Approved Laboratory Developed Tests by Test Name or by Gene(s) Tested

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GENE	EFFECTIVE DATE	COVERED FOR THE FOLLOWING INDICATIONS	CPT CODE(S)
ALK	Jan. 1, 2013	To determine response to Tyrosine Kinase Inhibitor (TKI) therapy in patients with adenocarcinoma of the lung or mixed lung cancer with adenocarcinoma component of the lung.	88271 88291
APC	Jan. 1, 2013	• Testing for APC variants in individuals with clinical symptoms consistent with: o Familial Adenomatous Polyposis (FAP) o Attenuated Familial Adenomatous Polyposis (AFAP). o Turcot's or Gardner's syndromes. • Testing individuals with an APC-associated polyposis syndrome for the purpose of identifying a variant that may be used to screen at-risk relatives. • For the presymptomatic testing of at-risk relatives for a known familial variant.	81201 81202 81203
ATXN1	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 1 (SCA1) in patients with cerebellar ataxia of unknown etiology, along with extracerebellar symptoms associated with SCA1 and/or a family history consistent with autosomal dominant inheritance. • Diagnosis of SCA1 in symptomatic family members of known SCA1 patients.	81401
ATXN2	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 2 (SCA2) in patients with cerebellar ataxia of unknown etiology, along with extracerebellar symptoms associated with SCA2 and/or a family history consistent with autosomal dominant inheritance. • Diagnosis of SCA2 in symptomatic family members of known SCA2 patients.	81401
ATXN3	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 3 (SCA3) in patients with cerebellar ataxia of unknown etiology, along with extracerebellar symptoms associated with SCA3 and/or a family history consistent with autosomal dominant inheritance. • Diagnosis of SCA3 in symptomatic family members of known SCA3 patients.	81401
ATXN7	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 7 (SCA7) in patients with cerebellar ataxia and visual disturbance. • Diagnosis of SCA7 in symptomatic family members of known SCA7 patients.	81401
ATXN10	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 10 (SCA10) in ataxia patients whose ancestry is of American Indian origin, and whose family history is consistent with autosomal dominant inheritance. • Diagnosis of SCA10 in symptomatic family members of known SCA10 patients.	81401

GENE	EFFECTIVE DATE	COVERED FOR THE FOLLOWING INDICATIONS	CPT CODE(S)
BCR/ABL1	Jan. 1, 2013	• Diagnostic assessment of individuals with suspected Chronic Myelogenous Leukemia (CML) by quantitative RT-PCR (RQ-PCR) • Diagnostic assessment of individuals with suspected CML by qualitative RT-PCR. • Monitoring response to TKI therapy, such as imatinib, in individuals with CML by RQPCR. • Testing for the presence of the following in CML patients to guide treatment selection following resistance to first-line imatinib therapy o BCR/ABL1 p.Thr315Ile o BCR/ABL1 variants other than p.Thr315Ile	81170 81206 81207 81208
BMPR1A	Jan. 1, 2013	• To clarify the diagnosis of individuals with Juvenile Polyposis Syndrome. • If a known SMAD4 mutation is in the family, genetic testing should be performed in the first six months of life due to hereditary hemorrhagic telangiectasia risk.	81479
BRAF	Jan. 1, 2013	• To predict response to: o vemurafenib therapy in patients with a positive cobas 4800 BRAF mutation test result. o trametinib monotherapy in advanced melanoma patients with a positive BRAF p.Val600Glu and/or p.Val600Lys test result. o dabrafenib monotherapy in advanced melanoma patients with a positive BRAF p.Val600Glu test result. o trametinib and dabrafenib combination therapy in advanced melanoma patients with a positive BRAF p.Val600Glu and/or p.Val600Lys test result. • For individuals with indeterminate thyroid Fine-Needle Aspiration biopsy cytology for diagnosis of papillary thyroid carcinoma.	81210

