GENETICS
TEST MENU

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Genetics Test Menu

Test Name	Test N°	Use
Biochemical Genetics — Metabolite Tests		
Acetylcholinesterase (AChE), Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)*	510354	Analysis of midtrimester amniotic fluid for detection of open neural tube defects and ventral wall defects.
Acylcarnitine Profile, Quantitative, Plasma	070228	Used in the diagnosis and monitoring of inherited disorders of fatty acid oxidation and organic acidurias. May be used as a follow-up test to some abnormal newborn screen results.
NeuroSURE® Metabolites: Alpha Aminoadipic Semialdehyde, Cerebrospinal Fluid (CSF)	620037	CSF Alpha aminoadipic semialdehyde is useful for diagnosing pyridoxine-dependent seizures (PDS) and folinic acid-responsive seizures (FRS). This testing may also be used for assessment of VUS identified during genetic testing. Pyridoxine dependent seizures is a genetic disorder characterized by seizures in neonates or infants up to 3 years of age, which in general, respond to a pharmacologic dose of pyridoxine (vitamin B6). Alpha - aminoadipic semialdehyde dehydrogenase (antiquin) deficiency is the underlying defect. Biochemical testing should be done prior to gene sequencing, and can be done regardless of pyridoxine therapy.
Alpha Aminoadipic Semialdehyde (Urine)	620046	Urine Alpha aminoadipic semialdehyde (AASA) is useful for diagnosing pyridoxine-dependent seizures (PDS) and folinic acid-responsive seizures (FRS). Elevation of AASA can also occur in molybdenum cofactor deficiency. Urine AASA may also be used for assessment of VUS identified during genetic testing (e.g. Next Generation Sequencing or Capillary Sequencing Testing). Pyridoxine dependent seizures is a genetic disorder characterized by seizures in neonates or infants up to 3 years of age, which in general, respond to a pharmacologic dose of pyridoxine (vitamin B6). AASA dehyrogenase (antiquin) deficiency is the underlying defect. Piperideine-6-Carboxylate (P6C) is the cyclic isomer of AASA and the equilibrium between P6C and AASA is PH dependent. P6P reacts with pyridoxal 5'-phosphate and leads to deficiency of this cofactor. Folinic responsive seizures and PDS are allelic, and caused mutations in the ALDH7A1 gene. Biochemical testing should be done prior to gene sequencing, and can be done regardless of pyridoxine therapy.
Amino Acid Profile, Quantitative, Cerebrospinal Fluid	700180	Diagnosis and monitoring of inherited aminoacidurias. Most notably, quantitation of amino acids in CSF is useful in the diagnosis of glycine encephalopathy. Note: Diagnosis of glycine encephalopathy requires the calculation of a CSF : plasma glycine ratio.
Amino Acid Profile, Quantitative, Plasma	700068	Diagnosis and monitoring of inherited aminoacidurias, organic acidurias, and urea cycle defects. May be used as a follow-up confirmatory test to some abnormal newborn screen results
Amino Acid Profile, Quantitative, Urine	700140	Diagnosis and monitoring of inherited aminoacidurias, organic acidurias, and urea cycle defects. Screening for aminoacidopathies in urine alone is discouraged unless a disorder is suspected that predominantly manifests abnormalities in urine (eg, cystinuria, renal Fanconi syndrome).
Ammonia, Plasma	007054	Ammonia measurements are of use in the diagnosis of urea cycle deficiencies (any neonate with unexplained nausea, vomiting, or neurological deterioration appearing after first feeding), and they play an important part in the detection of Reye syndrome.
Carnitine, Total and Free	706500	Useful for diagnosis of primary and secondary carnitine deficiencies. Test includes measurement of total and free carnitine and calculation of the esterified-to-free carnitine ratio.

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Test Name	Test N°	Use
Creatine and Guanidinoacetate (Plasma)	620180	Disorders of creatine synthesis (deficiency of arginine:glycine amidinotransferase [AGAT] and guanidinoacetate methyltransferase [GAMT]) and creatine transporter (SLC6A8) deficiency are collectively described creatine deficiency syndromes (CDS). AGAT and GAMT deficiencies are inherited in an autosomal recessive manner, while the creatine transporter defect is X-linked. Diagnosis is possible by measuring guanidinoacetate (GAA), creatine (Crn) in plasma and urine. The profiles are specific for each clinical entity. Patients with GAMT deficiency typically exhibit normal to low Cr, very elevated GAA, and low Crn. Patients with AGAT deficiency typically exhibit normal to low Cr, low GAA, and normal to low Crn. In comparison, elevated Cr, normal GAA, normal to low Crn, and an elevated Cr:Crn ratio characterize patients with creatine transporter defect. AGAT, GAMT and the creatine transporter defect result in a depletion of cerebral creatine and typically present with global developmental delays, intellectual disability, and severe speech delay. Some patients with CDS develop seizures. Patients with GAMT and the creatine transporter deficiency exhibit behavioral problems and features of autism.
Creatine and Guanidinoacetate (Urine)	620170	Disorders of creatine synthesis (deficiency of arginine:glycine amidinotransferase [AGAT] and guanidinoacetate methyltransferase [GAMT]) and creatine transporter (SLC6A8) deficiency are collectively described creatine deficiency syndromes (CDS). AGAT and GAMT deficiencies are inherited in an autosomal recessive manner, while the creatine transporter defect is X-linked. Diagnosis is possible by measuring guanidinoacetate (GAA), creatine (Crn) in plasma and urine. The profiles are specific for each clinical entity. Patients with GAMT deficiency typically exhibit normal to low Cr, very elevated GAA, and low Crn. Patients with AGAT deficiency typically exhibit normal to low Cr, low GAA, and normal to low Crn. In comparison, elevated Cr, normal GAA, normal to low Crn, and an elevated Cr:Crn ratio characterize patients with creatine transporter defect. AGAT, GAMT and the creatine transporter defect result in a depletion of cerebral creatine and typically present with global developmental delays, intellectual disability, and severe speech delay. Some patients with CDS develop seizures. Patients with GAMT and the creatine transporter deficiency exhibit behavioral problems and features of autism.
α-Fetoprotein (AFP), AChE, Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)*	510305	Analysis of midtrimester amniotic fluid for detection of open neural tube and ventral wall defects.
α-Fetoprotein (AFP), Amniotic Fluid*	002428	Analysis of midtrimester amniotic fluid for detection of open neural tube defects and ventral wall defects. This test reflexes to AChE and fetal hemoglobin if AF-AFP is abnormal.
Lactate (CSF)	620044	CSF Lactate is useful for investigating possible disorders of mitochondrial metabolism, when used in conjunction with cerebrospinal fluid pyruvate collected at the same time to determine the Lactate:Pyruvate (L:P) ratio. The CSF L:P ratio is considered a helpful (not diagnostic) tool in the evaluation of patients with possible disorders of mitochondrial metabolism, especially in patients with normal blood L:P ratios. Pyruvic acid levels alone have little clinical utility. An elevated L:P ratio may indicate inherited disorders of the respiratory chain complex, tricarboxylic acid cycle disorders and pyruvate carboxylase deficiency. The L:P ratio is characteristically normal in other patients. An artificially high ratio can be found in acutely ill patients.
Lactic Acid, Plasma	004770	Lactic acidosis may be due to inborn errors of metabolism. Evaluate metabolic acidosis, regional or diffuse tissue hypoperfusion, hypoxia, ketoacidosis or nonketotic acidosis in diabetes mellitus, enzyme defects, glycogen storage disease (type I), thiamine deficiency, and hepatic failure.
3-O-Methyldopa (Plasma)	620176	Aromatic L-amino acid decarboxylase (AADC) is a pyridoxal 5'-phosphate dependent enzyme responsible for the formation of dopamine and serotonin. AADC deficiency is a congenital autosomal recessive metabolic disorder that causes hypotonia, hypokinesia, oculogyric crises, and signs of autonomic dysfunction beginning in infancy. In AADC deficiency can be detected by measuring 3-OMD in blood.

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Methylmalonic Acid, Serum or Plasma	706961	Serum methylmalonic acid (MMA) measurement is used to evaluate individuals with signs and symptoms associated with vitamin B12 deficiency or congenital methylmalonic acidemia.
Methylmalonic Acid, Urine	716365	Diagnose cobalamin (vitamin B12) deficiency or congenital methylmalonic aciduria.
NeuroSURE® Metabolites: 5-Methyltetrahydrofolate (CSF)	620008	CSF 5-Methyltetrahydrofolate (5-MTFH) is useful for determining a deficiency of folate in the central nervous system. CSF 5-MTFH may also be used for assessment of VUS identified during genetic testing. 5-MTFH is the predominant form of folate in cerebrospinal fluid (CSF). Low CSF 5-MTHF levels are associated with inborn errors of metabolism affecting folate metabolism, dietary deficiency of folate, cerebral folate syndromes and Kearns-Sayre syndrome. Symptoms may include, anemia, developmental delay, seizures, depression and dementia.
NeuroSURE® Metabolites: Neopterin (CSF)	620009	CSF Neopterin is useful for diagnosis of certain disorders of neurotransmitter metabolism and as a marker for immune system stimulation. This testing may also be used for assessment of VUS identified during genetic testing. Neopterin is released from macrophages and astrocytes following stimulation by interferon gamma. It is a non-specific marker for immune system stimualtion. An elevation in cerebrospinal fluid can be useful to help differentiate between immune problems and other causes of neurological disease.
NeuroSURE® Metabolites: Neopterin / Tetrahydrobiopterin (CSF)	620010	CSF Neopterin/Tetrahydrobiopterin is useful for diagnosis of certain disorders of neurotransmitter metabolism. This testing may also be used for assessment of VUS identified during genetic testing. Tetrahydrobiopterin (BH4) serves as a cofactor for the hydroxylation of phenylalanine and in the biosynthesis of biogenic amines. Deficiency of BH4 may occur as a result of mutations causing a reduction in one of the three biosynthetic enzymes, guanosine triphosphate cyclohydrolase. 6-pyruvoyl-tetrahydropterin synthase, sepiapterin reductase, or the two regenerating enzymes, pterin-4-carbinolamine dehydratase, and dihydropteridine reductase. Defects in BH4 metabolism can result in hyperphenylalaninemia and deficiency of the neurotransmitters dopamine and serotonin. Changes in CSF neopterin may also occur in deficiency of the BH4 synthesis pathway. Disorders of BH4 metabolism are characterized by a wide range of symptoms that may include developmental delay, mental disability, behavioral disturbances, dystonia, Parkinsonian symptoms, gait disturbances, speech delay, psychomotor retardation and ptosis.
NeuroSURE® Metabolites: Neurotransmitter Metabolites (5 HIAA, HVA, 30MD) (CSF)	620011	CSF Neurotransmitter Metabolites (5HIAA, HVA, 3OMD) is useful in diagnosing pediatric neurotransmitter diseases affecting dopamine and serotonin metabolism in the brain. These disorders are characterized by a wide range of symptoms that may include developmental delay, mental disability, behavioral disturbances, dystonia, seizures, encephalopathy, athetosis and ptosis. This testing may also be used for assessment of VUS identified during genetic testing.
Organic Acid Analysis, Urine	716720	Useful in the diagnosis and monitoring of inborn errors of organic acid metabolism, amino acid metabolism, urea cycle defects, and defects of the mitochondrial respiratory chain.
Orotic Acid, Urine	007010	Elevated levels of orotic acid help lead to positive diagnoses of specific urea cycle disorders and rare hereditary disorders such as orotic aciduria and uridine monophosphate synthase deficiency.
NeuroSURE® Metabolites: Pyridoxal 5'-phosphate, Cerebrospinal Fluid (CSF)	620034	Pyridoxal 5'-phosphate (PLP) (a member of the vitamin B6 family) is required as a cofactor for more than 100 different enzymes in the body. These may involve the metabolism of various neurotransmitters and amino acids. Inadequate PLP may occur due to genetic, nutritional deficiencies as well as reaction with various drugs. Inherited disorders that affect the CSF PLP level include pyriodx(am) ine phosphate oxidase (PNPO) deficiency, alpha amnioadipic semialdehyde dehydrogenase deficiency, hyperprolinermia type 2 and hypophoshatasia due to alkaline phosphatase deficiency.

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Pyruvate (CSF)	620045	CSF Pyruvate is useful when used in conjunction with CSF Lactate collected at the same time to determine the Lactate:Pyruvate (L:P) ratio. Pyruvic acid levels alone have little clinical utility. The CSF L:P ratio is considered a helpful (not diagnostic) tool in the evaluation of patients with possible disorders of mitochondrial metabolism, especially in patients with normal blood L:P ratios. An elevated L:P ratio may indicate inherited disorders of the respiratory chain complex, tricarboxylic acid cycle disorders and pyruvate carboxylase deficiency. The L:P ratio is characteristically normal in other patients. An artificially high ratio can be found in acutely ill patients.
Pyruvic Acid, Whole Blood	004788	Increased pyruvic acid levels have been associated with diabetes mellitus, vitamin deficiencies, uremia, congestive heart failure, liver diseases, muscular dystrophy, thiamine deficiency, and neoplastic conditions. Pyruvic acid is useful in assessing oxygen deprivation and provides an index of the severity of circulatory failure.
NeuroSURE® Metabolites: Sialic Acid, Cerebrospinal Fluid (CSF)	620036	CSF Sialic Acid is useful for diagnosing free sialic acid storage diseases (SASD). Mutations in the SLC17A5 gene encoding the lysosomal transporter sialin are associated with the free SASD: Salla disease (or the Finish type of sialuria), the more severe infantile free sialic acid storage disease (ISSD), and intermediate phenotypes with clinical findings of both Salla disease and ISSD. SASD are characterized by the abnormal retention of free sialic acid in the lysosome (OMIM 604369 and 269920). Patients with SASD usually present with nystagmus, progressive cerebellar ataxia, spasticity, and severe psychomotor delay. Cerebellar ataxia may be the primary symptom. These symptoms are associated with diffuse supratentorial hypomyelination, thin corpus callosum, and cortical and cerebellar atrophy. In some patients, sialic acid increases are identified only in CSF. This testing may also be used for assessment of VUS identified during genetic testing.
NeuroSURE® Metabolites: Succinyladenosine, Cerebrospinal Fluid (CSF)	620035	CSF Succinyladenosine is useful for diagnosing Adenylosuccinate Lyase (ADSL) Deficiency. Succinyladenosine is elevated in ADSL deficiency and results in succinylpurinemic autism, intellectual disability, and, in some cases, growth retardation associated with muscle wasting and epilepsy. In the absence of ADSL deficiency, succinyladenosine is either not detected or at very low levels in the CSF. Small elevations of succinyladenosine in spinal fluid have been reported in AICA-Ribosiduria (deficiency of AICAR transformylase) a devastating condition involving profound mental retardation, epilepsy, dysmorphic features and congenital blindness. Small elevations are also seen secondary to fumarase deficiency.
Thymidine and Deoxyuridine Analytes (Plasma)	620173	Plasma Thymidine/Deoxyuridine analyte is used for diagnosis of Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). MNGIE is an autosomal recessive disorder caused by mutations in the gene encoding thymidine phosphorylase (TP). The disease is characterized clinically by impaired eye movements, gastrointestinal dysmotility, cachexia, peripheral neuropathy, myopathy and leukoencephalopathy. TP is a cytosolic enzyme required for nucleoside homeostatis. In MNGIE, TP activity is severely reduced and consequently levels of thymidine and deoxyuridine in plasma are dramatically elevated. MNGIE patients may benefit from hematopoietic stem cell transplantation.
Biochemical Genetics — Enzyme Activity Tests		
Arylsulfatase A Deficiency, Leukocytes	402396	Diagnose patient with metachromatic leukodystrophy (MLD).
Enzyme Biotinidase Deficiency	402362	Diagnosis of biotinidase deficiency. This test is appropriate for the confirmation of newborn screen-positive biotinidase deficiency results.
α-Galactosidase A Deficiency, Leukocytes	402388	Diagnosis of patients with Fabry disease
β-Galactosidase Deficiency, Leukocytes	402370	Diagnose patients with β-galactosidase deficiency, Morquio disease type B (MPS IVb), and combined β-galactosidase/neuraminidase deficiency (galactosialidosis).
Lysosomal Acid Lipase (LAL) Deficiency	402300	Diagnose Wolman disease and cholesteryl ester storage disease (CESD) caused by LAL deficiency.
Tay-Sachs Disease, Biochemical, Leukocytes	511246	Identification of Tay-Sachs disease gene carriers and affected individuals. Identification of Sandhoff disease gene carriers and affected individuals.

