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	CPT Code	Generic Test Name	Test Description
1	81200	ASPA Gene	Test for leukodystrophies (Canavan disease), an autosomal recessive disease (most common in Ashkenazi Jewish population) with life expectancy into childhood; only supportive treatment.
2	81205	BCKHD gene	Test for Branched-Chain Ketoacid Dehydrogenase Deficiency or Maple Syrup Urine disease, an autosomal recessive disease. Patients must be diagnosed to be put on a special diet which allows them to have a healthy life and avoid the severe neurological damage associated with the disease.
3	81206	BRC/ABL1 Gene Major bp tested together with 81207	This test is used to quantify BCR-ABL1 transcript levels of the major and minor fusion transcripts. These BCR-ABL1 fusion transcripts are found in patients with Chronic Myelogenous Leukemia (CML) and Philadelphia-positive Acute Lymphocytic Leukemia (ALL). This quantitative test is used to monitor the response of patients to imatinib mesylate or other therapies.
4	81207	BRC/ABL1 Gene Minor tested together with 81206	This test is used to quantify BCR-ABL1 transcript levels of the major and minor fusion transcripts. These BCR-ABL1 fusion transcripts are found in patients with CML and Philadelphia-positive ALL. This quantitative test is used to monitor the response of patients to imatinib mesylate or other therapies.
5	81209	BLM Gene	Test for Bloom's disease, an autosomal recessive disorder. It is characterized by multiple breaks and rearrangements in the affected person's chromosomes. This DNA damage greatly increases the risk of developing multiple cancers early in life. Incidence is high in Ashkenazi Jewish population. Early and accurate diagnosis of Bloom Syndrome patients will help to get them into appropriate cancer screening protocols.
6	81210	BRAF Gene	Mutations in BRAF are frequently found in melanomas, colorectal cancer, lung cancer, ovarian cancer, and thyroid gland cancer. Studies have

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			shown that patients with BRAF mutations respond differently to some therapies. Testing allows identification of the appropriate patients for treatment.
7	81220	Cystic Fibrosis Common variants	Cystic Fibrosis (CF) is a common genetic disorder resulting in chronic pulmonary and gastrointestinal pancreatic disease. ACOG recommends CF screening of common mutations be offered to all couples seeking preconception or prenatal care.
8	81221	Cystic Fibrosis Known family variants	Testing family members of those previously diagnosed with CF
9	81222	Cystic Fibrosis Deletion/Duplication	Used to identify causal mutations when only a single common mutation or rare variant of CF are detected. (Previous tests with a screening assay for common mutations and variants followed by CFTR full gene sequence analysis revealed only heterozygosity –a single mutation)
10	81223	Cystic Fibrosis Full Gene Sequencing	Used to identify rare mutations in individuals suspected of having CF but where only a single common mutation or variant has been identified
11	81225	CYP2c19 Gene common variants	The cytochrome P450 (CYP450) enzymes catalyze the oxidation of many drugs and chemicals. Individual differences of cytochrome P450 activity can result in total absence of metabolism of certain drugs to ultrafast metabolism of drugs. This can lead to adverse drug reactions or a lack of therapeutic effect under standard therapy conditions. CYP2C19 is a gene within the family of the CYP450 superfamily. It metabolizes 15% of all prescribed drugs, such as clopidogrel (Plavix).
12	81226	CYP2d6 Gene common variants	The cytochrome P450 (CYP450) enzymes catalyze the oxidation of many drugs and chemicals. Individual differences of cytochrome P450 activity can result in total absence of metabolism of certain drugs to ultrafast metabolism of drugs. This can lead to adverse drug reactions or a lack of therapeutic effect under standard therapy conditions. CYP2D6 is a gene within the family of the CYP450 superfamily. It metabolizes 25% of all prescribed drugs, such as codeine, tricyclic antidepressants, classical antipsychotics, and β-blockers. Specific variants in this gene also influence the metabolism of the breast cancer drug, tamoxifen, in postmenopausal women.