GENE	EFFECTIVE DATE	COVERED FOR THE FOLLOWING INDICATIONS	CPT CODE(S)
BCR/ABL1	Jan. 1, 2013	• For individuals from families transmitting a known BRCA1/2 variant. • For individuals with a history of breast cancer and at least one of the following: o Breast cancer diagnosed ≤ 45 years of age. o Breast cancer diagnosed ≤ 50 years of age and a close family member with breast cancer. o Two breast primaries with one diagnosed at or before age 50. o A diagnosis of triple negative breast cancer at or before age 60. o Breast cancer diagnosed at any age and at least one close relative with breast cancer before age 50 and/or epithelial ovarian cancer at any age. o Breast cancer diagnosed at any age and at least two close relatives diagnosed with breast, pancreatic, and/or prostate (Gleason ≥ 7) cancer at any age. o A close male relative, which includes first-, second-, and third-degree relatives, with breast cancer. o An ethnic background associated with a higher frequency of BRCA1/2 variants (i.e., Ashkenazi Jewish). • For individuals with a personal history of epithelial ovarian cancer. • For individuals with male breast cancer. • For individuals with a personal history of pancreatic cancer or prostate (Gleason ≥ 7) and at least two close relatives with breast, ovarian, prostate (Gleason ≥ 7), and/or pancreatic cancer. • For unaffected individuals (with no personal history of cancer) who have one of the following:	81162 81211 81212 81213 81214 81215 81216 81217
CACNA1A	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 6 (SCA6) in patients with cerebellar ataxia with dysarthria and/or nystagmus. • Diagnosis of SCA6 in symptomatic family members of known SCA6 patients.	81401
CALM1, CASQ2, RYR2, and TRDN	Jan. 1, 2013	To confirm a diagnosis of Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) in patients with clinically diagnosed or suspected CPVT.	81405 81408 81479
CDH1	Jan. 1, 2013	For large rearrangements in the CDH1 gene for the treatment of Hereditary Diffuse Gastric Cancer.	81406
CEBPA	Jan. 1, 2013	To guide the treatment decisions for individuals with Acute Myeloid Leukemia.	81218
Chromosome 22q11.2	Jan. 1, 2013	Confirmation of diagnosis in an individual suspected of chromosome 22q11.2 deletion syndrome based on clinical findings.	88271 88291

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CFTR	Jan. 1, 2013	• Confirmation of diagnosis in individuals showing clinical symptoms of Cystic Fibrosis (CF) or having a high sweat chloride level. • Identification of newborns who are affected with CF. • Identification of individuals with the p.Gly551Asp variant who will respond to treatment with ivacaftor. • Male infertility testing and treatment. • Preconception and prenatal carrier screening in accordance with the most current ACOG guidelines.	81220 81221 81222 81223 81224
Chimerism Analysis	Jan. 1, 2013	For the management and treatment of stem cell transplant patients.	81265 81266 81267 81268
COL1A1/ COL1A2	Jan. 1, 2013	For sequence variants in the COL1A1/COL1A2 genes for the diagnosis of Osteogenesis Imperfecta (OI) when clinical and radiological examination and family history provide inadequate information for diagnosis of OI.	81408
COL3A1	Jan. 1, 2013	To confirm or establish a diagnosis of Ehlers-Danlos Syndrome Type 4 (EDS IV), also known as vascular EDS, in patients with clinical symptoms or features of EDS IV.	81479
CYP2C9	Jan. 1, 2013	For the initiation and management of warfarin treatment.	81227
CYP2C19	Jan. 1, 2013	To manage dosing of clopidogrel.	81225
Cytogenomic Constitutional Microarray Analysis	Jan. 1, 2013	• Diagnostic evaluation of patients suspected of having a genetic syndrome (i.e., have congenital anomalies, dysmorphic features, Developmental Delay (DD), and/or intellectual disability). • Diagnostic evaluation of individuals with Autism Spectrum Disorder (ASD), including autism, Asperger syndrome, and pervasive developmental disorder	81228 81229 81406
DAZ/SRY	Jan. 1, 2013	To detect submicroscopic deletions involving the Y chromosome in the evaluation of men with infertility secondary to azoospermia, oligozoospermia, or teratozoospermia.	81403
DMD	Nov. 20, 2014	For diagnostic DMD testing (deletion and duplication analysis with reflex to complete gene sequencing) in males or females exhibiting symptoms of Duchenne Muscular Dystrophy (DMD) or Becker Muscular Dystrophy (BMD).	81161 81408
DMPK	Jan. 1, 2013	• Confirmation of a diagnosis of Myotonic Dystrophy Type 1 (DM1) or Type 2 (DM2) in symptomatic patients. • Diagnosis of DM1 or DM2 in asymptomatic adults who are at an increased risk of DM1 or DM2 through a positive family history.	81401 81404