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Tay-Sachs Disease, Biochemical	510412	Determine Tay-Sachs carrier and affected status. This serum assay should not be performed on women who are pregnant or who are taking oral contraceptives. Identification of Sandhoff carrier and affected status. May be used in the diagnosis of I-cell disease.
NeuroSURE® Metabolites: Thymidine Phosphorylase Enzyme Analysis (Blood)	620038	Thymidine phosphorylase Enzyme Analysis is used for the diagnosis of Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). MNGIE is an autosomal recessive disorder caused by mutations in the gene encoding thymidine phosphorylase (TP). The disease is characterized clinically by impaired eye movements, gastrointestinal dysmotility, cachexia, peripherl neuropathy, myopathy, and leukoencephalopathy. TP is a cytosolic enzyme required for nucleoside homeostasis. In MNGIE, TP activity is severely reduced and consequently levels of thymidine and deoxyuridine in plasma are drmatically elevated. MNGIE may benefit from hematopoietic stem cell transplantation.
Maternal Serum Screening		
α-Fetoprotein (AFP) Tetra Profile	017319	Screening test for open neural tube defects (detects 80% of open spina bifida, 90% of anencephaly), Down syndrome (detects 75% to 80%), and trisomy 18 (detects 73%).
α-Fetoprotein (AFP), Maternal Serum for Open Spina Bifida	010801	Screening test for open neural tube defects. Detects 80% of open spina bifida and 90% of anencephaly. Please note that this test does not provide screening for Down syndrome or trisomy 18.
First Trimester Screen With Nuchal Translucency	017500	Screening test for Down syndrome and trisomy 18 for use during the first trimester of pregnancy. Detects 86% of Down syndrome and 75% of trisomy 18. Test includes total human chorionic gonadotropin (hCG), pregnancy-associated plasma protein A (PAPP-A), and dimeric inhibin A (DIA) with maternal age risk and fetal nuchal translucency (NT) measurement. Performed from10.0 to 13.9 weeks of gestation. The NT must be performed by a sonographer credentialed by the NTQR program or other equivalent entity.
Integrated 1	017100	Screening test for Down syndrome and trisomy 18. Requires two specimens: one collected in the first trimester and one in the second trimester. This test number is for the first trimester portion of the test. Test measures PAPP-A and requires a fetal nuchal translucency (NT) measurement. Performed from 10.0 to 13.9 weeks of gestation. The NT measurement must be performed by a sonographer credentialed by the NTQR program or equivalent entity.
Integrated 2	017170	Screening test for Down syndrome and trisomy 18. Integrated screening requires two specimens: one collected in the first trimester and one in the second trimester. This test number is for the second trimester portion of the test. Detects 92.4% of Down syndrome and 90% of trisomy 18. Test combines results of Integrated 1 with AFP, hCG, uE3, and DIA. Performed from 15.0 to 21.9 weeks of gestation.
Sequential 1	017700	Screening test for Down syndrome and trisomy 18. Test measures PAPP-A and hCG and requires a fetal nuchal translucency (NT) measurement. Performed from 10.0 to 13.9 weeks of gestation. Patients who are not screen positive for this test must have Sequential 2 testing in the second trimester in order to receive a final risk assessment. The NT measurement must be performed by a sonographer credentialed by the NTQR program or equivalent entity.
Sequential 2	017750	Screening test for Down syndrome and trisomy 18. This test is for the second trimester portion of the test. Detects 92.3% of Down syndrome and 90% of trisomy 18. Test combines results of Sequential 1 with AFP, hCG, uE3, and DIA. Performed from 15.0 to 21.9 weeks of gestation.
Serum Integrated 1	017200	Screening test for Down syndrome and trisomy 18. Requires two specimens: one collected in the first trimester and one in the second trimester. Serum Integrated 1 is the first trimester portion of the test. Test measures PAPP-A. Performed from 10.0 to 13.9 weeks gestation. Test does not incorporate a fetal nuchal translucency (NT) measurement.

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Test Name	Test N°	Use
Serum Integrated 2	017270	Screening test for Down syndrome and trisomy 18. Requires two specimens: one collected in the first trimester and one in the second trimester. This test number is for the second trimester portion of the test. Detects 88.1% of Down syndrome and 90% of trisomy 18. Test combines results of Serum Integrated 1 with AFP, hCG, uE3, and DIA. Performed from 15.0 to 21.9 weeks of gestation.
Noninvasive Prenatal Testing		
MaterniT Genome	451941	The MaterniT Genome test provides comprehensive chromosome copy number analysis including unbalanced derivatives, and information about deletions or duplications of chromosome material 7 Mb or larger, as well as analysis of seven clinically relevant microdeletions less than 7 Mb in size.
MaterniT21 Genome NO Gender	452106	
MaterniT21 PLUS Core (chr21,18,13,sex)	451927	For pregnancies at increased risk of fetal abnormalities, the MaterniT21 PLUS test delivers a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, fetal sex.
MaterniT21 PLUS Core (chr21,18,13) NO Gender	451951	The MaterniT21 PLUS test is a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, for pregnancies at increased risk of fetal abnormalitites.
MaterniT21 PLUS Core + SCA	451934	For pregnancies at increased risk of fetal abnormalities, the MaterniT21 PLUS test delivers a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, fetal sex, and sex chromosome aneuploidies.
MaterniT21 PLUS Core + SCA, NO Gender	452112	For pregnancies at increased risk of fetal abnormalities, the MaterniT21 PLUS test delivers a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, and sex chromosome aneuploidies.
MaterniT21 PLUS Core + ESS	451931	For pregnancies at increased risk of fetal abnormalities, the MaterniT21 PLUS test delivers a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, fetal sex, and an enhanced sequencing series that examines seven clinically relevant microdeletions and two additional chromosomal regions, trisomies 22 and 16.
MaterniT21 PLUS Core + ESS, NO Gender	452136	For pregnancies at increased risk of fetal abnormalities, the MaterniT21 PLUS test delivers a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, and an enhanced sequencing series that examines seven clinically relevant microdeletions and two additional chromosomal regions, trisomies 22 and 16.
MaterniT21 PLUS Core + ESS + SCA	451937	For pregnancies at increased risk of fetal abnormalities, the MaterniT21 PLUS test delivers a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, fetal sex, sex chromosome aneuploidies, and an enhanced sequencing series that examines seven clinically relevant microdeletions and two additional chromosomal regions, trisomies 22 and 16.
MaterniT21 PLUS Core + ESS + SCA, NO Gender	452122	For pregnancies at increased risk of fetal abnormalities, the MaterniT21 PLUS test delivers a comprehensive NIPT for the analysis of chromosomal regions including trisomies 21, 18, and 13, sex chromosome aneuploidies, and an enhanced sequencing series that examines seven clinically relevant microdeletions and two additional chromosomal regions, trisomies 22 and 16.
MaterniT21 Genome Add On Redraw (GENOME-Flex)	452114	The MaterniT Genome test provides comprehensive chromosome copy number analysis including unbalanced derivatives, and information about deletions or duplications of chromosome material 7 Mb or larger, as well as analysis of seven clinically relevant microdeletions less than 7 Mb in size.
MaterniT21 Genome Add On (GENOME-Flex)	452104	
Cytogenetics — Prenatal and Postnatal Testing		
Chromosome Analysis and AFP, Amniotic Fluid*	510185	Prenatal detection of chromosome abnormalities in at-risk pregnant women. AFP analysis of midtrimester amniotic fluid for detection of open neural tube and ventral wall defects. This test reflexes to AChE and fetal hemoglobin if AF-AFP is abnormal. While chromosome analysis is being performed, additional biochemical or molecular analysis can be performed.
Chromosome Analysis, AFP, AChE, Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)*	511580	Determine fetal karyotype; prenatal diagnosis of Down syndrome or other chromosomal abnormalities; analysis of midtrimester amniotic fluid for detection of open neural tube and ventral wall defects.

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Chromosome Analysis, Amniotic Fluid*	052040	Determines fetal karyotype. The test allows prenatal detection of chromosomal rearrangements, aneuploidy, or mosaicism. Such groups include women who: are age 35 years of age or older; have a previous child having chromosome abnormality or multiple congenital abnormalities; have had two or more previous spontaneous abortions; have a family history of a chromosome abnormality; are known carriers of an X-linked disorder; are 31 years of age or older with twin pregnancies; have abnormal fetal ultrasound findings; or have a positive maternal serum marker screen. Additional biochemical or molecular tests may be performed on the cultured amniocytes.
Chromosome Analysis, Amniotic Fluid With Reflex to SNP Microarray (Reveal®)*	052104	The chromosome analysis determines fetal karyotype. A normal chromosome analysis will reflex to a high-resolution SNP microarray analysis targeting 2.695 million copy-number and allele-specific genome sites. The microarray test allows prenatal detection of clinically relevant alterations below the resolution of chromosome analysis. If specimens from a twin pregnancy are submitted by request, it can be reported if these are DZ or MZ twins. The genotyping portion of the SNP microarray will also screen for UPD for all chromosomes and estimate identity by descent.
Chromosome Analysis, Chorionic Villi Biopsy With Reflex to SNP Microarray (Reveal®)	511033	The chromosome analysis determines fetal karyotype. A normal chromosome analysis will reflex to a high-resolution SNP microarray analysis targeting 2.695 million copy-number and allele-specific genome sites. The microarray test allows prenatal detection of clinically relevant alterations below the resolution of chromosome analysis. If specimens from a twin pregnancy are submitted by request, it can be reported if these are DZ or MZ twins. The genotyping portion of the SNP microarray will also screen for UPD for all chromosomes and estimate identity by descent.
Chromosome Analysis, Instability Syndrome	511045	Chromosome analysis with DEB-induced breakage to assist in the diagnosis of Fanconi anemia (FA).
Chromosome Analysis, Prenatal Cordocentesis and Fetal Hemoglobin	511025	Rapid analysis of fetal chromosomes, used most frequently following the determination of fetal amniocyte mosaicism.
Chromosome Analysis, Products of Conception (POC) With Reflex to SNP Microarray (Reveal®)	052065	Evaluate possible chromosomal abnormalities as cause of miscarriage
Chromosome Analysis, Tissue Biopsies (Products of Conception, Skin)	052052	Evaluate possible chromosomal abnormalities as the cause of miscarriage. Extended study of mosaicism found in blood chromosome analysis.
Chromosome Analysis With Reflex to SNP Microarray – Pediatric (Reveal®)	052045	Detects microscopically visible chromosomal abnormalities and if normal; array reflex detects submicroscopic imbalance associated with developmental delay/ autism using 2.695 million genomic targets. The SNP microarray also provides detection of UPD (uniparental disomy) and the degree of consanguinity, as well as the genomic locations of recessive allele risk.
Chromosome Five-cell Count Plus Microarray (Reveal®), Amniotic Fluid	511590	Detects chromosomal imbalance that could be associated with developmental delay and congenital anomalies. The test allows prenatal detection of chromosomal aneuploidy, and is used to rule out tetraploidy and rearrangements not detected by array, such as balanced translocations and inversions. Also helps to clarify array abnormalities to determine if a structural rearrangement, marker or isochromosome is present. Array can only provide copy number imbalances and cannot determine structure of an abnormality. This test provides detection of uniparental disomy of any chromosome, the percentage and location of homozygosity, including the degree of identity by descent.
Chromosome Five-cell Count Plus Microarray (Reveal®), CVS	511555	Microarray detects chromosomal imbalance that could be associated with developmental delay and congenital anomalies. Test provides detection of uniparental disomy of any chromosome, the percentage and location of homozygosity, including the degree of identity by descent. The test allows prenatal detection of chromosomal aneuploidy, and is used to rule out tetraploidy and rearrangements not detected by array, such as balanced translocations and inversions. Also helps to clarify array abnormalities to determine if a structural rearrangement, marker or isochromosome is present. Array can only provide copy number imbalances and cannot determine structure of an abnormality.

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Test Name	Test N°	Use
Chromosome Five-cell Count Plus Microarray (Reveal®), Whole Blood	511535	Detects chromosomal imbalance that may be present in newborns or children with developmental delay and congenital anomalies and autism; provides detection of uniparental disomy of any chromosome and the degree of consanguinity as well as the genomic locations of recessive allele risk. Positive evaluation criteria include: DNA copy gain/loss within known clinically significant gene region of 50 Kb or greater or DNA copy number loss >200 kb or gain >500 kb outside known clinically significant regions with at least one OMIM-annotated gene or within a region of clear clinical significance. UPD testing is recommended for patient results demonstrating a long contiguous region of homozygosity in a single chromosome >20 Mb interstitially or >10 Mb telomerically (15 and 8 Mb, respectively, for imprinted chromosomes). Contiguous homozygosity >10 Mb within multiple chromosomes suggests common descent. These regions of potential recessive allele risk are designated. Abbreviated chromosome analysis detects balanced rearrangements and inversions. Also helps to clarify array abnormalities to determine if a structural rearrangement, marker or isochromosome is present. Array can only provide copy number imbalances and cannot determine structure of an abnormality.
InSight: Prenatal Amnio Aneuploid FISH Testing for Chromosomes 13, 18, 21, and XY	511894	Rapid direct identification of common prenatal aneuploidy (specific for 13, 18, 21, and XY). If specimen volume is too small, then direct FISH may not be performed and results may be obtained if cultured chromosome studies are ordered.
Microdeletion Syndromes*, FISH	510770	Confirmation/identification of deletions below the resolution of cytogenetics (Call 800-345-4363 for list of available probes.)
Prenatal Aneuploid Evaluation, Chorionic Villus Sampling*, FISH	510960	Rapid identification of common prenatal aneuploidy (specific for X, Y, 13, 18, and 21).
SNP Microarray (Direct)–Prenatal (Reveal®)	510200	This test detects chromosomal imbalance that could be associated with developmental delay/congenital anomalies. It provides detection of uniparental disomy of any chromosome, the percentage and location of homozygosity, including the degree of identity by descent.
SNP Microarray – Pediatric (Reveal®)	510002	Detects chromosomal imbalance that may be present in newborns or children with developmental delay/congenital anomalies/autism; genotyping in the array allows detection of uniparental disomy of autosomes, the presence of consanguinity, and the associated genomic location of recessive allele risk.
SNP Microarray– Prenatal (Reveal®)*	510100	This test will detect chromosomal imbalance that could be associated with developmental delay/congenital anomalies. Provides detection of possible uniparental disomy of any chromosome, and location of homozygosity including the degree of identity by descent.
SNP Microarray – Products of Conception (POC)/Tissue (Reveal®)	510110	Detects chromosomal imbalance that may be associated with fetal loss and is ideal for detection of complete or partial moles
Molecular Genetics — Carrier, Diagnostic, and Prenatal Testing		
Angelman and Prader-Willi Syndromes, DNA Analysis*	511210	This test detects all major causes of the Prader-Willi and Angelman syndromes.
α_1-Antitrypsin Deficiency, DNA Analysis*	511881	DNA-based determination of the two common alleles underlying α1-antitrypsin deficiency associated with chronic obstructive pulmonary disease (COPD) and childhood-onset liver disease. Prenatal testing is available.
Ashkenazi Jewish Carrier Profile	333561	Identification of carriers for Jewish heritage diseases, specifically Canavan disease, cystic fibrosis, familial dysautonomia, and Tay-Sachs disease.
Ashkenazi Jewish Carrier Profile Plus	332859	Identification of carriers for nine genetic diseases with elevated prevalence among people with Jewish heritage. The profile includes Bloom Syndrome, DNA Analysis (512145); Canavan Disease, DNA Analysis (511147); Cystic Fibrosis Profile, DNA Analysis (480533); Familial Dysautonomia, DNA Analysis (511352); Fanconi Anemia (Type C), DNA Analysis (511212); Gaucher Disease, DNA Analysis (511048); Mucolipidosis Type IV Mutation Detection (511386); Niemann-Pick Disease, DNA Analysis (511329); Tay-Sachs Disease, Biochemical, Leukocytes (511246).
Autoimmune Polyglandular Syndrome Type 1 (APS1/APECED): *AIRE* (Full Gene Sequencing)	252532	Confirm a clinical diagnosis of APS1/APECED; detect carriers; allow early diagnosis in family members.

