myoclonic dystonia), full gene sequence

SH3TC2 (SH3 domain and tetratricopeptide repeats 2) (eg, Charcot-Marie-Tooth disease), full gene sequence

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

SLC37A4 (solute carrier family 37 [glucose-6-phosphate transporter], member 4) (eg, glycogen storage disease type Ib), full gene sequence

SMAD4 (SMAD family member 4) (eg, hemorrhagic telangiectasia syndrome, juvenile polyposis), full gene sequence

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

SPAST (spastin) (eg, spastic paraplegia), full gene sequence

SPG7 (spastic paraplegia 7 [pure and complicated autosomal recessive]) (eg, spastic paraplegia), full gene sequence

STXBP1 (syntaxin-binding protein 1) (eg, epileptic encephalopathy), full gene sequence

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

TCF4 (transcription factor 4) (eg, Pitt-Hopkins syndrome), full gene sequence

TH (tyrosine hydroxylase) (eg, Segawa syndrome), full gene sequence

TMEM43 (transmembrane protein 43) (eg, arrhythmogenic right ventricular cardiomyopathy), full gene sequence

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

TRPC6 (transient receptor potential cation channel, subfamily C, member 6) (eg, focal segmental glomerulosclerosis), 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

UMOD (uromodulin) (eg, glomerulocystic kidney disease with hyperuricemia and isosthenuria), full gene sequence

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

WAS (Wiskott-Aldrich syndrome [eczema-thrombocytopenia]) (eg, Wiskott-Aldrich syndrome), full gene sequence

▲ **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

AGL (amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase) (eg, glycogen storage disease type III), full gene sequence

AHI1 (Abelson helper integration site 1) (eg, Joubert syndrome), full gene sequence

ASPM (asp [abnormal spindle] homolog, microcephaly associated [Drosophila]) (eg, primary microcephaly), full gene sequence

CACNA1A (calcium channel, voltage-dependent, P/Q type, alpha 1A subunit) (eg, familial hemiplegic migraine), full gene sequence

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

COL4A4 (collagen, type IV, alpha 4) (eg, Alport syndrome), full gene sequence

COL4A5 (collagen, type IV, alpha 5) (eg, Alport syndrome), duplication/deletion analysis

COL6A1 (collagen, type VI, alpha 1) (eg, collagen type VI-related disorders), full gene sequence

COL6A2 (collagen, type VI, alpha 2) (eg, collagen type VI-related disorders), full gene sequence

COL6A3 (collagen, type VI, alpha 3) (eg, collagen type VI-related disorders), full gene sequence

CREBBP (CREB binding protein) (eg, Rubinstein-Taybi syndrome), full gene sequence

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

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

KDM5C (lysine [K]-specific demethylase 5C) (eg, X-linked mental retardation), full gene sequence

KIAA0196 (KIAA0196) (eg, spastic paraplegia), full gene sequence

L1CAM (L1 cell adhesion molecule) (eg, MASA syndrome, X-linked hydrocephaly), full gene sequence

LAMB2 (laminin, beta 2 [laminin S]) (eg, Pierson 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

MYO7A (myosin VIIA) (eg, Usher syndrome, type 1), full gene sequence

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

NPHS1 (nephrosis 1, congenital, Finnish type [nephrin]) (eg, congenital Finnish nephrosis), 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

PKD1 (polycystic kidney disease 1 [autosomal dominant]) (eg, polycystic kidney disease), full gene sequence

PLCE1 (phospholipase C, epsilon 1) (eg, nephrotic syndrome type 3), 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

SLC12A1 (solute carrier family 12 [sodium/potassium/chloride transporters], member 1) (eg, Bartter syndrome), full gene sequence

SLC12A3 (solute carrier family 12 [sodium/chloride transporters], member 3) (eg, Gitelman syndrome), full gene sequence

SPG11 (spastic paraplegia 11 [autosomal recessive]) (eg, spastic paraplegia), full gene sequence

SPTBN2 (spectrin, beta, non-erythrocytic 2) (eg, spinocerebellar ataxia), full gene sequence

TMEM67 (transmembrane protein 67) (eg, Joubert syndrome), 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

VPS13B (vacuolar protein sorting 13 homolog B [yeast]) (eg, Cohen syndrome), duplication/deletion analysis

WDR62 (WD repeat domain 62) (eg, primary autosomal recessive microcephaly), full gene sequence

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

ABCA4 (ATP-binding cassette, sub-family A [ABC1], member 4) (eg, Stargardt disease, age-related macular degeneration), full gene sequence