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			Genetic variants of CYP2D6 can be used to predict the altered enzyme activity and address the potential effects of metabolized drugs.
13	81227	CYP2c9 Gene common variants	The presence of certain variants in the CYP2C9 gene can result in poor metabolizer phenotypes that are associated with lack of enzyme activity and drugs may be metabolized slowly or not at all. This results in increased concentrations of the drug with a reduced or absent therapeutic response and the potential for serious side effects. Warfarin metabolism is reduced by 30% to 50% by the *2 variant and 90% by the *3 variant. Individuals with at least one copy of *2 or *3 have an increased risk of bleeding compared to individuals without *2 or *3. A lower maintenance dose may be required.
14	81229	Cytogen Microarray Copy no.& snp	This test is wide clinical use in the postnatal population for patients with congenital anomalies. In addition, for patients with mental retardation and developmental delay, this technology greatly improves diagnosis. The increased detection rate not only improves patient care but is cost effective. Once the patient receives a definitive genetic diagnosis no additional testing is needed, and key family members also are risk can be identified and evaluated. This test is performed based on clinical signs and symptoms.
15	81240	F2 Gene	Many of the abnormalities that cause some patients to have an increased risk for thrombosis have been defined at the molecular level. A point mutation in the factor II (prothrombin) gene is the second most common cause of inherited thrombosis (after factor VLeiden) and accounts for up to 20% of inherited thrombophilia. Six percent to 8% of people with a first-time venous clot have this mutation. The mutation has been reported in patients with idiopathic portal vein thrombosis or cerebral vein thrombosis, in patients using oral contraceptives, and in pregnant patients with placental abruptions and fetal growth restrictions.
16	81241	F5 Gene	Details of how the blood coagulation system is regulated have become well understood in recent years. Many of the abnormalities that cause some patients to have an increased risk for thrombosis have been defined at the molecular level. There is a single mutation in the Factor V gene that is found in

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			10 to 20% of people with a first-time venous clot . Heterozygous carriers of this mutation have a four- to eightfold increased risk of thrombosis. Individuals homozygous for the mutation (ie, they have a copy of the mutation on each chromosome) carry an 80- to 100-fold risk of thrombosis. Beyond the risk for thrombophilia, this mutation has also been associated with an increased risk for recurrent pregnancy loss, severe pre-eclampsia, fetal growth retardation, stillbirth, and placental problems (infarction and abruption).
17	81242	FANCC Gene	Test for Fanconi Anemia, an autosomal recessive disorder more frequent in Ashkenazi Jewish population. Patients with FA are at a very high risk for developing leukemia at a young age and several types of solid tumors in adults. FA can result in bone marrow failure and hematopoietic malignancy. Early genetic diagnosis allows for screening bone marrow biopsies to guide treatment and for genetic counseling of this condition.
18	81243	FMR1 Gene	Somatic inactivation of this gene is the most common inherited change associated with developmental and/or intellectual disabilities in children, and several other adult onset conditions. This test has been in existence for more than 30 years and is well established as a reliable test for identifying affected patients and carrier females. Definitive diagnosis is essential for proper patient management.
19	81245	FLT3	FLT3 mutation detection testing is necessary for stratifying high and low risk acute myeloid leukemia. It is necessary after recurrence of leukemia on patients not initially screened for FLT3 mutations. At relapse in order to make a decision about whether stem cell transplantation is possible a number of risk factors need to be reviewed. These include duration of first complete remission, result of cytogenetic testing, as well as age and FLT3 mutation status
20	81250	C6PC Gene	Prevalent mutations in the glucose-6-phosphatase gene are the cause of glycogen storage disease Type 1a. Some infants who are untreated develop severe low blood sugar. Long-term complications of untreated GSD1a include short stature, osteoporosis, delayed puberty, kidney disease, liver disease, seizures, and mental retardation. This condition can be treated by making dietary changes and