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Autoimmune Polyglandular Syndrome Type 1 (APS1/APECED): *AIRE* (Known Mutation)	252737	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Bloom Syndrome, DNA Analysis*	512145	Identification of carrier and affected individuals by testing for the 2281del6ins7 variant associated with Bloom syndrome in the Ashkenazi Jewish population.
C9orf72 Genetic Testing (Repeat Expansion)	620017	Varients in the C9orf72 gene have been found to cause amyotrophic lateral sclerosis (ALS), a condition characterized by progressive muscle weakness, a loss of muscle mass, and an inability to control movement.
Canavan Disease, DNA Analysis*	511147	Identification of carrier and affected individuals for four variants, E285A, Y231X, 433-2A>G, and A305E, associated with Canavan disease. Prenatal testing is available.
Chronic Granulomatous Disease (CGD): *CYBB* (Full Gene Sequencing)	252529	Confirm a clinical diagnosis of CGD; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Chronic Granulomatous Disease (CGD): *CYBB* (Known Mutation)	252733	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Comprehensive Spinocerebellar Ataxia Repeat Expansion Panel	620167	See individual test components.
Congenital Adrenal Hyperplasia (CAH) 21-Hydroxylase (CYP21) Mutation (Endocrine Sciences)	500768	Identifies most common variants that cause congenital adrenal hyperplasia.
Cystic Fibrosis (CF) Profile, 32 Mutations, Fetal Analysis*	480541	Determine carrier or affected status for the 32 most common cystic fibrosis pathogenic variants. Includes the current mutation panel recommended by the ACMG and ACOG.
Cystic Fibrosis (CF) Profile, 97 Mutations, CF*plus*®	450020	Determine affected or carrier status for 97 CF variants. This assay may be used for individuals whose family history or ethnicity requires testing for less common variants. Also available for routine screening of pregnant couples. Discriminates between ΔF508 and the following polymorphisms: F508C, I506V, and I507V.
Cystic Fibrosis (CF) Profile, 97 Mutations, CF*plus*®, Fetal Analysis*	480819	An expanded mutation profile of 97 variants for cystic fibrosis for prenatal testing, diagnostic testing, and for testing in those individuals whose family history or ethnicity requires testing for less common variants.
Cystic Fibrosis (CF) Profile, 32 Mutations, DNA Analysis	480533	Determine affected or carrier status for the 32 most common CF variants. Routine screening for pregnant couples.
Cystic Fibrosis (CF) Profile, DNA Analysis and 5T Allele Genotyping	480555	Determine affected or carrier status for the 32 most common CF variants (includes the panel currently recommended by the ACMG and the ACOG); determine the presence of the 5T allele.
Dihydrolipoamide Dehydrogenase (DLD)*	450080	Detect dihydrolipoamide dehydrogenase deficiency (DLD), an autosomal-recessive disorder that occurs at an increased frequency in individuals of Ashkenazi Jewish descent, with a carrier frequency of 1 in 96.
DRPLA (ATN1) Genetic Testing (Repeat Expansion)	620158	Dentatorubral-pallidoluysian atrophy (DRPLA) is a progressive disorder of ataxia, myoclonus, epilepsy, and progressive intellectual deterioration in children and ataxia, choreoathetosis, and dementia or character changes in adults. The diagnosis of DRPLA is established in a proband with suggestive clinical findings and a family history of DRPLA or by the identification of a heterozygous pathogenic CAG trinucleotide expansion in ATN1.
Factor II (Prothrombin), DNA Analysis	511162	Mutation detection in factor II gene (OMIM 176930) assoicated with increased risk of thrombosis.
Factor V Leiden Mutation Analysis	511154	Detection of Leiden (R506Q) mutation in factor V gene (OMIM 227400), associated with increased risk of thrombosis.
Factor V Leiden With Reflex to R2	503853	Detection of the factor V Leiden mutation, followed by testing for the factor V R2 polymorphism in individuals positive for factor V Leiden (heterozygous). Factor V R2 further increases risk for venous thrombosis.

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Test Name	Test N°	Use
Factor V R2 DNA Analysis	503940	Follow-up evaluation in individuals with hyperhomocysteinemia; evaluation of patients with venous thrombosis.
Familial Dysautonomia, DNA Analysis*	511352	Identification of carrier and affected individuals by testing for two variants associated with familial dysautonomia in the Ashkenazi Jewish population.
Familial Hyperinsulinism (FHI)*	450070	Detect familial hyperinsulinism (FHI), which occurs at increased frequency in individuals of Ashkenazi Jewish descent, with a carrier frequency of 1 in 66.
Fanconi Anemia (Type C), DNA Analysis*	511212	Identification of carrier and affected individuals for two variants, IVS4+4A>T and 322delG, associated with Fanconi anemia, type C. Prenatal testing is available.
FBN1 (Marfan Syndrome) Full Gene Sequencing	452028	Confirm a clinical diagnosis of MFS; identify presymptomatic family members, guiding prophylactic measures
Fragile X Syndrome, DNA Analysis, Prenatal With Southern Blot Analysis*	510300	Testing performed on fetal sample (amnio or CVS) for fetus at risk for fragile X syndrome.
Fragile X Syndrome, PCR With Reflex to Southern Blot	511919	Carrier screening for individuals who are pregnant or considering pregnancy and who have NO family history of: fragile X syndrome, fragile X-related disorders (primary ovarian insufficiency, or late-onset ataxia) or unexplained intellectual disabilities, developmental delay, or autism.
Fragile X, PCR and Southern Blot Analysis	511655	Carrier screening for individuals with a family history of fragile X syndrome, fragile X-related disorders (primary ovarian insufficiency, or late-onset ataxia), or unexplained intellectual disabilities (including mental retardation), developmental delay, or autism. Diagnostic testing for: individuals with unexplained intellectual disabilities, developmental delay, or autism; women with primary ovarian insufficiency or failure, premature menopause, or infertility associated with elevated FSH levels before the age of 40 with no known cause; or individuals with late-onset intention tremor and/or cerebellar ataxia of unknown origin .
Friedreich Ataxia Genetic Testing (Trinucleotide Repeat Expansion)	620077	Friedreich ataxia is a genetic condition that affects the nervous system and causes movement problems.
α-Galactosidase A Deficiency (Full Gene Sequencing	252225	Patients with clinical features of Fabry disease, both male and female; carrier testing for females with affected male relatives; patients with left ventricular hypertrophy or cardiomyopathy who otherwise do not have a classic Fabry disease phenotype; parents, siblings, and possibly children of a patient known to carry a variant in *GLA* gene; prenatal testing when a parent is diagnosed with Fabry disease and has an identified *GLA* variant.
α-Galactosidase A Deficiency (Known Mutation)	252230	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Gaucher Disease, DNA Analysis*	511048	Identifies carriers and affected individuals using eight variants associated with Gaucher disease in the Ashkenazi Jewish population. DNA testing may be used to confirm affected status.
GeneSeq®: Cardio-Early-onset Coronary Artery Disease/Familial Hypercholesterolemia Profile	451416	Confirm a clinical diagnosis of coronary artery disease and identify presymptomatic family members, guiding prophylactic measures.
GeneSeq®: Cardio-Familial Aortopathy Profile	451432	Confirm a clinical diagnosis of aortopathy and identify presymptomatic family members, guiding prophylactic measures.
GeneSeq®: Cardio-Familial Arrhythmia Profile	451412	Confirm a clinical diagnosis of arrhythmia and identify presymptomatic family members, guiding prophylactic measures.
GeneSeq®: Cardio-Familial Cardiomyopathy Profile	451422	Confirm a clinical diagnosis of cardiomyopathy and identify presymptomatic family members, guiding prophylactic measures.
GeneSeq®: Cardio-Familial Congenital Heart Disease Profile	451402	Confirm a clinical diagnosis of congenital heart disease and identify presymptomatic family members, guiding prophylactic measures.
GeneSeq®: Cardio-Familial Hypercholesterolemia Profile	452040	Confirm a clinical diagnosis of familial hypercholesterolemia (FH) and allow early diagnosis in family members, thus promoting early intervention, which may prevent or repair atherosclerotic damage and lower the risk of coronary artery disease.

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Test Name	Test N°	Use
GeneSeq®: Cardio-Gene Specific Sequencing	452053	Full gene sequencing is available for all the genes included in any of the GeneSeq®: Cardio profiles: GeneSeq®: Cardio-Familial Arrhythmia Profile (451412); GeneSeq®: Cardio-Familial Cardiomyopathy Panel (451422); GeneSeq®: Cardio-Noonan Syndrome and Related Conditions Profile (451441); GeneSeq®: Cardio-Familial Aortopathy Profile (451432); GeneSeq®: Cardio-Early-onset Coronary Artery Disease/FamilialHypercholesterolemia Profile (451416); GeneSeq®: Cardio-Familial Hypercholesterolemia Profile (452040); and GeneSeq®: Cardio-Familial Congenital Heart Disease Profile (451402). Links to these tests are in Related Information.
GeneSeq®: Cardio-Noonan Syndrome / RASopathies Profile	451441	Confirm a clinical diagnosis of Noonan syndrome and identify presymptomatic family members, guiding prophylatic measures. See Prenatal Noonan Syndrome (451890) for fetal testing.
GeneSeq® PLUS	630068	Full gene sequencing is available for genes included in the Inheritest®500 PLUS panel. See related Inheritest® test codes: Inheritest® Carrier Screen, Comprehensive (144 genes) (451950); Inheritest® Carrier Screen, Ashkenazi Jewish (48 genes) (451920); Inheritest® Carrier Screen, Society-guided (14 genes) (451960). For HBA1 and HBA2 (alpha-thalassemia) see α-Thalassemia, DNA Analysis (511172); for SMN1 see Spinal Muscular Atrophy (SMA) Carrier Testing (450010); and for FMR1 see Fragile X Syndrome, PCR with Reflex to Southern Blot (511919). Links to these tests are in Related Information.
GeneSeq® PLUS, Prenatal*	630119	
GeneSeq® PLUS without VUS	630085	
GeneSeq® PLUS without VUS, Prenatal*	630102	
GJB2 Sequencing, Full Gene Sequencing*	511405	Confirms a diagnosis of GJB2-related nonsyndromic sensioneural hearing loss (NSHL); detects carriers.
GJB2 Sequencing, Family-targeted (Single Exon Sequencing–Known Mutation)*	511414	Detects known familial variants in the connexin 26 (GJB2) gene associated with nonsyndromic sensorineural hearing loss (NSHL). This option is available when the variant is known and can be documented by the ordering physician.
Glycogen Storage Disease 1a*	511290	Glycogen storage disease type 1a (GSD1a), also called von Gierke disease (OMIM 232200), is a recessive inherited disorder characterized by an enlarged liver and kidneys due to the accumulation of glycogen and fat. Testing encompasses two variants associated with GSD1a in the Ashkenazi Jewish population.
Hereditary Hemochromatosis, DNA Analysis	511345	Follow-up evaluation in individuals with elevated saturated transferrin; detection of affected individuals and carriers of hereditary hemochromatosis.
Huntington Disease (HTT) Genetic Testing (Repeat Expansion)	620016	Huntington disease (HD) is a neurodegenerative disease of mid-life onset that produces choreic movements and cognitive decline, often accompanied by psychiatric changes. The disease is caused by an expansion of the CAG repeats in 3-5 out of 100,000 individuals. However, the prevalence of HD exceeds 15 per 100,000 in some populations, mostly of Western European origin. Juvenile-onset HD occurs in approximately 5% of affected patients, is rapidly progressive, and presents with rigidity, spasticity, and intellectual decline before the age of 20 years. The symptoms result from the selective loss of neurons, most notably in the caudate nucleus and putamen, and there is currently no effective treatment.
Hyper-IgE Syndrome (HIES): *STAT3* (Full Gene Sequencing)	252449	Confirm a clinical diagnosis of HIES; detect carriers; allow early diagnosis of family members.
Hyper-IgE Syndrome (HIES): *STAT3* (Known Mutation)	252680	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Hyper-IgM Syndrome (HIGM): (*AICDA* for HIGM2) (Full Gene Sequencing)	252425	Confirm a clinical diagnosis of HIGM; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Hyper-IgM Syndrome (HIGM): (*AICDA* for HIGM2) (Known Mutation)	252663	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Hyper-IgM Syndrome (HIGM): (*CD40* [TNFRSF5] for HIGM3) (Full Gene Sequencing)	252432	Confirm a clinical diagnosis of HIGM; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Hyper-IgM Syndrome (HIGM): (*CD40* [TNFRSF5] for HIGM3) (Known Mutation)	252670	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.

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Hyper-IgM Syndrome (HIGM): (*CD40LG* [TNFSF5] for HIGM1) (Full Gene Sequencing)	252435	Confirm a clinical diagnosis of HIGM; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Hyper-IgM Syndrome (HIGM): (*CD40LG* [TNFSF5] for HIGM1) (Known Mutation)	252673	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Hyper-IgM Syndrome (HIGM): (*UNG* for HIGM5) (Full Gene Sequencing)	252428	Confirm a clinical diagnosis of HIGM; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Hyper-IgM Syndrome (HIGM): (*UNG* for HIGM5) (Known Mutation)	252666	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Hyper-IgM Syndrome (HIGM): Four-gene Profile (*AICDA, UNG, CD40, CD40LG*) (Full Gene Sequencing)	252446	Confirm a clinical diagnosis of HIGM; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Hyper-IgM Syndrome (HIGM): Three-gene Profile (*AICDA, UNG, CD40*) (Full Gene Sequencing)	252442	
Hyper-IgM Syndrome (HIGM): Two-gene Profile (*AICDA, UNG*) (Full Gene Sequencing)	252439	
Hypohidrotic Ectodermal Dysplasia With Immune Deficiency (HED-ID): *IKBKG* (NEMO) (Full Gene Sequencing)	252539	Confirm a clinical diagnosis of HED-ID; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Hypohidrotic Ectodermal Dysplasia With Immune Deficiency (HED-ID): *IKBKG* (NEMO) (Known Mutation)	252744	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Infertility—Male, Y Deletion Analysis	512053	Determine the genetic basis for oligospermia or azoöspermia. Azoöspermia may also be associated with cystic fibrosis mutations, primarily the 5T allele.
Inheritest® 500 PLUS Panel	630049	Carrier testing by analyzing 525 genes, each associated with a clinically relevant disorder, including fragile X syndrome and spinal muscular atrophy.
Inheritest® 500 PLUS with Repro Partners Report	630217	Carrier testing by analyzing 525 genes, each associated with a clinically relevant disorder, including Fragile X syndrome and spinal muscular atrophy. This test code should be utilized if a combined partners' report is desired.
Inheritest® Carrier Screen, Ashkenazi Jewish Panel (48 Genes)	451920	Carrier screening by analyzing 48 genes for more than 2,300 pathogenic variants associated with more than 47 autosomal recessive or X-linked disorders including genes for fragile X syndrome, spinal muscular atrophy, and diseases specific to individuals of Ashkenazi Jewish descent.
Inheritest® Carrier Screen, Comprehensive Panel (144 Genes)	451950	Carrier testing by analyzing 144 genes for more than 9,400 pathogenic variants associated with more than 116 autosomal recessive or X-linked disorders, including fragile X syndrome and spinal muscular atrophy.
Inheritest® Core Panel	451964	Carrier screening for Cystic Fibrosis (97 mutations), Spinal Muscular Atrophy, and Fragile X Syndrome.
Inheritest® Gene-specific Sequencing, NGS	451910	Full gene sequencing is available for all the genes included in the Inheritest® NGS panels. See related Inheritest® test codes: Inheritest® Carrier Screen, Comprehensive Panel (144 Genes) (451950); or Inheritest® Carrier Screen, Ashkenazi Jewish Panel (48 Genes) (451920); or Inheritest® Carrier Screen, Society-guided Panel (14 Genes) (451960). For HBA1 and HBA2 (alpha-thalassemia) see α-Thalassemia, DNA Analysis (511172); for SMN1 see Spinal Muscular Atrophy (SMA) Carrier Testing (450010); and for FMR1 see Fragile X Syndrome, PCR With Reflex to Southern Blot (511919).
Inheritest® Carrier Screen, Society-guided Panel (14 Genes)	451960	Carrier testing by analyzing 14 genes for more than 1,200 pathogenic variants associated with more than 13 autosomal recessive or X-linked disorders including fragile X syndrome and spinal muscular atrophy.
Interferon-γ Receptor Deficiency: *IFNGR1* (Full Gene Sequencing)	252519	Carrier testing by analyzing 14 genes for more than 1,200 pathogenic variants associated with more than 13 autosomal recessive or X-linked disorders including fragile X syndrome and spinal muscular atrophy.