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DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, and TMEM43	Jan. 1, 2013	• For sequence variants in the DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, and TMEM43 genes to confirm a diagnosis of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C) in probands. • For a known familial sequence variant in the DSC2, DSG2, DSP, PKP2, or TMEM43 gene for at-risk relatives of probands with International Task Force (ITF)-confirmed ARVD/C to confirm a diagnosis of ARVD/C in those whose symptoms meet the ITF diagnostic criteria.	81406 81408
EGFR	Jan. 1, 2013	To help guide administration of Epidermal Growth Factor Receptor (EGFR) TKIs in the first-line treatment of non-small cell lung cancer.	81235
F2	Jan. 1, 2013	• Diagnostic evaluation of individuals with a prior Venous Thromboembolism (VTE) during pregnancy or puerperium. • For patients with VTE with a personal or family history of recurrent VTE (more than two in the same person). • For patients with their first VTE before age 50 with no precipitating factors. • For venous thrombosis at unusual sites such as the cerebral, mesenteric, portal, or hepatic veins. • For VTE associated with the use of estrogen-containing oral contraceptives, Selective Estrogen Receptor Modulators (SERMs), or Hormone Replacement Therapy (HRT). • To diagnose an inherited thrombophilia in female family members of individuals with an inherited thrombophilia if the female family member is pregnant or considering pregnancy or oral contraceptive use.	81240 81400
FBN1	Jan. 1, 2013	• To facilitate the diagnosis of Marfan syndrome in patients who do not fulfill the Ghent diagnostic criteria, but have at least one major feature of the condition. • To facilitate the diagnosis of Marfan syndrome in the at-risk relatives of patients carrying known disease- causing variants.	81408
FLCN	Jul. 31, 2014	To confirm a diagnosis of Birt-Hogg-Dubé Syndrome (BHD) in patients with suspected BHD.	81479
FLT3	Oct. 7, 2013	For diagnosis and prognosis in AML.	81245 81246

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F5	Jan. 1, 2013	• Diagnostic evaluation of individuals with a prior VTE during pregnancy or puerperium. • For patients with VTE with a personal or family history of recurrent VTE (more than two in the same person). • For patients with their first VTE before age 50 with no precipitating factors. • For venous thrombosis at unusual sites such as the cerebral, mesenteric, portal, or hepatic veins. • For VTE associated with the use of estrogen-containing oral contraceptives, Selective Estrogen Receptor Modulators (SERMs), or Hormone Replacement Therapy (HRT). • To diagnose an inherited thrombophilia in female family members of individuals with an inherited thrombophilia if the female family member is pregnant or considering pregnancy or oral contraceptive use.	81241 81400
FMR1	Jan. 1, 2013	• FMR1 gene testing is covered for testing for CGG repeat length for diagnosis of patients of either sex with mental retardation, intellectual disability, developmental delay, or autism. • FMR1 gene testing for Fragile X-Associated Tremor/Ataxia Syndrome is covered for the following individuals: o Males and females older than age 50 years who have progressive cerebellar ataxia and intention tremor with or without a positive family history of FMR1-related disorders in whom other common causes of ataxia have been excluded. o Women with unexplained Premature Ovarian Insufficiency (POI).	81243 81244
GCK	Jan. 1, 2013	Diagnosis of Maturity-Onset Diabetes of the Young Type 2 (MODY2) in patients with hyperglycemia or non-insulin-dependent diabetes who have a family history of abnormal glucose metabolism in at least two consecutive generations, with the patient or ≥ 1 family member(s) diagnosed before age 25.	81406
GJB2	Jan. 1, 2016	Diagnosis of DFNB1 or DFNA3 in individuals with nonsyndromic hearing loss to aid in treatment.	81252 81253
GJB6	Jan. 1, 2016	Diagnosis of DFNB1 or DFNA3 in individuals with nonsyndromic hearing loss to aid in treatment.	81254
HBA1/HBA2	Jan. 1, 2016	• To confirm the diagnosis of alpha-thalassemia in a symptomatic individual. • To confirm the diagnosis in a pregnant woman with low hemoglobin when alphathalassemia is suspected.	81257 81404 81405
HEXA	Jan. 1, 2013	As an adjunct to biochemical testing in patients with low hexosaminidase A levels in blood. When individuals are identified with apparent deficiency of hexosaminidase A enzymatic activity, targeted mutation analysis can then be used to distinguish pseudodeficiency alleles from disease-causing alleles.	81255 81406
HFE	Jan. 1, 2013	Diagnosis of patients with or without symptoms of iron overload with a serum transferrin saturation >45% and/or elevated serum ferritin.	81256