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			maintaining normal levels of glucose to prevent hypoglycemia. Individuals who are treated can be expected to have normal growth and many live into adulthood If not diagnosed and managed well, these patients may need liver and/or kidney transplantation and suffer from other morbid conditions.
21	81251	GBA Gene	This test detects certain mutations associated with Gaucher's disease and others associated with Parkinsonism. Gaucher's disease can be treated with gene therapy and Parkinson's patients with GBA mutations respond differently to treatment.
22	81255	HEXA Gene	Tay-Sachs disease is an autosomal recessive lysosomal storage disorder that causes progressive neurological deterioration ranging in severity from forms with infantile onset to those with adult onset. Presently there is no treatment for Tay-Sachs disease. Anticonvulsant medicine may initially control seizures. Other supportive treatment includes proper nutrition and hydration and techniques to keep the airway open. Children may eventually need a feeding tube.
23	81256	HFE Gene	Hereditary hemochromatosis is a disorder of iron metabolism resulting in the accumulation of excess iron in a variety of organs. Necessary treatment is to remove excess iron from the body before there is any organ damage usually by phlebotomy.
24	81257	HBA1/HBA2 Gene	Alpha-Thalassemia is the most common inherited disorder of hemoglobin synthesis in the world. Treatments, when needed for moderate and severe forms of thalassemia include blood transfusions, iron chelation therapy and folic acid supplements.
25	81260	IKBKAP Gene	Riley-Day syndrome is a disorder that is characterized by absence of papillae of the tongue, decreased tearing, erythematous blotching of the skin, difficulties with swallowing, relative insensitivity to pain, and reduced life expectancy. Treatment of this syndrome remains preventative, symptomatic and supportive.
26	81261	IGH Gene Rearrange amp meth	81261 through 81264 as well as 81341and 81342 should be used in conjunction to diagnose B- and T cell malignancies and determine leukemia and lymphoma lineage. Once diagnosed, testing continues to support treatment by detecting minimal residual disease or recurrent disease.
27	81262	IGH Gene	81261 through 81264 as well as 81341and

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		rearrange dir probe	81342 should be used in conjunction to diagnose B- and T cell malignancies and determine leukemia and lymphoma lineage. Once diagnosed, testing continues to support treatment by detecting minimal residual disease or recurrent disease.
28	81263	IGH vari regional mutations	Determines the mutation status of IgVH gene in B lymphocytes, including those of CLL (chronic lymphocytic leukemia). The IgVH gene mutation status is one of the discriminators of clinical outcome in patients with CLL. The mutational status of the immunoglobulin genes expressed by CLL cells can be used to segregate patients into two subsets that have significantly different tendencies for disease progression. Patients with leukemic cells that express unmutated immunoglobulin heavy-chain variable region genes have a greater tendency for disease progression than those who have leukemic cells that express IgVH genes with less than 98% nucleic acid homology with their germ-line counterparts.
29	81264	IGK rearrange abn clonal pop	81261 through 81264 as well as 81341and 81342 should be used in conjunction to diagnose B- and T cell malignancies and determine leukemia and lymphoma lineage. Once diagnosed, testing continues to support treatment by detecting minimal residual disease or recurrent disease.
30	81265	Str markers specimen anal	Testing uses markers that can compare the patient and a comparative specimen for a variety of purposes. In transplant patients can differentiate donor from recipient and also can determine maternal cell contamination of fetal cells from amniocentesis. Necessary to determine whose cells and DNA the test results originate from.
31	81270	JAK2 Gene	Mutations in Janus kinase 2 (JAK2), and in particular JAK2 V617F, are common in Philadelphia chromosome-negative myeloproliferative neoplasms. JAK2 exon 12 mutation status is associated with erythrocytosis and atypical bone marrow morphology and may be used to differentiate reactive conditions from malignant erythrocytosis. In the past several years, JAK2 inhibitors have been rapidly developed as targeted therapies for myeloproliferative neoplasms and testing insures the right treatment.
32	81275	KRAS Gene	This assay is intended to aid in the identification of CRC patients for treatment with cetuximab