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Interferon-γ Receptor Deficiency: *IFNGR1* (Known Mutation)	252727	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Interferon-γ Receptor Deficiency: *IFNGR2* (Full Gene Sequencing)	252522	Confirm a clinical diagnosis of IFNGR2; guide therapy; detect carriers; allow early diagnosis in family members.
Interferon-γ Receptor Deficiency: *IFNGR2* (Known Mutation)	252730	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Interferon-γ Receptor Deficiency: Two-gene Profile (*IFNGR1, IFNGR2*) (Full Gene Sequencing)	252525	Confirm a clinical diagnosis of IFNGR; guide therapy; detect carriers; allow early diagnosis in family members.
Joubert Syndrome Type II, DNA Analysis*	511490	Detect the presence of the R12L mutation (also called R73L) in the *TMEM216* gene.
Maple Syrup Urine Disease Carrier Test, DNA*	511310	Maple syrup urine disease (MSUD, OMIM 248600) is an inherited recessive disease caused by deficient activity of branched-chain α-ketoacid dehydrogenase. Testing encompasses four variants associated with MSUD in either the Ashkenazi Jewish or Mennonite populations.
Maternal Cell Contamination*	511402	Quality assurance for interpretation of prenatal molecular genetic test results.
Maturity-Onset Diabetes of the Young (MODY) Genetic Profile	504603	Maturity-onset diabetes of the young (MODY) is a suspected diagnosis in young non-obese patients who lack an autoimmune cause for diabetes and who have a family history of diabetes in successive generations. The majority of MODY cases are due to mutations in one of four genes. Identifying a mutation in one of these MODY genes can lead to improved treatment, increased surveillance for related symptoms, and earlier detection in currently asymptomatic family members. GCK encodes the enzyme glucokinase, a key regulator of glucose metabolism in pancreatic beta cells. The three HNF (hepatic nuclear factor) genes encode transcription factors that regulate gene expression in the pancreas.
Methylenetetrahydrofolate Reductase (MTHFR) Thermolabile Variant, DNA Analysis	511238	Follow-up evaluation in individuals with hyperhomocysteinemia; evaluation of patients with venous thrombosis.
Mucolipidosis Type IV Mutation Detection*	511386	Carrier testing for mucolipidosis type IV in the Ashkenazi Jewish population. DNA testing may be used to confirm affected status. Prenatal testing is available.
Mutation-specific Sequencing, Whole Blood	451382	This test is available for family testing when a variant has been specifically identified through universal carrier screening (Inheritest® Gene-specific Sequencing, NGS [451910]; Inheritest® Ashkenazi Jewish Carrier Screening Panel, NGS [451920]; Inheritest® Comprehensive Panel, NGS [451950]; or Inheritest® Society-guided Screening Panel, NGS [451960]); or VistaSeq® Hereditary Cancer Panel [481220] or VistaSeq® Hereditary Cancer Panel Without *BRCA* [481240]); or through GeneSeq: Cardio testing. (See links to tests in Related Information).
Mutation-specific Sequencing, Prenatal*	451385	This test is available for partner testing when a carrier is identified through universal carrier screening (Inheritest® Comprehensive, NGS [451950] or Inheritest® Ashkenazi Jewish Carrier Screening, NGS [451920] or Inheritest® Society-guided Screening, NGS [451960] or Inheritest® Gene-specific Sequencing, NGS [451910]).
Myotonic Dystrophy 1 (DMPK) Genetic Testing (Repeat Expansion)	620084	Type 1 myotonic dystrophy results from a mutation in the DMPK gene known as a trinucleotide repeat expansion. This mutation increases in the size of the repeated CTG segment in the DMPK gene. People with type 1 myotonic dystrophy have from 50 to 5,000 CTG repeats in most cells. The number of repeats may be even greater in certain types of cells, such as muscle cells.
Myotonic Dystrophy 2 (ZNF9/CNBP) Genetic Testing (Repeat Expansion)	620087	Type 2 myotonic dystrophy results from a mutation in the CNBP gene known as a tetranucleotide repeat expansion. This mutation increases in size of the repeated CCTG segment in the CNBP gene. People with type 2 myotonic dystrophy have from 75 to more than 11,000 CCTG repeats.
Nemaline Myopathy*	450040	Detect nemaline myopathy, which occurs at increased frequency in individuals of Ashkenazi Jewish descent, with a carrier frequency of 1 in 149. Nemaline myopathy is a disorder characterized by weakness and poor muscle tone.

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Niemann-Pick Disease, DNA Analysis*	511329	Identification of carrier and affected individuals for four variants associated with Neimann-Pick disease, types A and B. Prenatal testing is available.
PMP22 MLPA Deletion/Duplication Analysis	620081	Pathogenic variants in the PMP22 gene cause several forms of a neurological disorder called Charot-Marie-Tooth disease. This disorder damamges the peripheral nerves, which can result in loss of sensation and wasting (atrophy) of muscles in the feet, legs and hands.
Prenatal Noonan Syndrome*	451890	Prenatal diagnosis for at-risk pregnancies when a parent is affected or when abnormalities are seen on fetal ultrasound.
SCA1 (ATXN1) Genetic Testing (Repeat Expansion)	620114	Spinocerebellar ataxias (SCAs), and episodic ataxias are the most common types of autosomal dominant cerebellar ataxias (ADCAs). SCAs are numbered based upon their time of identification. SCA3 is the most common type of SCA worldwide, followed by SCA2, SCA1, and SCA6. Some of the complicated forms have not been given a SCA number, like Dentatorubral Pallidoluysian Atrophy (DRPLA). Anticipation can be observed in the autosomal dominant ataxias in which CAG trinucleotide repeats occur. Anticipation results from expansion in the number of CAG repeats with transmission of the gene to subsequent generations. Most ADCAs have an overlap in clinical clinical presentation, which makes it hard to differentiate. The most frequent clinical symptoms in all ADCAs are progressive adult-onset gait ataxia (often with hand dysmetria), and dysarthria associated with cerebellar atrophy. The episodic ataxias are characterized by periods of unsteady gait and often associated with nystagmus or or dysarthria. Myokymia, vertigo, or hearing loss may occur in some of the subtypes. Permanent ataxia and even cerebellar atrophy may result late in the disease course.
SCA2 (ATXN2) Genetic Testing (Repeat Expansion)	620118	
SCA3 (ATXN3) Genetic Testing (Repeat Expansion)	620123	
SCA6 (CACNA1A) Genetic Testing (Repeat Expansion)	620127	
SCA7 (ATXN7) Genetic Testing (Repeat Expansion)	620131	
SCA8 (ATXN8) Genetic Testing (Repeat Expansion)	620135	
SCA10 (ATXN10) Genetic Testing (Repeat Expansion)	620140	
SCA12 (PPP2R2B) Genetic Testing (Repeat Expansion)	620144	
SCA17 (TBP) Genetic Testing (Repeat Expansion)	620149	
SCA36 (NOP56) Genetic Testing (Repeat Expansion)	620154	
SCN1A Sequencing, Full Gene	511236	Confirms a diagnosis of a *SCN1A*-related seizure disorder, including but not limited to severe myoclonic epilepsy of infancy (SMEI, also known as Dravet Syndrome), intractable childhood epilepsy with generalized tonic-cloinic seizures (ICE-GTC), generalize epilepsy with febrile seizures plus (GEFS+).
SCN1A Family-targeted Sequencing	511274	This test is intended for testing of additional family members once a pathogenic variant or variant of uncertain significance has been identified in an affected individual. This option is available when the mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.
Sex Determination (SRY), DNA Analysis*	510222	Resolution of unexplained sex reversal or infertility through detection of the SRY gene; can help rule out mosaicism for 46,XY cells in Turner syndrome patients.
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): *DCLRE1C* (Artemis) for RS-SCID or SCIDA (Full Gene Sequencing)	252492	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): *DCLRE1C* (Artemis) for RS-SCID or SCIDA (Known Mutation)	252723	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.

*This test can be used for prenatal diagnosis on amniotic fluid and/or CVS. **Maternal and prenatal specimens should be submitted on separate LabCorp test request forms when testing for both specimens is desired.** For questions regarding genetic testing, please call 800-345-4363. Many of the assays listed here are discussed further in other publications; consult your LabCorp service representative to learn more.

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Test Name	Test N°	Use
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Eight-gene Profile (*IL2RG, JAK3, RAG1, RAG2, IL7R, ADA, CD3D, CD3E*) (Full Gene Sequencing)	252513	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Nine-gene Profile (*IL2RG, JAK3, RAG1, RAG2, IL7R, ADA, CD3D, CD3E, DCLREC1C* [Artemis]) (Full Gene Sequencing)	252516	
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): *RAG1* (Full Gene Sequencing)	252470	
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): *RAG1* (Known Mutation)	252701	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): *RAG1, RAG2, DCLRE1C* (Artemis) (Full Gene Sequencing)	252503	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): *RAG2* (Full Gene Sequencing)	252472	
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): *RAG2* (Known Mutation)	252704	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Three-gene Profile (*IL2RG, ADA, IL7R*) (Full Gene Sequencing)	252509	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Two-gene Profile (*RAG1, RAG2*) (Full Gene Sequencing)	252499	
Severe Combined Immunodeficiency (SCID): *ADA* (Full Gene Sequencing)	252475	
Severe Combined Immunodeficiency (SCID): *ADA* (Known Mutation)	252707	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID): *CD3D* (Full Gene Sequencing)	252482	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID): *CD3D* (Known Mutation)	252713	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID): *CD3E* (Full Gene Sequencing)	252485	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID): *CD3E* (Known Mutation)	252716	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID): *IL2RG* for XSCID (Full Gene Sequencing)	252463	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID): *IL2RG* for XSCID (Known Mutation)	252694	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID): *IL7R* (Full Gene Sequencing)	252479	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.

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Genetics Test Menu

Test Name	Test N°	Use
Severe Combined Immunodeficiency (SCID): *IL7R* (Known Mutation)	252710	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID): *JAK3* (Full Gene Sequencing)	252466	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID): *JAK3* (Known Mutation)	252697	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Severe Combined Immunodeficiency (SCID): Three-gene Profile (*IL7R, CD3D, CD3E*) (Full Gene Sequencing)	252506	Confirm a clinical diagnosis of SCID; detect carriers; allow early diagnosis in family members.
Severe Combined Immunodeficiency (SCID): Two-gene Profile (*IL2RG, JAK3*) (Full Gene Sequencing)	252496	
Severe Combined Immunodeficiency (SCID): *ZAP70* (Full Gene Sequencing)	252489	
Severe Combined Immunodeficiency (SCID): *ZAP70* (Known Mutation)	252720	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
***SHOX*, DHPLC (Endocrine Sciences)**	500110	Identifies variants causing short stature related to SHOX deficiency. SHOX deficiency is an indication for somatotropin (Humatrope®).
Sickle Cell Anemia Mutation Analysis, Fetal*	451391	DNA analysis to detect variants known to cause sickle cell anemia.
Spinal Muscular Atrophy (SMA)	450010	Carrier testing for individuals in the general population, or individuals with a family history of SMA, or couples who are planning a pregnancy or who are already pregnant. Pediatric or adult diagnostic testing when a diagnosis of SMA is suspected. Test 452140, Prenatal Spinal Muscular Atrophy (SMA) Testing, should be used for prenatal diagnosis for at-risk pregnancies, when both parents are carriers or when severe joint contractures are found on fetal ultrasound.
Tay-Sachs Disease, Biochemical, Leukocytes	511246	Identification of Tay-Sachs disease gene carriers and affected individuals. Identification of Sandhoff disease gene carriers and affected individuals.
Tay-Sachs Disease, Biochemical	510412	Determine Tay-Sachs carrier and affected status. This serum assay should not be performed on women who are pregnant or who are taking oral contraceptives. Identification of Sandhoff carrier and affected status. May be used in the diagnosis of I-cell disease.
Tay-Sachs Disease, DNA Analysis*	510404	Identifies Tay-Sachs disease carriers and affected individuals in specific ethnic groups. The test identifies three variants associated with the Ashkenazi Jewish population, one variant associated with the French Canadian population, one associated with non-Jewish Caucasians, and two pseudodeficiency variants.
α-Thalassemia, DNA Analysis*	511172	Detects α-thalassemia, the most common inherited disorder of hemoglobin (Hb) synthesis in the world. Gene frequencies vary between 1% and 98% throughout the tropics and subtropics.
β-Thalassemia: *HBB* (Full Gene Sequencing)	252823	Confirm a clinical diagnosis of β-thalassemia; detect carriers; help to establish a prognosis.
β-Thalassemia: *HBB* (Known Mutation)	252827	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
β-Thalassemia: *HBB* Prenatal Test (Full Gene Sequencing)*	252867	Use for prenatal analysis. Can confirm a clinical diagnosis of β-thalassemia, detect carriers, and help to establish a prognosis.
β-Thalassemia: *HBB* Prenatal Test (Known Mutation)*	252870	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.