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HLA	Jan. 1, 2013	• To determine histocompatibility of tissue between organ and bone marrow donors and recipients prior to transplant. • For platelet transfusion for patients refractory to treatment due to alloimmunization. • Diagnosis of celiac disease in symptomatic patients with equivocal results on small bowel biopsy and serology, or in previously symptomatic patients who are asymptomatic while on a gluten-free diet. • Testing for the HLA-B*1502 allele prior to initiating treatment with carbamazepine in patients from high-risk ethnic groups. • Testing for the HLA-B*5701 allele for hypersensitivity reactions in patients prior to initiation or reinitiation with treatments containing abacavir. • Testing for the HLA-B*58:01 allele in patients prior to initiating treatment with allopurinol.	81370 81371 81372 81373 81374 81375 81376 81377 81378 81379 81380 81381 81382 81383
HNF1A	Jan. 1, 2013	Diagnosis of Maturity-Onset Diabetes of the Young Type 3 (MODY3) in patients with hyperglycemia or non- insulin-dependent diabetes who have a family history of abnormal glucose metabolism in at least two consecutive generations, with the patient or ≥ 1 family member(s) diagnosed before age 25.	81405
HTT	Jan. 1, 2013	To test for CAG repeat length for diagnosis of Huntington Chorea/Disease (HD) in patients suspected of having HD in the absence of a family history of HD.	81401
IGH	Jan. 1, 2013	• For medical management of patients with Acute Lymphoblastic Leukemia (ALL) through analysis of rearrangements in the IGH gene to estimate Minimal Residual Disease (MRD) levels. • For diagnostic evaluation of rearrangements in the IGH gene in patients with suspected B-cell Non- Hodgkin's Lymphoma (NHL), but in whom clinical, immunophenotypic, and histologic evaluation have provided inconclusive results.	81261 81262 81263
IGK	Jan. 1, 2013	• For medical management of patients with ALL through analysis of rearrangements in the IGK gene to estimate MRD levels. • For diagnostic evaluation of rearrangements in the IGK gene in patients with suspected B-cell NHL, but in whom clinical, immunophenotypic, and histologic evaluation have provided inconclusive results.	81264
JAK2	Jan. 1, 2013	• Diagnostic evaluation of individuals presenting with clinical, laboratory, or pathological findings suggesting classic forms of myeloproliferative neoplasms (MPN) • Diagnostic evaluation of PV through JAK2 Exon 12 variant detection in JAK2 p.Val617Phe negative individuals.	81270 81403