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			(Erbitux®), based on a K-ras "no mutation detected" test result. The presence of these mutations correlates with a lack of response for certain EGFR inhibitor cancer therapies in patients with metastatic colorectal cancer.
33	81290	MCOLN1 Gene	Mucolipidosis type IV is a neurodegenerative lysosomal storage disorder associated with growth and psychomotor retardation, as well as ophthalmologic abnormalities. There is no treatment for this disorder but the symptoms can be alleviated for example by corneal transplant.
34	81291	MTHFR Gene	High blood levels of homocysteine is a risk factor for cerebrovascular disease, cerebral vein thrombosis, coronary artery disease, myocardial infarction, and venous thrombosis. The levels of homocysteine in the serum are influenced by both genetic and environmental factors. One of the genetic factors involves point mutations in the MTHFR gene. Some variants of the MTHFR enzyme result in an elevation of serum homocysteine levels, especially in individuals with insufficient folate. Hyperhomocysteinemia has been found in women who have experienced two or more early pregnancy losses, placental infarction, and fetal growth retardation. Dietary folic acid supplementation before the fourth week of gestation is well documented in reducing the recurrence risk for open neural tube defects by approximately 75%. It may act by normalizing homocysteine levels.
35	81292	MLH1 Gene full sequence	Lynch syndrome, often called hereditary nonpolyposis colorectal cancer (HNPCC). It is an inherited disorder that increases the risk of many types of cancer, particularly colon cancer. People with Lynch syndrome also have an increased risk of cancers of the stomach, small intestine, liver, gallbladder ducts, upper urinary tract, brain, and skin. Identification of the mutations in **MLH1, MSH2 and MSH6** that cause Lynch syndrome allows the appropriate screening for early detection for CRC for these high risk patients.
36	81293	MLH1 Gene known variants	Follow up testing in family members of those who have been identified as having Lynch Syndrome and known mutations in MLH1, MSH2 or MSH6.
37	81295	MSH2 Gene full sequence	Lynch syndrome, often called hereditary nonpolyposis colorectal cancer (HNPCC). It is an

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			inherited disorder that increases the risk of many types of cancer, particularly colon cancer. People with Lynch syndrome also have an increased risk of cancers of the stomach, small intestine, liver, gallbladder ducts, upper urinary tract, brain, and skin. Identification of the mutations that cause Lynch syndrome allows the appropriate screening for CRC for these high risk patients.
38	81296	MSH2 Gene known variants	Follow up testing in family members of those who have been identified as having Lynch Syndrome and known mutations in MLH1, MSH2 or MSH6.
39	81298	MSH6 Gene full sequence	Lynch syndrome, often called hereditary nonpolyposis colorectal cancer (HNPCC). It is an inherited disorder that increases the risk of many types of cancer, particularly colon cancer. People with Lynch syndrome also have an increased risk of cancers of the stomach, small intestine, liver, gallbladder ducts, upper urinary tract, brain, and skin. Identification of the mutations that cause Lynch syndrome allows the appropriate screening for CRC for these high risk patients.
40	81299	MSH6 Gene known variants	Follow up testing in family members of those who have been identified as having Lynch Syndrome and known mutations in MLH1, MSH2 or MSH6.
41	81301	Microsatellite instability	This test identifies tumors with microsatellite instability. High frequency microsatellite instability (MSI-H) is associated with hereditary nonpolyposis colorectal cancer (HNPCC), but it is also found in 15% to 20% of sporadic colorectal cancers. The presence of MSI-H is associated with a more favorable prognosis. Determines treatment option strategies.
42	81302	MECP2 Gene Full sequence	Rett syndrome, a severe neurological disorder leads to regression of developmental behaviors and language skills. It is the second most common cause of mental retardation in females. Mutations in the gene for MECP2 are the most common cause of Rett syndrome. There is no cure but treatments and medications can controls some of the signs and symptoms.
43	81304	MECP2 Gene dup/delet variants	Testing for specific mutations in the MECP2 gene. Duplications cause a very severe form of the Rett syndrome disorder.
44	81310	NPM1 Gene	The NPM1 mutation is one of the most common recurring genetic lesions in acute myeloid leukemia