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Test Name	Test N°	Use
Thrombotic Risk Profile, DNA Analysis	512103	Evaluation appropriate for patients with venous thrombosis. Molecular analysis of factor V Leiden factor II (prothrombin), and methylenetetrahydrofolate reductase (MTHFR) is performed.
Uniparental Disomy (UPD), Proband, DNA Analysis	470074	Establishes the chromosome parent of origin to rule out syndromes that result from single-parent inheritance of a specific chromosome pair.
Usher Syndrome Type IF*	450060	Detect Usher syndrome type IF, which occurs at increased frequency in individuals of Ashkenazi Jewish descent, with a carrier frequency of 1 in 141. This type of Usher syndrome causes profound deafness at birth, severe balance problems, as well as vision impairment. Blindness progresses over time.
Usher Syndrome Type III*	450050	Detect Usher syndrome type III, which occurs at increased frequency in individuals of Ashkenazi Jewish descent, with a carrier frequency of 1 in 107. This type of Usher syndrome causes hearing problems that progressively worsen, although the rate of detection varies.
von Hippel-Lindau Disease (VHL): *VHL* (OPT) (Full Gene Sequencing)	252559	Confirm a clinical diagnosis of VHL; identify presymptomatic family members.
von Hippel-Lindau Disease (VHL): *VHL* (OPT) (Known Mutation)	252562	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Walker-Warburg Syndrome*	511480	Detection of the c.1167insA variant in the *FKTN* gene, which accounts for approximately 99% of Walker-Warburg carriers in the Ashkenazi Jewish population.
Whole Exome Sequencing - DUO (Proband)	620023	Whole Exome Sequencing (WES) is a genetic test used to identify a heritable cause of a disorder. WES searches through all coding regions of all genes currently identified; thus, it has a higher chance to find the cause of a heritable disease. WES can be used if a patient has symptoms, which, after exhaustive testing, cannot be linked to a diagnosis and corrective treatment is necessary to change the prognosis. WES can also be used if, upon clinical presentation, multiple disease states may be suspected and a clinician wishes to improve his/her testing approach. Once a genetic variant has been identified, this information can then be linked back to the phenotype of the patient, which will improve the pathway to a correct diagnosis and a suitable treatment plan can be administered. DUO testing consists of a proband or patient sample, and one biological parent or family member in the case that both parents are not available for testing.
Whole Exome Sequencing - Proband Only	620024	Whole Exome Sequencing (WES) is a genetic test used to identify a heritable cause of a disorder. WES searches through all coding regions of all genes currently identified thus it has a higher chance to find the cause of a heritable disease. WES can be used if a patient has symptoms which, after exhaustive testing, cannot be linked to a diagnosis and corrective treatment is necessary to change the prognosis. WES can also be used if upon clinical presentation, multiple disease states may be suspected and a clinician wishes to improve his/her testing approach. Once a genetic variant has been identified, this information can then be linked back to the phenotype of the patient, which will improve the pathway to a correct diagnosis and a suitable treatment plan can be administered.
Whole Exome Sequencing - TRIO (Proband)	620022	Whole Exome Sequencing (WES) is a genetic test used to identify a heritable cause of a disorder. WES searches through all coding regions of all genes currently identified; thus, it has a higher chance to find the cause of a heritable disease. WES can be used if a patient has symptoms, which, after exhaustive testing, cannot be linked to a diagnosis and corrective treatment is necessary to change the prognosis. WES can also be used if, upon clinical presentation, multiple disease states may be suspected and a clinician wishes to improve his/her testing approach. Once a genetic variant has been identified, this information can then be linked back to the phenotype of the patient, which will improve the pathway to a correct diagnosis and a suitable treatment plan can be administered. TRIO testing consists of a proband or patient sample, and both biological parents. In the case both parents are not available for testing, up to two family member samples are also accepted. Trios are preferred for better diagnostic sensitivity.

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Genetics Test Menu

Test Name	Test N°	Use
Whole Exome Sequencing Comparator - Additional FM	620194	Whole exome sequencing (WES) is a genetic test used to identify a hertiable cause of disorder. This test should be used in cases of WES Duo or Trio sequencing where the parental samples are unavailable and an additional family member can be used as a comparator to inform the diagnosis of the proband.
Whole Exome Sequencing Comparator - Father	620197	Whole exome sequencing (WES) is a genetic test used to identify a hertiable cause of disorder. This test should be used in cases of WES Duo or Trio sequencing where the father can be used as a comparator to inform the diagnosis of the proband.
Whole Exome Sequencing Comparator - Mother	620192	Whole exome sequencing (WES) is a genetic test used to identify a hertiable cause of disorder. This test should be used in cases of WES Duo or Trio sequencing where the mother can be used as a comparator to inform the diagnosis of the proband.
Wiskott-Aldrich Syndrome (WAS): *WAS* (Full Gene Sequencing)	252459	Confirm a clinical diagnosis of WAS; detect carriers; allow early diagnosis in family members.
Wiskott-Aldrich Syndrome (WAS): *WAS* (Known Mutation)	252690	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
X-linked Agammaglobulinemia (XLA): *BTK* (Full Gene Sequencing)	252453	Confirm a clinical diagnosis of XLA; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
X-linked Agammaglobulinemia (XLA): *BTK* (Known Mutation)	252683	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
X-linked Lymphoproliferative Disease (XLP): *SH2D1A* (Full Gene Sequencing)	252535	Confirm a clinical diagnosis of XLP; detect carriers; allow early diagnosis in family members.
X-linked Lymphoproliferative Disease (XLP): *SH2D1A* (Known Mutation)	252740	This option is available when the variant is known and can be documented by the ordering physician. If the variant cannot be documented, please order full gene sequencing.
Cancer Genetics — Germline (Hereditary) Testing		
***BRCA1/2* Comprehensive Analysis (BRCA*ssure*®)**	485030	According to the National Comprehensive Cancer Network, testing is indicated if one of the features mentioned below is present in the family: Early-age-onset (age <50 years) breast cancer, including both invasive and ductal carcinoma in situ (DCIS) breast cancers; two breast primaries or breast and ovarian/fallopian tube/primary peritoneal cancer in a single individual or two or more breast primaries or breast and ovarian/fallopian tube/primary peritoneal cancers in close (first-, second-, and third-degree) relatives(s) from the same side of the family; populations at risk (eg, Ashkenazi Jewish); member of a family with a known *BRCA1* or *BRCA2* mutation; any male breast cancer; ovarian/fallopian tube/primary peritoneal cancer at any age.
***BRCA1/2* Deletion/Duplication Analysis (BRCA*ssure*®)**	485050	This test code should be used when an individual has had previous sequence analysis of the *BRCA1* and *BRCA2* genes but did not have previous testing for large deletions or duplications of the *BRCA1* and/or *BRCA2* genes. It may also be used for those individuals who have a known familial variant that is a large deletion or duplication. If ordering for familial analysis, a copy of the positive family member's report or genetic counseling letter is requested for documentation of the familial variant.
***BRCA1/2* Ashkenazi Jewish Profile (BRCA*ssure*®)**	485097	Screens for three founder variants in *BRCA*1 (c.68_69delAG and c.5266dupC) and *BRCA*2 (c.5946delT) genes in the Ashkenazi Jewish population. These variants are also known by their previous nomenlcature, namely 187delAG and 5382insC for the *BRCA1* and 6174delT for the *BRCA2* gene.
***BRCA1* Targeted Analysis (BRCA*ssure*®)**	485066	This test code is intended for those individuals who have a family member with a known *BRCA1* variant and wished to be tested only for that variant. A copy of the positive family member's laboratory report or genetic counseling letter documenting the variant is required for this testing. Only the specific region of the *BRCA1* gene containing the familial variant will be tested. If the familial variant is a large deletion or duplication of *BRCA1*, *BRCA1/2* Deletion/Duplication Analysis (BRCAssure®) (test code 252888) should be ordered. If there is no family member with a known *BRCA1* variant or if there is no documentation of the familial variant, *BRCA1/2* Comprehensive Analysis (BRCA*ssure*®) (test code 252911) should be ordered. Please call 800-345-GENE (4343) for more information regarding documentation requirements or other questions.

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Test Name	Test N°	Use
***BRCA2* Targeted Analysis (BRCA*ssure*®)**	485081	This test code is intended for those individuals who have a family member with a known *BRCA2* variant and wished to be tested only for that variant. A copy of the positive family member's laboratory report or genetic counseling letter documenting the variant is required for this testing. Only the specific region of the *BRCA2* gene containing the familial variant will be tested. If the familial variant is a large deletion or duplication of *BRCA2*, *BRCA1/2* Deletion/Duplication Analysis (BRCAssure®) (test code 252888) should be ordered. If there is no family member with a known *BRCA1* variant or if there is no documentation of the familial variant, *BRCA1/2* Comprehensive Analysis (BRCA*ssure*®) (testcode 252911) should be ordered. Please call 800-345-GENE (4343) for more information regarding documentation requirements or other questions.
***MLH1* Comprehensive Analysis**	511615	Can confirm a clinical diagnosis of HNPCC and allow early diagnosis in family members, guiding preventive measures. Hereditary nonpolyposis colorectal cancer (HNPCC) is an autosomal-dominant, genetically heterogeneous syndrome caused by heterozygous mutations in mismatch repair genes (MMR). HNPCC is estimated to account for 4% to 6% of colorectal cancer and is characterized by early onset, a predominant proximal location of colon cancer, multiple primary cancers, and significantly improved survival when compared stage for stage to sporadic colon cancer survival rates. HNPCC has been linked to variants in the genes *MLH1*, *MSH2*, *PMS2*, and *MSH6*, which are involved in DNA mismatch repair. Genetic testing can confirm the diagnosis of HNPCC and can also identify presymptomatic individuals among the patient's relatives.
***MLH1* Deletion/Duplication Analysis**	511690	This test is intended for individuals who have had previous negative sequencing of the *MLH1* gene and have not had previous deletion/duplication analysis or who have a family member with an identified large deletion or duplication of the *MLH1* gene. If testing for a known family mutation, please submit a copy of the laboratory report from the index family member documenting the familial mutation.
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): *MLH1* (Known Mutation)	511635	Identify who in a family harbors the familial variant and is at high risk of the disease and who does not harbor the familial variant and is not at increased risk of the disease. Family testing for known familial variants can identify presymptomatic carriers within affected families who are at high risk of developing the familial disease.
***MSH2* Comprehensive Analysis**	511632	Can confirm a clinical diagnosis of HNPCC and allow early diagnosis in family members, guiding preventive measures.
***MSH2* Deletion/Duplication Analysis**	511705	This test is intended for individuals who have had previous negative sequencing of the *MSH2* gene and have not had previous deletion/duplication analysis or who have a family member with an identified large deletion or duplication of the *MSH2* gene. If testing for a known family mutation, please submit a copy of the laboratory report from the index family member documenting the familial mutation.
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): *MSH2* (Known Mutation)	511750	Identify who in a family harbors the familial variant and is at high risk of the disease and who does not harbor the familial variant and is not at increased risk of the disease. Family testing for known familial variants can identify presymptomatic carriers within affected families who are at high risk of developing the familial disease.
***MSH6* Comprehensive Analysis**	511636	Can confirm a clinical diagnosis of HNPCC and allow early diagnosis in family members, guiding preventive measures.
***MSH6* Deletion/Duplication Analysis**	511720	This test is intended for individuals who have had previous negative sequencing of the *MSH6* gene and have not had previous deletion/duplication analysis or who have a family member with an identified large deletion or duplication of the *MSH6* gene. If testing for a known family mutation, please submit a copy of the laboratory report from the index family member documenting the familial mutation.

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Test Name	Test N°	Use
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): *MSH6* (Known Mutation)	511765	Identify who in a family harbors the familial variant and is at high risk of the disease and who does not harbor the familial variant and is not at increased risk of the disease. Family testing for known familial variants can identify presymptomatic carriers within affected families who are at high risk of developing the familial disease.
***PMS2* Comprehensive Analysis**	511630	Can confirm a clinical diagnosis of HNPCC and allow early diagnosis in family members, guiding preventive measures. Hereditary nonpolyposis colorectal cancer (HNPCC) is an autosomal-dominant, genetically heterogeneous syndrome caused by heterozygous mutations in mismatch repair genes (MMR). HNPCC is estimated to account for 4% to 6% of colorectal cancer and is characterized by early onset, a predominant proximal location of colon cancer, multiple primary cancers, and significantly improved survival when compared stage for stage to sporadic colon cancer survival rates. HNPCC has been linked to mutations in the genes *MLH1*, *MSH2*, *PMS2*, and *MSH6*, which are involved in DNA mismatch repair. Genetic testing can confirm the diagnosis of HNPCC and can also identify presymptomatic individuals among the patient's relatives.
***PMS2* Deletion/Duplication Analysis**	511725	This test is intended for individuals who have had previous negative sequencing of the *PMS2* gene and have not had previous deletion/duplication analysis or who have a family member with an identified large deletion or duplication of the *PMS2* gene. If testing for a known family mutation, please submit a copy of the laboratory report from the index family member documenting the familial mutation.
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): *PMS2* (Known Mutation)	511776	Identify who in a family harbors the familial variant and is at high risk of the disease and who does not harbor the familial variant and is not at increased risk of the disease. Family testing for known familial variants can identify presymptomatic carriers within affected families who are at high risk of developing the familial disease.
***MLH1/MSH2* Comprehensive Analysis**	511660	Can confirm a clinical diagnosis of HNPCC and allow early diagnosis in family members, guiding preventive measures. Hereditary nonpolyposis colorectal cancer (HNPCC) is an autosomal-dominant, genetically heterogeneous syndrome caused by heterozygous mutations in mismatch repair genes (MMR). HNPCC is estimated to account for 4% to 6% of colorectal cancer and is characterized by early onset, a predominant proximal location of colon cancer, multiple primary cancers, and significantly improved survival when compared stage for stage to sporadic colon cancer survival rates. HNPCC has been linked to mutations in the genes *MLH1*, *MSH2*, *PMS2*, *MSH6*, and *EPCAM*. Genetic testing can confirm the diagnosis of HNPCC and can also identify presymptomatic individuals among the patient's relatives.
***MLH1/MSH2/MSH6* Comprehensive Analysis**	511673	
***MLH1/MSH2/MSH6/PMS2* Comprehensive Analysis**	511700	
VistaSeq® Hereditary Cancer Panel	481220	VistaSeq℠ provides an assessment of inherited genetic variants within a panel of 27 genes known to be associated with hereditary cancer syndromes.
VistaSeq® Hereditary Cancer Panel Without *BRCA*	481240	The VistaSeq℠ Hereditary Cancer Panel Without *BRCA* provides an assessment of inherited genetic variants within a panel of 25 genes known to be associated with hereditary cancer syndromes. The test is intended for individuals who have already had a *BRCA1* and *BRCA2* gene assessment, but for whom results of that testing were negative and/or personal or family history warrant assessment of additional genes.

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Test Name	Test N°	Use
VistaSeq® Breast Cancer Panel	481319	These assays are intended for patients with a family history consistent with an inherited cancer syndrome.
VistaSeq® High/Moderate Risk Breast Cancer Panel	481452	
VistaSeq® GYN Cancer Panel	481330	
VistaSeq® Breast and GYN Cancer Panel	481341	
VistaSeq® High Risk Colorectal Cancer Panel	481352	
VistaSeq® Colorectal Cancer Panel	481363	
VistaSeq® Endocrine Cancer Panel	481374	
VistaSeq® Brain/CNS/PNS Cancer Panel	481386	
VistaSeq® Pancreatic Cancer Panel	481385	
VistaSeq® Renal Cell Cancer Panel	481407	
Cancer Genetics — Somatic Mutation Testing		
1p,19q Oncology FISH	510360	Confirmation/identification of cancer-related alterations (associated with oligoglioma).
Adult Acute Lymphoblastic Leukemia (ALL) Profile, FISH	511077	Diagnostic and prognostic test for acute lymphoblastic leukemia in the pediatric population; detection rate is improved from 50% with a chromosome study to 90% with fluorescence in situ hybridization (FISH)
Aggressive B-Cell Lymphoma Profile, FISH	510344	Diagnostic test for non-Hodgkin's lymphoma (NHL). Detects primary genetic changes associated with various types of NHL, including follicular lymphoma (FL), Burkitt's lymphoma (BL), mantle cell lymphoma (MCL), diffuse large B-cell lymphoma (DLBCL), and those with *BCL6* rearrangement.
ALK FISH, Non–Small-cell Lung Cancer	510950	Confirmation/identification of non–small-cell lung cancer. This is an FDA-approved test for the identification of NSCLC patients who may be eligible for treatment.
B-Cell Gene Rearrangements Profile, IgH and IgK	481222	This profile can be used to detect clonal B-cell immunoglobulin heavy chain (IgH) and immunoglobulin κ light chain (IgK) gene rearrangements in blood, bone marrow, and tissue specimens with combined B-cell clonality detection rate of 99%. The presence of a monoclonal gene rearrangement usually, but not always, reflects the presence of a B-lymphocytic neoplasm, while polyclonal gene rearrangement patterns are found in benign reactive condition. Thus, the results of these studies can assist in the diagnosis of lymphoproliferative disorders.
B-Cell, IgH Gene Rearrangements	480716	Detects IgH (immunoglobulin heavy chain) gene rearrangement. Could be used to identify clonal B-cell populations highly suggestive of B-cell malignancies, determine the lineage of leukemias and lymphomas, monitor and evaluate disease recurrence, and detect and assess residual disease.
B-Cell, IgK Gene Rearrangements	480812	This assay can be used to detect clonal immunoglobulin receptor kappa-chain gene rearrangements in blood, bone marrow, and tissue specimens with combined B-cell clonality detection rate of 94% to 99%. The presence of a monoclonal gene rearrangement usually, but not always, reflects the presence of a T-lymphocytic neoplasm, while polyclonal gene rearrangement patterns are found in benign reactive conditions. Thus, the results of these studies can assist in the diagnosis of lymphoproliferative disorders.
BCR-ABL1 Kinase Domain Mutation Analysis	480510	Mutations within the *BCR-ABL1* kinase domain in imatinib-treated chronic myeloid leukemia are the main mechanism of acquired resistance. The early detection of mutations should provide clinical benefit by allowing early intervention.