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

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

CEP290 (centrosomal protein 290kDa) (eg, Joubert syndrome), full gene sequence

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

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

COL4A1 (collagen, type IV, alpha 1) (eg, brain small-vessel disease with hemorrhage), full gene sequence

COL4A3 (collagen, type IV, alpha 3 [Goodpasture antigen]) (eg, Alport syndrome), full gene sequence

COL4A5 (collagen, type IV, alpha 5) (eg, Alport syndrome), full gene sequence

DMD (dystrophin) (eg, Duchenne/Becker muscular dystrophy), 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

ITPR1 (inositol 1,4,5-trisphosphate receptor, type 1) (eg, spinocerebellar ataxia), full gene sequence

LAMA2 (laminin, alpha 2) (eg, congenital muscular dystrophy), full gene sequence

LRRK2 (leucine-rich repeat kinase 2) (eg, Parkinson disease), full gene sequence

MYH11 (myosin, heavy chain 11, smooth muscle) (eg, thoracic aortic aneurysms and aortic dissections), full gene sequence

NEB (nebulin) (eg, nemaline myopathy 2), full gene sequence

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

PKHD1 (polycystic kidney and hepatic disease 1) (eg, autosomal recessive polycystic kidney disease), full gene sequence

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

RYR2 (ryanodine receptor 2 [cardiac]) (eg, catecholaminergic polymorphic ventricular tachycardia, arrhythmogenic right ventricular dysplasia), full gene sequence or targeted sequence analysis of > 50 exons

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

VPS13B (vacuolar protein sorting 13 homolog B [yeast]) (eg, Cohen syndrome), full gene sequence

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

● **81504** Code added

● **81507** Code added

▲ **84112** ~~Placental~~Evaluation of cervicovaginal fluid for specific amniotic fluid protein(s) (eg, placental alpha microglobulin-1 ~~(PAMG-1)~~ [PAMG-1], ~~cervicovaginal secretion~~placental protein 12 [PP12], alpha-fetoprotein), qualitative, each specimen;

▲ **87498** enterovirus, amplified probe technique, includes reverse transcription ~~and amplified probe technique~~when performed

▲ **87521** hepatitis C, amplified probe technique, includes reverse transcription ~~and amplified probe technique~~when performed

▲ **87522** hepatitis C, quantification, includes reverse transcription ~~and quantification~~when performed

▲ **87535** HIV-1, amplified probe technique, includes reverse transcription ~~and amplified probe technique~~when performed

▲ **87536** HIV-1, quantification, includes reverse transcription ~~and quantification~~when performed

▲ **87538** HIV-2, amplified probe technique, includes reverse transcription ~~and amplified probe technique~~when performed

▲ **87539** HIV-2, quantification, includes reverse transcription ~~and quantification~~when performed

● **87661** Code added

▲ **88342** Immunohistochemistry ~~(including tissue immunoperoxidase)~~or immunocytochemistry, each separately identifiable antibody per block, cytologic preparation, or hematologic smear; first separately identifiable antibody per slide

✚● **88343** Code added

Medicine

#● **90673** Code added

● **90685** Code added

● **90686** Code added

● **90687** Code added

● **90688** Code added

▲ **91065** Breath hydrogen or methane test (eg, for detection of lactase deficiency, fructose intolerance, bacterial overgrowth, or oro-cecal gastrointestinal transit);

92506 ~~Evaluation of speech, language, voice, communication, and/or auditory processing;~~

● **92521** Code added

● **92522** Code added

● **92523** Code added

● **92524** Code added

⊙● **93582** Code added

⊙● **93583** Code added

⊙▲ **93653** Comprehensive electrophysiologic evaluation including insertion and repositioning of multiple electrode catheters with induction or attempted induction of an arrhythmia with right atrial pacing and recording, right ventricular pacing and recording (when necessary), and His bundle recording (when necessary) with intracardiac catheter ablation of arrhythmogenic focus; with treatment of supraventricular tachycardia by ablation of fast or slow atrioventricular pathway, accessory atrioventricular connection, cavo-tricuspid isthmus or other single atrial focus or source of atrial re-entry

⊙▲ **93654** with treatment of ventricular tachycardia or focus of ventricular ectopy including intracardiac electrophysiologic 3D mapping, when performed, and left ventricular pacing and recording, when performed