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KCNQ1, KCNH2, SCN5A, KCNE1, and KCNE2	Jan. 1, 2013	For patients with suspected familial Long QT Syndrome for confirmation of diagnosis and treatment.	81280 81281 81282
KIT	Jan. 1, 2013	• To confirm a diagnosis of a gastrointestinal stromal tumor (GIST) in patients who are negative by immunostaining. • To determine primary resistance to treatment with TKIs in patients with an advanced metastatic or unresectable GIST. • To determine primary resistance to preoperative or postoperative treatment of a GIST with TKIs.	81272 81273
KRAS	Jan. 1, 2013	To help guide administration of anti-EGFR monoclonal antibodies.	81275 81276
MECP2	Jan. 1, 2013	• Testing for MECP2 sequence variants in patients who: o Meet established clinical diagnostic criteria for classic or variant Rett Syndrome (RS). o Have symptoms of RS, but don't meet established clinical diagnostic criteria.	81302 81303 81304

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MLH1, MSH2, MSH6, MSI, PMS2, and EPCAM	Jan. 1, 2013	Genetic testing for Lynch syndrome is covered for a symptomatic or asymptomatic beneficiary who meets one of the following criteria: 1. Amsterdam II criteria for Lynch syndrome genetic testing. At least three relatives of the affected beneficiary must have a cancer associated with Lynch syndrome; and all of the following criteria must be present: • One must be a first-degree relative of the other two; • At least two successive generations must be affected; • At least one relative with cancer associated with Lynch syndrome should be diagnosed before age 50 years; • FAP should be excluded in the colorectal cancer case(s) (if any); and • Tumors should be verified whenever possible. 2. Revised Bethesda guidelines: • Colorectal cancer diagnosed in a beneficiary at less than 50 years old. • Presence of synchronous or metachronous Lynch syndrome-associated cancers, regardless of age. • Colorectal cancer with the MSI-H histology diagnosed in a beneficiary who is less than 60 years of age. • Colorectal cancer diagnosed in a beneficiary with one or more first-degree relatives with a Lynch syndrome- associated cancer, with one of the cancers being diagnosed under age 50 years. • Colorectal cancer diagnosed in a beneficiary with two or more first- or second-degree relatives with Lynch syndrome-associated cancers, regardless of age. 3. Has a known Lynch syndrome mutation in the family. 4. Endometrial cancer diagnosed in a beneficiary at less than 50 years old. 5. If any of the revised Bethesda guidelines are met, Microsatellite Instability (MSI) and/or Immunohistochemistry (IHC) testing on the colon cancer tissue may be clinically appropriate. If the tumor is MSI positive or mutation of one of the mismatch repair genes is indicated by failure of IHC staining, then genetic testing should be undertaken. Further unnecessary testing can often be avoided by performance of IHC prior to any MSI testing. • Genetic testing is covered for symptomatic or asymptomatic patients > 18 years of age who are at risk of having a known familial sequence variant in a Mismatch Repair (MMR) gene.	81288 81292 81293 81294 81295 81296 81297 81298 81299 81300 81301 81317 81318 81319 81403
MPL	Jan. 1, 2013	Diagnostic evaluation of Myeloproliferative Leukemia (MPL) variants to include Trp515Leu and Trp515Lys in JAK2 p.Val617Phe-negative individuals showing symptoms.	81402 81403
MUTYH	Jan. 1, 2013	• Diagnosis of MYH-Associated Polyposis (MAP) in APC-negative colorectal polyposis patients, or in polyposis patients who have a family history consistent with autosomal recessive inheritance. • Diagnosis of MAP in asymptomatic siblings of patients with known MYH variants.	81401 81406