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Test Name	Test N°	Use
***BCR-ABL1*, Transcript Detection for Chronic Myelogenous Leukemia (CML) and Acute Lymphocytic Leukemia (ALL), Quantitative**	480481	This assay can detect three different types of *BCR-ABL1* fusion transcripts associated with CML, ALL, and AML:e13a2 (previously b2a2) and e14a2 (previously b3a2) (major breakpoint, p210), as well as e1a2 (minor breakpoint, p190). The e13a2 and e14a2 transcript values are titrated to the current International Scale (IS). The standardized baseline is 100% *BCR-ABL1* (IS) and major molecular response (MMR) is equivalent to 0.1% *BCR-ABL1* (IS) corresponding to a 3-log reduction. Results should be correlated with appropriate clinical and laboratory information as indicated.
Bladder Cancer FISH, Pathologist Review	130080	The assay is designed to detect aneuploidy for chromosomes 3, 7, 17, and loss of the 9p21 locus via fluorescence in situ hybridization (FISH) in urine specimens from subjects with transitional cell carcinoma of the bladder. This assay does not detect other chromosomal or genetic alterations. Results are intended for use as a noninvasive method of monitoring for tumor recurrence in conjunction with cystoscopy in patients previously diagnosed with bladder cancer. The clinical interpretation of test results should be evaluated within the context of the patient's medical history and other diagnostic laboratory test results.
***BRAF* Gene Mutation Analysis, Melanoma**	481110	The US Food and Drug Administration (FDA) has approved TKI inhibitor vemurafenib and debrafenib for the first-line treatment of patients with unresectable or metastatic melanoma whose tumors have a *BRAF* V600E mutation, and trametinib for tumors with either V600E or V600K mutations. These mutations make up greater than 90% of identified *BRAF* mutations. In addition, pembrolizumab and nivolumab have been approved by the FDA for treatment for disease progression after treatment with ipilimumab and V600 mutation positive patients with unresectable or metastatic melanoma with disease progression and prior treatment with a *BRAF* inhibitor. The NCCN guideline also suggests using both pembrolizumab and nivolumab as options for first-line treatment as both drugs have higher response rates and less toxicity compared to ipilimumab. *BRAF* is an important member of the mitogen-activated protein kinase (MAPK) pathway that influences cell proliferation. *BRAF* mutations are found in approximately 50% of melanoma tumors.
***BRAF* Gene Mutation Analysis**	481030	*BRAF* is an important member of the mitogen-activated protein kinase (MAPK) pathway that influences cell proliferation. This test will detect all V600 mutations of the BRAF oncogene frequently found in human cancers, such as melanoma, colorectal cancer, lung cancer, ovarian cancer, thyroid cancer, and hairy cell leukemia, allowing the determination of drug response, aiding the diagnosis and providing prognosis information. More than 90% of mutations are the V600E (c1799T>A) mutation, but other V600 mutations have been reported.This test can detect the following *BRAF* V600 mutations: V600E, V600E2, V600K, V600D, V600R, V600A, V600G, V600M, V600L.
Calreticulin (*CALR*) Mutation Analysis	489450	The detection of a *CALR* gene mutation aids in the specific diagnosis of a myeloproliferative neoplasm and helps distinguish this clonal disease from a benign, reactive, more indolent disease course with a lower thrombotic risk and longer overall survival (relative to those with a *JAK2* mutation).
***CEBPA* Mutation Analysis**	489170	The *CEBPA* (CCAAT/enhancer binding protein α) gene encodes a transcription factor important for granulocyte differentiation. *CEBPA* mutations are found in 6% to 15% of de novo acute myeloid leukemia (AML) and in 15% to 18% of AML with normal karyotypes. *CEBPA* mutations are associated with favorable prognosis in the absence of associated cytogenetic abnormalities and FLT3 internal duplication (FLT3-ITD). Germline *CEBPA* mutations are a cause of nonsyndromic, familial AML.
CHOP Oncology FISH	510349	Confirmation/identification of cancer-related alterations (associated with myxoid liposarcomas/round liposarcomas).
Chromosome Analysis, Leukemia/ Lymphoma	510999	Karyotyping, physical localization of copy number changes, and mapping of breakpoints involved in translocations.
Chromosome Analysis, Solid Tumor	510995	Detection of chromosomal abnormalities with subgroup-specific diagnostic and prognostic significance [eg, t(11;22) in Ewing sarcoma].
Chronic Lymphocytic Leukemia (CLL) Profile, FISH	510340	Diagnostic and prognostic test for chronic lymphocytic leukemia; detection rate is improved from 45% with a chromosome study to 80% with fluorescence in situ hybridization (FISH). Differentiates CLL from MCL.

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Test Name	Test N°	Use
Chronic Myelogenous Leukemia (CML) Profile: Chromosome Analysis and *BCR-ABL*, FISH	150500	Confirm the diagnosis of chronic myelogenous leukemia; establish the chronic-phase karyotype for comparison with blast crisis alterations; monitor residual disease.
c-KIT Mutation Analysis in Tumors of Hematopoietic Tissue	480940	c-KIT is a proto-oncogene that encodes a type III trans-membrane tyrosine kinase. c-KIT and its ligand stem cell factor have a key role in survival, proliferation, differentiation, and functional activation of hematopoietic progenitor cells. c-KIT mutations are reported in nearly all systemic mastocytosis, 20% to 40% core-binding factor (CBF) acute myeloid leukemia (AML), and approximately 20% high-grade myelodysplastic syndrome (MDS) and MDS-derived AML. c-KIT mutation in AML confers increased risk of relapse and decreased overall survival. Tyrosine kinase inhibitor, such as imatinib, has been evaluated to treat systemic mastocytosis and c-KIT-positive AML and MDS, and it was found effective as a single reagent or combination therapy.
CML FISH Reflex to *JAK2*^{V617F} Mutation Analysis, Qualitative, With Reflex to CALR Mutation Analysis, *JAK2* Exon 12-15 Mutation Analysis and MPL Mutation Analysis	511994	Confirm the diagnosis of CML; establish the chronic phase karyotype for comparison with blast crisis alterations; monitor residual disease.
EGFR Oncology FISH	510355	Confirmation/identification of cancer-related alterations (for lung and brain cancer).
***EPCAM* Deletion/Duplication Analysis**	511654	Confirm a clinical diagnosis of hereditary nonpolyposis colorectal cancer (HNPCC) and allow early diagnosis in family members, guiding preventive measures.
Epidermal Growth Factor Receptor (EGFR) Mutation Analysis for Non–Small-cell Lung Cancer (Single-base Extension)	489360	The presence of a somatic EGFR mutation is significantly associated with response to gefitinib and erlotinib, and it is strongly predictive of prolonged survival in NSCLC patients.
EWSR1 Oncology FISH	510379	Confirmation/identification of cancer-related alterations (Ewing sarcoma).
FKHR Oncology FISH	510371	Confirmation/identification of cancer-related alterations (alveolar rhabomyosarcoma).
Fluorescence in situ Hybridization (FISH), Oncology	510669	Confirmation/identification of leukemia/lymphoma-related alterations; leukemia/lymphoma monitoring of residual disease, transplants, and indolent clones (see list or call lab for probe availability).
Fluorescence in situ Hybridization (FISH), Paraffin Block	510825	For specific FISH probe analysis of tissue specimens. (Call the laboratory for a list of available probes.)
HER-2/CEP17, FISH	483248	Qualitative determination of HER-2/*neu* gene amplification; prognostic information regarding risk of recurrence and disease-related death; predict response to therapies, including targeted immunotherapy.
***JAK2* Exon 12, 13, 14 and 15 Mutation Analysis**	115101	The *JAK2*^{V617F} (exon 14) mutation analysis can be used in conjunction with bone marrow histology and cytogenetic analysis to assist in the diagnosis of myeloproliferative neoplasma (MPN). The *JAK2*^{V617F} mutation is found in almost all patients with polycythemia vera (PV) and in nearly one-half of those with idiopathic myelofibrosis (IMF) and with essential thrombocythemia (ET). A small percentage (~3.3%) of *JAK2* mutation positive patient contain other non-*V617F* mutations within exons 12 to 15.
***JAK2*^{V617F} Mutation Analysis, Qualitative, With Reflex to CALR Mutation Analysis, *JAK2* Exon 12-15 Mutation Analysis and MPL Mutation Analysis**	489421	This test will assess for the *JAK2*^{V617F} (exon 14) mutation first and will reflex to CALR mutation analysis, *JAK2* exon 12 to 15 mutation analysis and MPL mutation analysis when the *JAK2*^{V617F} mutation is negative.
***JAK2*^{V617F} Mutation Analysis, Quantitative**	481020	The quantitative real-time PCR assay detects *V617F* mutation (c.1849 G>T) observed in approximately 95% polycythemia vera (pv), 55% essential thrombocythemia (ET), and 55% primary myelofibrosis (PMF). It is also infrequently present (3% to 5%) in myelodysplastic syndrome, chronic myelomonocytic leukemia, and other atypical chronic myeloid disorders.

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Test Name	Test N°	Use
JAK2^{V617F} Mutation Analysis, Qualitative	489200	The *JAK2*^{V617F} (exon 14) mutation analysis can be used in conjunction with bone marrow histology and cytogenetic analysis to assist in the diagnosis of myeloproliferative neoplasms (MPNs). The *JAK2*^{V617F} mutation is found in almost all patients with polycythemia vera (PV) and in nearly one-half of those with idiopathic myelofibrosis (IMF) and with essential thrombocythemia (ET). The *V617F* mutation has also been detected, although infrequently, in other myeloid disorders, such as chronic myelomonocytic leukemia and chronic neutrophilic leukemia.
Microsatellite Instability Analysis	511855	Identify tumors with microsatellite instability. High-frequency microsatellite instability (MSI-H) is associated with Lynch syndrome, but it is also found in 15% to 20% of sporadic colorectal and endometrial cancers. Lynch syndrome is an autosomal-dominant inherited cancer syndrome that predisposes to colorectal, endometrial, gastric, ovarian, upper urinary tract, and other cancers.
MGMT (O^6-Methylguanine-DNA Methyltransferase) Gene Methylation Assay	489280	Approximately 40% to 50% of glioblastoma multiforme (GBM) tumors exhibit MGMT gene methylation. Retrospective studies have shown that detection of MGMT promoter methylation in tumor samples is associated with an increased likelihood of a favorable response to temozolomide (Temodar®).
MPL Mutation Analysis	489150	MPL (myeloproliferative leukemia virus oncogene homology) W515 mutations are present in patients with primary myelofibrosis (PMF) and essential thrombocythemia (ET) at a frequency of approximately 5% and 1%, respectively. The S505 mutation is detected in patients with hereditary thrombocythemia.
Multiple Myeloma (MM) Profile, FISH	510325	Diagnostic test for multiple myeloma. Plasma cell enrichment diagnosis increased as much as 50% to 100%. The FISH results on the enriched assay should not be used as a quantitative assay, since the abnormal cells do not represent the percentage of abnormal cells in the aspirate.
Multiple Myeloma Enrichment SNP Microarray–Oncology (Reveal®)	510195	Detects chromosomal imbalance that may be present in neoplastic clonal evolution; provides detection of acquired uniparental disomy of any chromosome.
MYCN Oncology FISH	510945	Confirmation/identification of leukemia/lymphoma-related alterations; leukemia/lymphoma monitoring of residual disease, transplants, and indolent clones (see list or call lab for probe availability)
Myelodysplastic Syndrome (MDS), FISH	511060	Diagnostic test for myelodysplastic syndrome. The principal use is for interphase analysis of cases with no (or low) mitotic activity in cytogenetic analysis or interphase analysis from blood in cases of inaspirable bone marrow. Detection rate is approximately 80% of clones detected in cytogenetic analysis.
Myeloproliferative Neoplasms / Chronic Myelogenous Leukemia (MPN / CML), FISH	511425	Confirmation/identification of chromosome abnormalities in interphase nuclei. Leukemia monitoring of residual disease.
Myeloproliferative Neoplasms With Hypereosinophilia (MPN / HES), FISH	511444	
Non–Small-cell Lung Cancer (NSCLC) Therapeutic Profile II	388103	Non–small-cell lung cancer (NSCLC) is the leading cause of death from cancer in both men and women in the US. A subgroup of NSCLC patients has shown clinical responsiveness to the epidermal growth factor receptor (EGFR) inhibitors gefitinib (Iressa®) and erlotinib (Tarceva®). In the majority of patients with highly responsive tumors, the tumor contains somatic mutations within the EGFR tyrosine kinase domain. The presence of a somatic EGFR mutation is significantly associated with deferential responsiveness or resistance to gefitinib and erlotinib, and is strongly predictive of prolonged survival in NSCLC patients.KRAS mutations in NSCLC are predictive of lack of therapeutic efficacy with EGFR tyrosine kinase inhibitor (erlotinib and gefitinib). Patients with mutations appear to have a shorter survival than patients with wild-type KRAS. A rearrangement of ALK is reported to be associated with the development of NSCLC. The FDA-approved ALK FISH probe is used to identify gene rearrangements involving the ALK gene in patients with NSCLC who are eligible for treatment with crizotinib (Xalkori®).