⊙▲ **93656** Comprehensive electrophysiologic evaluation including transseptal catheterizations, insertion and repositioning of multiple electrode catheters with induction or attempted induction of an arrhythmia ~~with atrial recording and~~including left or right atrial pacing;/recording when ~~possible~~necessary, right ventricular pacing/recording when necessary and ~~recording,~~ His bundle recording when necessary with intracardiac catheter ablation ~~of arrhythmogenic focus, with treatment~~ of atrial fibrillation ~~by ablation~~ by pulmonary vein isolation;

● **94669** Code added

● **97610** Code added

▲ **99170** Anogenital examination ~~with colposcopic magnification,~~ magnified, in childhood for suspected trauma, including image recording when performed;

Category II Codes

- ● **0580F** Code added
- ● **0581F** Code added
- ● **0582F** Code added
- ● **0583F** Code added
- ● **0584F** Code added
- ▲ **1040F** DSM-~~IV~~ V™ criteria for major depressive disorder documented at the initial evaluation (MDD, MDD ADOL)[1]
- ● **1500F** Code added
- ● **1501F** Code added
- ● **1502F** Code added
- ● **1503F** Code added
- ● **1504F** Code added
- ● **1505F** Code added
- ● **3751F** Code added
- ● **3752F** Code added
- ● **3753F** Code added
- ● **3754F** Code added
- ● **3755F** Code added
- ● **3756F** Code added
- ● **3757F** Code added
- ● **3758F** Code added
- ● **3759F** Code added
- ● **3760F** Code added
- ● **3761F** Code added
- ● **3762F** Code added
- ● **3763F** Code added
- ● **4540F** Code added
- ● **4541F** Code added
- ● **4550F** Code added
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- ● **4560F** Code added
- ● **4561F** Code added
- ● **4562F** Code added
- ● **4563F** Code added
- ● **9001F** Code added
- ● **9002F** Code added
- ● **9003F** Code added
- ● **9004F** Code added
- ● **9005F** Code added
- ● **9006F** Code added
- ● **9007F** Code added

Category III Codes

- **~~0078T~~** ~~Endovascular repair using prosthesis of abdominal aortic aneurysm, pseudoaneurysm or dissection, abdominal aorta involving visceral branches (superior mesenteric, celiac and/or renal artery[s]);~~
- **~~0079T~~** ~~Placement of visceral extension prosthesis for endovascular repair of abdominal aortic aneurysm involving visceral vessels, each visceral branch (List separately in addition to code for primary procedure);~~
- **~~0080T~~** ~~Endovascular repair using prosthesis of abdominal aortic aneurysm, pseudoaneurysm or dissection, abdominal aorta involving visceral vessels (superior mesenteric, celiac and/or renal artery[s]), radiological supervision and interpretation;~~
- **~~0081T~~** ~~Placement of visceral extension prosthesis for endovascular repair of abdominal aortic aneurysm involving visceral vessels, each visceral branch, radiological supervision and interpretation (List separately in addition to code for primary procedure);~~
- **~~0124T~~** ~~Conjunctival incision with posterior extrascleral placement of pharmacological agent (does not include supply of medication);~~
- **~~0183T~~** ~~Low frequency, non-contact, non-thermal ultrasound, including topical application(s), when performed, wound assessment, and instruction(s) for ongoing care, per day;~~
- **~~0185T~~** ~~Multivariate analysis of patient-specific findings with quantifiable computer probability assessment, including report;~~
- **~~0186T~~** ~~Suprachoroidal delivery of pharmacologic agent (does not include supply of medication);~~
- **~~0192T~~** ~~external approach~~
- **~~0260T~~** ~~Total body systemic hypothermia, per day, in the neonate 28 days of age or younger;~~
- **~~0261T~~** ~~Selective head hypothermia, per day, in the neonate 28 days of age or younger;~~
- **~~0318T~~** ~~Implantation of catheter-delivered prosthetic aortic heart valve, open thoracic approach, (eg, transapical, other than transaortic);~~
- ● **0319T** Code added

- **0320T** Code added
- **0321T** Code added
- **0322T** Code added
- **0323T** Code added
- **0324T** Code added
- **0325T** Code added
- **0326T** Code added
- **0327T** Code added
- **0328T** Code added
- **0329T** Code added
- **0330T** Code added
- **0331T** Code added
- **0332T** Code added
- **0333T** Code added
- **0334T** Code added
- ⊙ **0335T** Code added
- **0336T** Code added
- **0337T** Code added
- **0338T** Code added
- **0339T** Code added