GENE	EFFECTIVE DATE	COVERED FOR THE FOLLOWING INDICATIONS	CPT CODE(S)
Noninvasive Prenatal Screening for Trisomies 13, 18, 21, X & Y	Mar. 5, 2015	In singleton pregnancies with a high risk of fetal aneuploidy.	81420 81479 81507 81599
NPM1	Jan. 1, 2013	To guide treatment decisions for individuals with AML.	81310
NRAS	Oct. 3, 2014	For patients with metastatic colorectal cancer who are being considered for treatment with anti-EGFR monoclonal antibodies, and who have had negative KRAS gene testing.	81311
Oncotype DX® Breast Cancer Assay	Jan. 1, 2013	• Estrogen Receptor (ER) positive (+), lymph node (LN) negative (-), human EGFR 2 negative (HER2-) breast cancer patients who are considering whether to use adjuvant chemotherapy in addition to standard hormone therapy. • ER+, HER2- breast cancer patients with 1-3 involved ipsilateral axillary lymph nodes who are considering whether to use adjuvant chemotherapy in addition to hormonal therapy.	81519
PAX8	Jan. 1, 2013	For individuals with indeterminate thyroid FNA biopsy cytology for diagnosis of papillary thyroid carcinoma.	81401
PDGFRA	Jan. 1, 2013	• To confirm a diagnosis of a GIST in patients who are negative by immunostaining. • To determine primary resistance to treatment with TKIs in patients with an advanced metastatic or unresectable GIST. • To determine primary resistance to preoperative or postoperative treatment of a GIST with TKIs.	81314
PML/ RARalpha	Jan. 1, 2013	• Diagnostic assessment of individuals with suspected acute promyelocytic leukemia (APL) by quantitative RT- PCR (RQ-PCR). • Diagnostic assessment of individuals with suspected APL by qualitative RT-PCR. • Monitoring response to treatment and disease progression in individuals with APL by RQ-PCR.	81315 81316
PMP22	Jan. 1, 2013	For the accurate diagnosis and classification of hereditary polyneuropathies.	81324 81325 81326
PPP2R2B	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 12 (SCA12) in patients with action tremor of the upper extremities and signs of cerebellar and cortical dysfunction, in addition to Indian ancestry and a family history consistent with autosomal dominant inheritance. • Diagnosis of SCA12 in symptomatic family members of known SCA12 patients.	81401

GENE	EFFECTIVE DATE	COVERED FOR THE FOLLOWING INDICATIONS	CPT CODE(S)
PRSS1	Jan. 1, 2013	To confirm a diagnosis of hereditary pancreatitis in symptomatic patients with any of the following: • A family history of pancreatitis in a first-degree (parent, sibling, child) or second degree (aunt, uncle, grandparent) relative; • An unexplained episode of documented pancreatitis occurring in a child that has required hospitalization, and where there is significant concern that hereditary pancreatitis should be excluded; • Recurrent (two or more separate, documented episodes with hyper-amylasemia) attacks of acute pancreatitis for which there is no explanation (anatomical anomalies, ampullary or main pancreatic strictures, trauma, viral infection, gallstones, alcohol, drugs, hyperlipidemia, etc.); or • Unexplained (idiopathic) chronic pancreatitis.	81401
PTEN	Jan. 1, 2013	• For patients with ASDs and macrocephaly (Head circumference greater than 2 standard above the mean for age). • PTEN variant testing in individuals suspected of being affected with Cowden Syndrome (CS) or Bannayan- Riley-Ruvalcaba Syndrome.	81321 81322 81323
RET	Jan. 1, 2013	• Multiple endocrine neoplasia type 2 (MEN2) gene testing in patients with the clinical manifestations of MEN2A, MEN2B, or familial medullary thyroid carcinoma (FMTC), including those with apparently sporadic Medullary Thyroid Carcinoma (MTC) or pheochromocytoma. • MEN2 gene testing to confirm a diagnosis in the at-risk relatives of genetically confirmed MEN2 patients.	81404 81405
ROSI	Jan. 12, 2016	For patients who have wild type (negative) EGFR or ALK gene testing, reflex testing to ROS1 should be ordered for the treatment of non-small cell lung carcinoma.	88274
RYRI	Jan. 1, 2013	• To test clinically confirmed Malignant Hyperthermia Susceptibility (MHS) patients for variants in the RYR1 gene to facilitate diagnostic testing in at-risk relatives. • To diagnose MHS in at-risk relatives of patients with clinically confirmed MHS.	81408
SDHB	Jun. 16, 2014	To diagnose a hereditary paraganglioma (PGL) or pheochromocytoma (PCC) syndrome in patients with PGLs and/or PCCs.	81405
SDHD	Jun. 16, 2014	To diagnose a hereditary PGL or PCC syndrome in patients with PGLs and/or PCCs.	81404
SERPINA1	May 27, 2014	Diagnosis of inconclusive cases of Alpha-1 Antitrypsin Deficiency (AATD) in individuals with Chronic Obstructive Pulmonary Disease , unexplained liver disease, family history of AATD, or environmental exposures leading to airflow obstruction after serum Alpha-1 Ant	81332