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Test Name	Test N°	Use
NPM1 Mutation Analysis	489140	NPM1 (nucleophosmin) mutation is one of the most common recurring genetic lesions in acute myeloid leukemia (AML). This AML type frequently has myelomonocytic or monocytic features and typically presents de novo in older adults with a normal karyotype. Prevalence increases with age, occurring in 2% to 8% of childhood AML and 27% to 35% of adult AML. The most common mutation, insertion at nucleotide position 959 (exon 12), accounts for 90% to 95% of NPM1 mutations. NPM1 mutations in the absence of FLT3-ITD identify a prognostically favorable subgroup.
p53 Oncology FISH	510940	Confirmation/identification of leukemia/lymphoma-related alterations; leukemia/lymphoma monitoring of residual disease, transplants, and indolent clones (see list or call lab for probe availability)
PIK3CA Mutation Analysis, Breast Cancer	485113	The therascreen PIK3CA RGQ RT-PCR Kit is a real-time, qualitative PCR assay for the detection of 11 mutations in the phosphatidyl 3-kinases catalytic subunit alpha (PIK3CA) gene (Exon 7: C420R; Exon 9: E542K; E545A, E545D [1635G>T only], E545G, E545K, Q546E, Q546R; and Exon 20: H1047L, H1047R, H1047Y) using genomic DNA (gDNA) extracted from formalin-fixed paraffin-embedded (FFPE) breast tumor tissue. The test is intended to aid clinicians in identifying breast cancer patients who may be eligible for treatment with PIQRAY(R) (alpelisib) based on a PIK3CA Mutation Detected result. Patients whose FFPE tissue produce a positive therascreen PIK3CA RGQ PCR Kit test result for the presence of one or more PIK3CA mutations are eligible for treatment with PIQRAY® (alpelisib).
PML-RARA Transcript Detection for Acute Promyelocytic Leukemia, Quantitative	510840	The translocation t(15;17) (q22;q21) is the prototype rearrangement found in the vast majority of acute promyelocytic leukemia (APL), being found in >95% of APL cases. In this chromosomal rearrangement, the retinoic acid receptor (*RARA*) gene on chromosome 17 is fused with the PMLgene on chromosome 15. There are three common breakpoints within the *PML* gene, bcr1(intron 6), bcr2 (exon 6), and bcr3 (intron 3). All breakpoints fuse a portion of the *PML* gene to a consistent breakpoint region within the *RARA* gene. This assay will detect the *PML-RARA* transcripts associated with the *bcr1*, *bcr2*, and *bcr3* breakpoints using real-time RT-PCR in order to assist in the diagnosis and monitoring of APL. The results are reported as a normalized ratio of %PML-RARA copies/ABL1 copies. In vitro studies have indicated that this assay has an analytical sensitivity that allows for the detection of 0.001% PML-RARA/ABL1.
Prostate Cancer Gene 3 (PCA3)	489160	Prostate cancer gene 3 (PCA3) is strongly expressed in 95% of primary prostate cancer specimens. The PCA3 test is indicated for use in conjunction with other patient information to aid in the decision for repeat biopsy in men age 50 or older who have had one or more previous negative prostate biopsies and for whom a repeat biopsy would be recommended by a urologist based on current standard of care. The PCA3 result provides a risk assessment of a positive biopsy. This assay should not be used for men with atypical small acinar proliferation (ASAP) on their most recent biopsy. Men with ASAP on their most recent biopsy should be treated in accordance with current medical guidelines.
RB1 Oncology FISH	510374	Confirmation/identification of leukemia/lymphoma-related alterations; leukemia/lymphoma monitoring of residual disease, transplants, and indolent clones (see list or call lab for probe availability).
RET Oncology FISH	510315	Confirmation/identification of cancer-related alterations in non–small-cell lung cancer.
SNP Microarray – Oncology (Reveal®)	510146	High-resolution detection of genomic imbalance that may be present in neoplastic clonal evolution; provides detection of acquired uniparental disomy associated with cancer gene mutations.
SYT Oncology FISH	510384	Confirmation/identification of leukemia/lymphoma-related alterations; leukemia/lymphoma monitoring of residual disease, transplants, and indolent clones (see list or call lab for probe availability).
T-Cell Receptor Gene Rearrangements Profile, γ and β	481080 (combines 480985 and 480708)	See 480985 and 480708.

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Test Name	Test N°	Use
T-Cell Receptor β-Chain Gene Rearrangements	480985	Detects clonal T-cell receptor β-chain gene rearrangements in blood, bone marrow, and tissue specimens. The presence of a monoclonal gene rearrangement usually, but not always, reflects the presence of a T-lymphocytic neoplasm, while polyclonal gene rearrangement patterns are found in benign reactive conditions. Thus, the results of these studies can assist in the diagnosis of lymphoproliferative disorders.
T-Cell Receptor γ-Chain Gene Rearrangements	480708	Detects T-cell receptor γ-chain gene rearrangement. It could be used to identify clonal T-cell populations highly suggestive of T-cell malignancies, determine the lineage of leukemias and lymphomas, monitor and evaluate disease recurrence, and detect and assess residual disease.
Pharmacogenetics		
Cytochrome P450 *2C9* Genotyping	511893	This test provides genotype information for CYP2C9.
Cytochrome P450 *2C19* Genotyping	511675	The xTAG® CYP2C19 Kit v3 is a qualitative genotyping assay, which can be used as an aid to clinicians in determining therapeutic strategy for the therapeutics that are metabolized by the CYP2C19 gene product. CYP2C19 is involved in the metabolism of drugs including clopidogrel, anticonvulsants, diazepam, omeprazole, tricyclic antidepressants and proton pump inhibitors. The CYP2C19 gene is highly polymorphic. Many alleles of CYP2C19 encode enzymes that have non-functional, decreased or increased enzyme activity compared to wild-type. Depending on the combination of alleles in an individual, drug-metabolizing phenotypes associated with the CYP2C19 enzyme can vary.
Cytochrome P450 *2D6* Genotyping	511230	The xTAG® CYP2D6 Kit v3 is a qualitative genotyping assay, which can be used as an aid to clinicians in determining therapeutic stategy for therapeutics that are metabolized by the CYP2D6 gene product. CYP2D6 is involved in the metabolism of more than 65 commonly used drugs including β-blockers, antipsychotics, antidepressants, analgesics, and antiarrythmics. The CYP2D6 gene is highly polymorphic. Many alleles of 2D6 encode enzymes that have reduced or no function compared to the wild-type enzyme. Individuals can also have gene rearrangements with more than two copies of the CYP2D6 gene (gene duplication) or absence of both copies (gene deletion). Depending on the combination of alleles in an individual, drug-metabolizing phenotypes associated with the CYP2D6 enzyme can vary.
Cytochrome P450 *2D6/2C19* Genotyping	511905 (combines 511230 and 511675)	See 511230 and 511675.
DPD 5-Fluorouracil Toxicity	511176	Variability in response (efficacy and toxicity) to 5-fluorouracil (5-FU) chemotherapy has been linked to the rate-limiting enzyme in the drug's catabolic pathway, known as dihydropyrimidine dehydrogenase (DPD).
IFNL3 (IL28B) Genotyping (rs12979860)	480630	This assay is used for genotyping IL-28B rs12979860.
Thiopurine Methyltransferase (TPMT), Enzyme Activity, Erythrocytes	510750	Determination of TPMT levels that may be associated with toxicity of anticancer and anti-inflammatory drug.
UGT1A1 Irinotecan Toxicity	511200	Irinotecan (Camptosar®) is used, or under evaluation, in a broad spectrum of solid tumors, and is often prescribed for treating patients with metastatic cancer of the colon or rectum, especially when 5-fluorouracil treatment has not been entirely successful. 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. *UGT1A1* variants have also been reported in patients with disorders of bilirubin metabolism, such as Crigler-Najjar Types I and II, as well as Gilbert syndrome. Between 80% to 100% of Caucasian patients with Gilbert syndrome are reported to have either one or two copies of *UGT1A1**28. G71R (*6), a *UGT1A1* variant reported in Asian patients with Gilbert syndrome, is not detected by this assay.

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1p,19q Oncology FISH	510360
3-O-Methyldopa (Plasma)	620176
Acetylcholinesterase (AChE), Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)*	510354
Acylcarnitine Profile, Quantitative, Plasma	070228
Adult Acute Lymphoblastic Leukemia (ALL) Profile, FISH	511077
Aggressive B-Cell Lymphoma Profile, FISH	510344
ALK FISH, Non–Small-cell Lung Cancer	510950
Alpha Aminoadipic Semialdehyde (Urine)	620046
Amino Acid Profile, Quantitative, Cerebrospinal Fluid	700180
Amino Acid Profile, Quantitative, Plasma	700068
Amino Acid Profile, Quantitative, Urine	700140
Ammonia, Plasma	007054
Angelman and Prader-Willi Syndromes, DNA Analysis*	511210
α1-Antitrypsin Deficiency, DNA Analysis*	511881
Arylsulfatase A Deficiency, Leukocytes	402396
Ashkenazi Jewish Carrier Profile	333561
Ashkenazi Jewish Carrier Profile Plus	332859
Autoimmune Polyglandular Syndrome Type 1 (APS1/APECED): AIRE (Full Gene Sequencing)	252532
Autoimmune Polyglandular Syndrome Type 1 (APS1/APECED): AIRE (Known Mutation)	252737
B-Cell Gene Rearrangement Profile, IgH and IgK	481222
B-Cell, IgH Gene Rearrangements	480716
B-Cell, IgK Gene Rearrangements	480812
BCR-ABL1 Kinase Domain Mutation Analysis	480510
BCR-ABL1, Transcript Detection for Chronic Myelogenous Leukemia (CML) and Acute Lymphocytic Leukemia (ALL), Quantitative	480481
Bladder Cancer FISH, Pathologist Review	130080
Bloom Syndrome, DNA Analysis*	512145
BRAF Gene Mutation Analysis, Melanoma	481110
BRAF Gene Mutation Analysis	481030
BRCA1 Targeted Analysis (BRCAssure®)	485066
BRCA2 Targeted Analysis (BRCAssure®)	485081
BRCA1/2 Ashkenazi Jewish Profile (BRCAssure®)	485097
BRCA1/2 Comprehensive Analysis (BRCAssure®)	485030
BRCA1/2 Deletion/Duplication Analysis (BRCAssure®)	485050
C9orf72 Genetic Testing (Repeat Expansion)	620017
Calreticulin (CALR) Mutation Analysis	489450
Canavan Disease, DNA Analysis*	511147
Carnitine, Total and Free	706500
CEBPA Mutation Analysis	489170
CHOP Oncology FISH	510349
Chromosome Analysis and AFP, Amniotic Fluid*	510185
Chromosome Analysis, AFP, AChE, Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)*	511580
Chromosome Analysis, Amniotic Fluid*	052040
Chromosome Analysis, Amniotic Fluid With Reflex to SNP Microarray (Reveal®)*	052104
Chromosome Analysis, Chorionic Villi Biopsy With Reflex to SNP Microarray (Reveal®)	511033
Chromosome Analysis, Instability Syndrome	511045
Chromosome Analysis, Leukemia/Lymphoma	510999
Chromosome Analysis, Prenatal Cordocentesis and Fetal Hemoglobin	511025
Chromosome Analysis, Products of Conception (POC) With Reflex to SNP Microarray (Reveal®)	052065
Chromosome Analysis, Solid Tumor	510995
Chromosome Analysis, Tissue Biopsies (Products of Conception, Skin)	052052
Chromosome Analysis With Reflex to SNP Microarray – Pediatric (Reveal®)	052045
Chromosome Five-cell Count Plus Microarray (Reveal®), Amniotic Fluid	511590
Chromosome Five-cell Count Plus Microarray (Reveal®), CVS	511555
Chromosome Five-cell Count Plus Microarray (Reveal®), Whole Blood	511535
Chronic Granulomatous Disease (CGD): CYBB (Full Gene Sequencing)	252529
Chronic Granulomatous Disease (CGD): CYBB (Known Mutation)	252733
Chronic Lymphocytic Leukemia (CLL) Profile, FISH	510340
Chronic Myelogenous Leukemia (CML) Profile: Chromosome Analysis and BCR-ABL, FISH	150500

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Comprehensive List of Assays

c-KIT Mutation Analysis in Tumors of Hematopoietic Tissue	480940
CML FISH Reflex to JAK2V617F Mutation Analysis, Qualitative, With Reflex to CALR Mutation Analysis, JAK2 Exon 12-15 Mutation Analysis and MPL Mutation Analysis	511994
Comprehensive Spinocerebellar Ataxia Repeat Expansion Panel	620167
Congenital Adrenal Hyperplasia (CAH) 21-Hydroxylase (CYP21) Mutation (Endocrine Sciences)	500768
Creatine and Guanidinoacetate (Plasma)	620180
Creatine and Guanidinoacetate (Urine)	620170
Cystic Fibrosis (CF) Profile, 97 Mutations, CFplus®	450020
Cystic Fibrosis (CF) Profile, 97 Mutations, CFplus®, Fetal Analysis*	480819
Cystic Fibrosis (CF) Profile, 32 Mutations, DNA Analysis	480533
Cystic Fibrosis (CF) Profile, DNA Analysis and 5T Allele Genotyping	480555
Cystic Fibrosis (CF) Profile, 32 Mutations, Fetal Analysis*	480541
Cytochrome P450 2C9 Genotyping	511893
Cytochrome P450 2C19 Genotyping	511675
Cytochrome P450 2D6 Genotyping	511230
Cytochrome P450 2D6/2C19 Genotyping	511905
Dihydrolipoamide Dehydrogenase (DLD)*	450080
DPD 5-Fluorouracil Toxicity	511176
DRPLA (ATN1) Genetic Testing (Repeat Expansion)	620158
EGFR Oncology FISH	510355
Enzyme Biotinidase Deficiency	402362
EPCAM Deletion/Duplication Analysis	511654
Epidermal Growth Factor Receptor (EGFR) Mutation Analysis for Non–Small-cell Lung Cancer (Single-base Extension)	489360
EWSR1 Oncology FISH	510379
α-Galactosidase A Deficiency, Leukocytes	402388
Factor II (Prothrombin), DNA Analysis	511162
Factor V Leiden Mutation Analysis	511154
Factor V Leiden With Reflex to R2	503853
Factor V R2 DNA Analysis	503940
Familial Dysautonomia, DNA Analysis*	511352
Familial Hyperinsulinism (FHI)*	450070
Fanconi Anemia (Type C), DNA Analysis*	511212
FBN1 (Marfan Syndrome) Full Gene Sequencing	452028
α-Fetoprotein (AFP) Tetra Profile	017319
α-Fetoprotein (AFP), AChE, Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)*	510305
α-Fetoprotein (AFP), Amniotic Fluid*	002428
α-Fetoprotein (AFP), Maternal Serum for Open Spina Bifida	010801
First Trimester Screen With Nuchal Translucency	017500
FKHR Oncology FISH	510371
Fluorescence in situ Hybridization (FISH), Oncology	510669
Fluorescence in situ Hybridization (FISH), Paraffin Block	510825
Fragile X Syndrome, DNA Analysis, Prenatal With Southern Blot Analysis*	510300
Fragile X Syndrome, PCR With Reflex to Southern Blot	511919
Fragile X, PCR and Southern Blot Analysis	511655
Friedreich Ataxia Genetic Testing (Trinucleotide Repeat Expansion)	620077
α-Galactosidase A Deficiency (Full Gene Sequencing)	252225
α-Galactosidase A Deficiency (Known Mutation)	252230
β-Galactosidase Deficiency, Leukocytes	402370
Gaucher Disease, DNA Analysis*	511048
GeneSeq®: Cardio-Early-onset Coronary Artery Disease/ Familial Hypercholesterolemia Profile	451416
GeneSeq®: Cardio-Familial Aortopathy Profile	451432
GeneSeq®: Cardio-Familial Arrhythmia Profile	451412
GeneSeq®: Cardio-Familial Cardiomyopathy Profile	451422
GeneSeq®: Cardio-Familial Congenital Heart Disease Profile	451402
GeneSeq®: Cardio-Familial Hypercholesterolemia Profile	452040
GeneSeq®: Cardio-Gene Specific Sequencing	452053
GeneSeq®: Cardio-Noonan Syndrome / RASopathies Profile	451441
GeneSeq® PLUS	630068
GeneSeq® PLUS, Prenatal	630119
GeneSeq® PLUS without VUS	630085
GeneSeq® PLUS without VUS, Prenatal	630102
GJB2 Sequencing, Full Gene Sequencing*	511405
GJB2 Sequencing, Family-targeted (Single Exon Sequencing–Known Mutation)*	511414