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			(AML). The most common mutation, insertion at nucleotide position 959 (exon 12), accounts for 90% to 95% of NPM1 mutations. NPM1 mutations in absence of FLT3-ITD identify a prognostically favorable subgroup.
45	81315	PML/RARALPHA common breakpoints	The translocation rearrangements found in the vast majority (>95%) of acute promyelocytic leukemia (APL) cases. Identification of the rearrangements is necessary to assist in the diagnosis of APL. Early diagnosis and treatment of acute promyelocytic leukemia is important because patients with APL may develop serious blood-clotting or bleeding problems.
46	81330	SMPD1 Gene common variants	Niemann-Pick disease is a lysosomal storage disorder that is characterized by failure to thrive. There is no specific treatment except to manage symptoms.
47	81331	SNRPN/UBE3a Gene	Testing to detect Angelman Syndrome which is a developmental disorder. FISH testing is primary and molecular testing is performed if FISH is negative.
48	81332	SERPINA1 Gene	α1-antitrypsin (AAT)deficiency is inherited and is associated with COPD (chronic obstructive pulmonary disease), early onset emphysema, unexplained liver disease among other diseases. The clinical expression can be highly variable. AAT deficiency has no cure, but treatments are available and based on the type of disease you develop.
49	81341	TRB@ Gene rearrange dir probe	81261 through 81264 as well as 81341and 81342 should be used in conjunction with to diagnose B- and T cell malignancies and determine leukemia and lymphoma lineage. Once diagnosed, testing continues to support treatment by detecting minimal residual disease or recurrent disease.
50	81342	TRG Gene rearrangement analysis	81261 through 81264 as well as 81341and 81342 should be used in conjunction with to diagnose B- and T cell malignancies and determine leukemia and lymphoma lineage. Once diagnosed, testing continues to support treatment by detecting minimal residual disease or recurrent disease.
51	81350	UGT1a1 Gene	Severe toxicity (eg, grade 4 neutropenia) is commonly observed in cancer patients receiving irinotecan who carry the UGT1A1*28 allele, also called TA. This test result will provide valuable information to physicians prior to initiating or modifying treatment or supplementing treatment with additional drugs.

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			Gilbert's syndrome is an inherited metabolic disorder leading to hyperbilirubinemia syndromes. Between 80% to 100% of Caucasian patients with Gilbert syndrome are reported to have either one or two copies of UGT1A1*28. Patients require lifelong treatment and possibly liver transplant.
52	81355	VIKORE1 Gene	The presence of specific mutations in the VKORC1 gene reduces the gene's expression and leads to combined deficiency of vitamin K-dependent coagulation factors type 2. The risk of bleeding complication during oral anticoagulation is high. Low-dosage warfarin treatment should be considered.
53	81370	HLA i and ii Typing lr	histocompatibility (human leukocyte antigen, or HLA) required testing for bone marrow and other transplants
54	81372	HLA i typing complete lr	histocompatibility (human leukocyte antigen, or HLA) required testing for bone marrow and other transplants
55	81373	HLA i typing 1 locus lr	histocompatibility (human leukocyte antigen, or HLA) - required testing for bone marrow and other transplants
56	81374	HLA i typing 1 antigen lr	Human Leukocyte Antigen (HLA) B27 is associated with many disorders, including ankylosing spondylitis (Marie-Strumpell disease). A patient with consistent clinical and radiographic findings who is B27 positive has a greater chance of having or developing ankylosing spondylitis than a negative patient.
57	81376	HLA ii typing 1 locus lr	histocompatibility (human leukocyte antigen, or HLA) - required testing for bone marrow and other transplants
58	81377	HLA ii typing 1 ag equiv lr	Associated with celiac disease and sometimes used to support this diagnosis.
59	81380	HLA i typing 1 locus lr	histocompatibility (human leukocyte antigen, or HLA) - required testing for bone marrow and other transplants
60	81381	HLA i typing 1 allele hr	Abacavir hypersensitivity has been shown to be associated with the HLAB*57:91 of the major histocompatibility complex (MHC). The presence of HLA-B*57:91 allele increases the susceptibility to abacavir hypersensitivity in several populations studied.
61	81382	HLA ii typing 1 loc hr	histocompatibility (human leukocyte antigen, or HLA) - required testing for bone marrow and other transplants
62	81383	HLA ii typing 1	histocompatibility (human leukocyte antigen, or HLA)

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		allele hr	- required testing for bone marrow and other transplants
		Mopath procedures	Each of the levels is comprised of many tests that haven't been assigned individual CPT codes. The levels and tests within each level are grouped by method. The tests are similar in purpose to those described above. Mutation detection for diagnosis of disease or for determining appropriate treatment.
63	81400	Mopath procedure level 1	identification of single germline variant by techniques such as restriction enzyme digestion or melt curve analysis)
64	81401	Mopath procedure level 2	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)
65	81402	Mopath procedure level 3	>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])
66	81403	Mopath procedure level 4	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
67	81404	Mopath procedure level 5	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
68	81405	Mopath procedure level 6	analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons, regionally targeted cytogenomic array analysis
69	81406	Mopath procedure level 7	analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons, cytogenomic array analysis for neoplasia
70	81407	Mopath procedure level 8	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
71	81408	Mopath procedure level 9	analysis of >50 exons in a single gene by DNA sequence analysis