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SMAD4	Jan. 1, 2013	• To clarify the diagnosis of individuals with JPS. • If a known SMAD4 mutation is in the family, genetic testing should be performed in the first six months of life due to hereditary hemorrhagic telangiectasia risk.	81405 81406
SMN1/SMN2	Jan. 1, 2013	Diagnosis of patients with hypotonia and muscle weakness who are suspected of having Spinal Muscular Atrophy (SMA).	81400 81401 81403 81405
SNRPN/UBE3A	Jan. 1, 2013	When a clinical diagnosis of Prader-Willi Syndrome (PWS) is suspected, the following findings justify genetic testing: • From birth to age two: Hypotonia with poor suck (neonatal period). • From age two to age six: Hypotonia with history of poor suck, global developmental delay. • From age six to age 12: Hypotonia with history of poor suck, global developmental delay, excessive eating with central obesity if uncontrolled. • From age 13 years to adulthood: Cognitive impairment, usually mild intellectual disability; excessive eating with central obesity if uncontrolled, hypothalamic hypogonadism and/or typical behavior problems. When a clinical diagnosis of Angelman Syndrome is suspected, the following findings justify genetic testing: • As part of the evaluation of patients with developmental delay, regardless of age. • As part of the evaluation of patients with a balance or movement disorder such as ataxia of gait. May not appear as frank ataxia but can be forward lurching unsteadiness, clumsiness, or quick, jerky motions. • As part of the evaluation of patients with uniqueness of behavior: any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with uplifted hand-flapping or waving movements; hypermotoric behavior. • Speech impairment, none or minimal use of words; receptive and non-verbal communication skills higher than verbal ones.	81331
STK11	Jan. 1, 2013	To confirm a diagnosis of Peutz-Jeghers Syndrome (PJS) in proband patients with a presumptive or probable diagnosis of PJS.	81404 81405
TBP	Jan. 1, 2013	• Diagnosis of Spinocerebellar Ataxia Type 17 (SCA17) in ataxia patients exhibiting variable combinations of cognitive decline, psychiatric disturbance, and movement disorders. • Diagnosis of SCA17 in symptomatic family members of known SCA17 patients. • Diagnosis of SCA17 in patients suspected of having Huntington Disease (HD) who have tested negative for a pathogenic variant in the HD gene.	81401

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TP53	Jan. 1, 2013	• Diagnosis of patients satisfying the criteria for classic Li-Fraumeni Syndrome (LFS) or Li- Fraumeni-Like Syndrome (LFLS), or the Chompret criteria for TP53 gene testing.	81404 81405
TRG	Jan. 1, 2013	Diagnosis and treatment of T-cell neoplasms.	81342
UPD	Jan. 1, 2013	For neonates, infants, children or adults symptomatic for Beckwith-Wiedermann Syndrome (BWS) to diagnose Uniparental Disomy (UPD) for chromosome 11.	81402
UGT1A1	Jan. 1, 2013	• Prior to irinotecan administration in patients with CRC to lower the starting dose of irinotecan in patients with the UGT1A1*28/UGT1A1*28 genotype. • Prior to irinotecan administration in patients with CRC to increase the starting dose of irinotecan in patients with the UGT1A1*1/UGT1A1*1 or UGT1A1*1/UGT1A1*28 genotypes.	81350
VHL	Jan. 1, 2013	• Diagnosis of Von Hippel-Lindau (VHL) syndrome in patients presenting with pheochromocytoma, paraganglioma, or central nervous system hemangioblastoma. • Confirmation of diagnosis in individuals with symptoms consistent with VHL syndrome.	81403 81404
VKORC1	Jan. 1, 2013	For the initiation and management of warfarin treatment.	81355