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Glycogen Storage Disease 1a*	511290
HER-2/CEP17, FISH	483248
Hereditary Hemochromatosis, DNA Analysis	511345
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): MLH1 (Known Mutation)	511635
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): MSH2 (Known Mutation)	511750
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): MSH6 (Known Mutation)	511765
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): PMS2 (Known Mutation)	511776
Huntington Disease (HTT) Genetic Testing (Repeat Expansion)	620016
Hyper-IgE Syndrome (HIES): STAT3 (Full Gene Sequencing)	252449
Hyper-IgE Syndrome (HIES): STAT3 (Known Mutation)	252680
Hyper-IgM Syndrome (HIGM): (AICDA for HIGM2) (Full Gene Sequencing)	252425
Hyper-IgM Syndrome (HIGM): (AICDA for HIGM2) (Known Mutation)	252663
Hyper-IgM Syndrome (HIGM): (CD40 [TNFRSF5] for HIGM3) (Full Gene Sequencing)	252432
Hyper-IgM Syndrome (HIGM): (CD40 [TNFRSF5] for HIGM3) (Known Mutation)	252670
Hyper-IgM Syndrome (HIGM): (CD40LG [TNFSF5] for HIGM1) (Full Gene Sequencing)	252435
Hyper-IgM Syndrome (HIGM): (CD40LG [TNFSF5] for HIGM1) (Known Mutation)	252673
Hyper-IgM Syndrome (HIGM): (UNG for HIGM5) (Full Gene Sequencing)	252428
Hyper-IgM Syndrome (HIGM): (UNG for HIGM5) (Known Mutation)	252666
Hyper-IgM Syndrome (HIGM): Four-gene Profile (AICDA, UNG, CD40, CD40LG) (Full Gene Sequencing)	252446
Hyper-IgM Syndrome (HIGM): Three-gene Profile (AICDA, UNG, CD40) (Full Gene Sequencing)	252442
Hyper-IgM Syndrome (HIGM): Two-gene Profile (AICDA, UNG) (Full Gene Sequencing)	252439
Hypohidrotic Ectodermal Dysplasia With Immune Deficiency (HED-ID): IKBKG (NEMO) (Full Gene Sequencing)	252539
Hypohidrotic Ectodermal Dysplasia With Immune Deficiency (HED-ID): IKBKG (NEMO) (Known Mutation)	252744
Infertility-Male, Y Deletion Analysis	512053

Inheritest® 500 PLUS Panel	630049
Inheritest® 500 PLUS with Repro Partners Report	630217
Inheritest® Carrier Screen, Ashkenazi Jewish Panel (48 Genes)	451920
Inheritest® Carrier Screen, Comprehensive Panel (144 Genes)	451950
Inheritest® Core Panel	451964
Inheritest® Gene-specific Sequencing, NGS	451910
Inheritest® Carrier Screen, Society-guided Panel (14 Genes)	451960
InSight: Prenatal Amnio Aneuploid (FISH) Testing for Chromosomes 13, 18, 21, and XY	511894
Integrated 1	017100
Integrated 2	017170
Interferon-γ Receptor Deficiency: IFNGR1 (Full Gene Sequencing)	252519
Interferon-γ Receptor Deficiency: IFNGR1 (Known Mutation)	252727
Interferon-γ Receptor Deficiency: IFNGR2 (Full Gene Sequencing)	252522
Interferon-γ Receptor Deficiency: IFNGR2 (Known Mutation)	252730
Interferon-γ Receptor Deficiency: Two-gene Profile (IFNGR1, IFNGR2) (Full Gene Sequencing)	252525
IFNL3 (IL28B) Genotyping (rs12979860)	480630
JAK2 Exon 12, 13, 14, and 15 Mutation Analysis	115101
JAK2V617F Mutation Analysis, Qualitative, With Reflex to CALR Mutation Analysis, JAK2 Exon 12-15 Mutation Analysis and MPL Mutation Analysis	489421
JAK2V617F Mutation Analysis, Quantitative	481020
JAK2V617F Mutation Analysis, Qualitative	489200
Joubert Syndrome Type II, DNA Analysis*	511490
Lactate (CSF)	620044
Lactic Acid, Plasma	004770
Maple Syrup Urine Disease Carrier Test, DNA*	511310
Maternal Cell Contamination*	511402
MaterniT Genome	451941
MaterniT21 Genome Add On (GENOME-Flex)	452104
MaterniT21 Genome Add On Redraw (GENOME-Flex)	452114
MaterniT21 Genome NO Gender	452106
MaterniT21 PLUS Core (chr21,18,13) NO Gender	451951

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MaterniT21 PLUS Core (chr21,18,13,sex)	451927
MaterniT21 PLUS Core + ESS	451931
MaterniT21 PLUS Core + ESS + SCA	451937
MaterniT21 PLUS Core + ESS + SCA, NO Gender	452122
MaterniT21 PLUS Core + ESS, NO Gender	452136
MaterniT21 PLUS Core + SCA	451934
MaterniT21 PLUS Core + SCA, NO Gender	452112
Methylenetetrahydrofolate Reductase (MTHFR) Thermolabile Variant, DNA Analysis	511238
Methylmalonic Acid, Serum or Plasma	706961
Methylmalonic Acid, Urine	716365
Microdeletion Syndromes*, FISH	510770
Microsatellite Instability Analysis	511855
MGMT (O6-Methylguanine-DNA Methyltransferase) Gene Methylation Assay	489280
MLH1 Comprehensive Analysis	511615
MLH1 Deletion/Duplication Analysis	511690
MLH1/MSH2 Comprehensive Analysis	511660
MLH1/MSH2/MSH6 Comprehensive Analysis	511673
MLH1/MSH2/MSH6/PMS2 Comprehensive Analysis	511700
MPL Mutation Analysis	489150
Myeloproliferative Neoplasms / Chronic Myelogenous Leukemia (MPN / CML), FISH	511425
Myeloproliferative Neoplasms With Hypereosinophilia (MPN / HES), FISH	511444
MSH2 Comprehensive Analysis	511632
MSH2 Deletion/Duplication Analysis	511705
MSH6 Comprehensive Analysis	511636
MSH6 Deletion/Duplication Analysis	511720
Mucolipidosis Type IV Mutation Detection*	511386
Multiple Myeloma (MM) Profile, FISH	510325
Multiple Myeloma Enrichment SNP Microarray–Oncology (Reveal®)	510195
Mutation-specific Sequencing, Whole Blood	451382
Mutation-specific Sequencing, Prenatal	451385
MYCN Oncology FISH	510945
Myelodysplastic Syndrome (MDS), FISH	511060
Myotonic Dystrophy 1 (DMPK) Genetic Testing (Repeat Expansion)	620084
Myotonic Dystrophy 2 (ZNF9/CNBP) Genetic Testing (Repeat Expansion)	620087
Nemaline Myopathy*	450040
NeuroSURE® Metabolites: 5-Methyltetrahydrofolate (CSF)	620008
NeuroSURE® Metabolites: Alpha Aminoadipic Semialdehyde, Cerebrospinal Fluid (CSF)	620037
NeuroSURE® Metabolites: Neopterin (CSF)	620009
NeuroSURE® Metabolites: Neopterin/Tetrahydrobiopterin (CSF)	620010
NeuroSURE® Metabolites: Neurotransmitter Metabolites (5 HIAA, HVA, 30MD) (CSF)	620011
NeuroSURE® Metabolites: Pyridoxal 5'-phosphate, Cerebrospinal Fluid (CSF)	620034
NeuroSURE® Metabolites: Sialic Acid, Cerebrospinal Fluid (CSF)	620036
NeuroSURE® Metabolites: Succinyladenosine, Cerebrospinal Fluid (CSF)	620035
NeuroSURE® Metabolites: Thymidine Phosphorylase Enzyme Analysis (Blood)	620038
Niemann-Pick Disease, DNA Analysis*	511329
Non-Small-cell Lung Cancer (NSCLC) Therapeutic Profile II	388103
NPM1 Mutation Analysis	489140
Organic Acid Analysis, Urine	716720
Orotic Acid, Urine	007010
p53 Oncology FISH	510940
PIK3CA Mutation Analysis, Breast Cancer	485113
PML-RARA Transcript Detection for Acute Promyelocytic Leukemia, Quantitative	510840
PMP22 MLPA Deletion/Duplication Analysis	620081
PMS2 Comprehensive Analysis	511630
PMS2 Deletion/Duplication Analysis	511725
Prenatal Aneuploid Evaluation, Chorionic Villus Sampling*, FISH	510960
Prenatal Noonan Syndrome	451890
Prostate Cancer Gene 3 (PCA3)	489160

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Pyruvate (CSF)	620045
Pyruvic Acid, Whole Blood	004788
RB1 Oncology FISH	510374
RET Oncology FISH	510315
SCA1 (ATXN1) Genetic Testing (Repeat Expansion)	620114
SCA2 (ATXN2) Genetic Testing (Repeat Expansion)	620118
SCA3 (ATXN3) Genetic Testing (Repeat Expansion)	620123
SCA6 (CACNA1A) Genetic Testing (Repeat Expansion)	620127
SCA7 (ATXN7) Genetic Testing (Repeat Expansion)	620131
SCA8 (ATXN8) Genetic Testing (Repeat Expansion)	620135
SCA10 (ATXN10) Genetic Testing (Repeat Expansion)	620140
SCA12 (PPP2R2B) Genetic Testing (Repeat Expansion)	620144
SCA17 (TBP) Genetic Testing (Repeat Expansion)	620149
SCA36 (NOP56) Genetic Testing (Repeat Expansion)	620154
SCN1A Sequencing, Full Gene	511236
SCN1A Family-targeted Sequencing	511274
Sequential 1	017700
Sequential 2	017750
Serum Integrated 1	017200
Serum Integrated 2	017270
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): DCLRE1C (Artemis) for RS-SCID or SCIDA (Full Gene Sequencing)	252492
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): DCLRE1C (Artemis) for RS-SCID or SCIDA (Known Mutation)	252723
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Eight-gene Profile (IL2RG, JAK3, RAG1, RAG2, IL7R, ADA, CD3D, CD3E) (Full Gene Sequencing)	252513
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Nine-gene Profile (IL2RG, JAK3, RAG1, RAG2, IL7R, ADA, CD3D, CD3E, DCLREC1C [Artemis]) (Full Gene Sequencing)	252516
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): RAG1 (Full Gene Sequencing)	252470
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): RAG1 (Known Mutation)	252701
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): RAG1, RAG2, DCLRE1C (Artemis) (Full Gene Sequencing)	252503
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): RAG2 (Full Gene Sequencing)	252472
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): RAG2 (Known Mutation)	252704
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Three-gene Profile (IL2RG, ADA, IL7R) (Full Gene Sequencing)	252509
Severe Combined Immunodeficiency (SCID Including Omenn Syndrome): Two-gene Profile (RAG1, RAG2) (Full Gene Sequencing)	252499
Severe Combined Immunodeficiency (SCID): ADA (Full Gene Sequencing)	252475
Severe Combined Immunodeficiency (SCID): ADA (Known Mutation)	252707
Severe Combined Immunodeficiency (SCID): CD3D (Full Gene Sequencing)	252482
Severe Combined Immunodeficiency (SCID): CD3D (Known Mutation)	252713
Severe Combined Immunodeficiency (SCID): CD3E (Full Gene Sequencing)	252485
Severe Combined Immunodeficiency (SCID): CD3E (Known Mutation)	252716
Severe Combined Immunodeficiency (SCID): IL2RG for XSCID (Full Gene Sequencing)	252463
Severe Combined Immunodeficiency (SCID): IL2RG for XSCID (Known Mutation)	252694
Severe Combined Immunodeficiency (SCID): IL7R (Full Gene Sequencing)	252479
Severe Combined Immunodeficiency (SCID): IL7R (Known Mutation)	252710
Severe Combined Immunodeficiency (SCID): JAK3 (Full Gene Sequencing)	252466
Severe Combined Immunodeficiency (SCID): JAK3 (Known Mutation)	252697
Severe Combined Immunodeficiency (SCID): Three-gene Profile (IL7R, CD3D, CD3E) (Full Gene Sequencing)	252506
Severe Combined Immunodeficiency (SCID): Two-gene Profile (IL2RG, JAK3) (Full Gene Sequencing)	252496
Severe Combined Immunodeficiency (SCID): ZAP70 (Full Gene Sequencing)	252489

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Severe Combined Immunodeficiency (SCID): ZAP70 (Known Mutation)	252720
Sex Determination (SRY), DNA Analysis*	510222
SHOX, DHPLC (Endocrine Sciences)	500110
Sickle Cell Anemia Mutation Analysis, Fetal*	451391
SNP Microarray (Direct)–Prenatal (Reveal®)	510200
SNP Microarray – Oncology (Reveal®)	510146
SNP Microarray – Pediatric (Reveal®)	510002
SNP Microarray– Prenatal (Reveal®)*	510100
SNP Microarray – Products of Conception (POC)/Tissue (Reveal®)	510110
Spinal Muscular Atrophy (SMA)	450010
SYT Oncology FISH	510384
Tay-Sachs Disease, Biochemical, Leukocytes	511246
Tay-Sachs Disease, Biochemical	510412
Tay-Sachs Disease, DNA Analysis*	510404
T-Cell Receptor β-Chain Gene Rearrangements	480985
T-Cell Receptor γ-Chain Gene Rearrangements	480708
T-Cell Receptor Gene Rearrangements Profile, γ and β (481080 [combines 480985 and	80708]
α-Thalassemia, DNA Analysis*	511172
β-Thalassemia: HBB (Full Gene Sequencing)	252823
β-Thalassemia: HBB (Known Mutation)	252827
β-Thalassemia: HBB Prenatal Test (Full Gene Sequencing)	252867
β-Thalassemia: HBB Prenatal Test (Known Mutation)	252870
Thiopurine Methyltransferase (TPMT), Enzyme Activity Erythrocytes	510750
Thrombotic Risk Profile, DNA Analysis	512103
Thymidine and Deoxyuridine Analytes (Plasma)	620173
UGT1A1 Irinotecan Toxicity	511200
Uniparental Disomy (UPD), Proband, DNA Analysis	470074
Usher Syndrome Type IF*	450060
Usher Syndrome Type III*	450050
VistaSeq® Hereditary Cancer Panel	481220
VistaSeq® Brain/CNS/PNS Cancer Panel	481386
VistaSeq® Breast and GYN Cancer Panel	481341
VistaSeq® Breast Cancer Panel	481319
VistaSeq® Colorectal Cancer Panel	481363
VistaSeq® Endocrine Cancer Panel	481374
VistaSeq® GYN Cancer Panel	481330
VistaSeq®SM Hereditary Cancer Panel Without *BRCA*	481240
VistaSeq® High Risk Colorectal Cancer Panel	481352
VistaSeq® High/Moderate Risk Breast Cancer Panel	481452
VistaSeq® Pancreatic Cancer Panel	481385
VistaSeq® Renal Cell Cancer Panel	481407
von Hippel-Lindau Disease (VHL): VHL (OPT) (Full Gene Sequencing)	252559
von Hippel-Lindau Disease (VHL): VHL (OPT) (Known Mutation)	252562
Walker-Warburg Syndrome*	511480
Whole Exome Sequencing - DUO (Proband)	620023
Whole Exome Sequencing - Proband Only	620024
Whole Exome Sequencing - TRIO (Proband)	620022
Whole Exome Sequencing Comparator - Additional FM	620194
Whole Exome Sequencing Comparator - Father	620197
Whole Exome Sequencing Comparator - Mother	620192
Wiskott-Aldrich Syndrome (WAS): WAS (Full Gene Sequencing)	252459
Wiskott-Aldrich Syndrome (WAS): WAS (Known Mutation)	252690
X-linked Agammaglobulinemia (XLA): BTK (Full Gene Sequencing)	252453
X-linked Agammaglobulinemia (XLA): BTK (Known Mutation)	252683
X-linked Lymphoproliferative Disease (XLP): SH2D1A (Full Gene Sequencing)	252535
X-linked Lymphoproliferative Disease (XLP): SH2D1A (Known Mutation)	252740

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