GENETICS
TEST MENU

LabCorp
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Genetics Test Menu

Test Name	Test N°	Use
General Assays (Includes tests for family history concern, developmental delay, infertility)		
Amino Acid Profile, Quantitative, CSF	700180	Aids in the diagnosis of some inborn errors of amino acid metabolism such as glycine encephalopathy (nonketotic hyperglycinemia). **Note:** Diagnosis of glycine encephalopathy requires the calculation of a CSF:plasma glycine ratio.
Amino Acid Profile, Quantitative, Plasma	700068	Diagnosis and monitoring of patients with inborn errors of amino acid metabolism and urea cycle defects.
Amino Acid Profile, Quantitative, Urine	700140	Diagnosis and monitoring of patients with inborn errors of amino acid metabolism and urea cycle defects.
Angelman and Prader-Willi Syndromes, DNA Analysis*	511210	All major causes of the Angelman and Prader-Willi syndromes are detected by this methodology. This study is not offered for chorionic villus sampling.
Arylsulfatase A Deficiency, Leukocytes	402396	Aids in the diagnosis of patients with metachromatic leukodystrophy (MLD).
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.
Congenital Adrenal Hyperplasia (CAH) 21-Hydroxylase (CYP21) Mutation	500768	Identifies most common mutations that cause congenital adrenal hyperplasia.
Congenital Sucrase-Isomaltase Deficiency (CSID)	511570	Detects sucrase and isomaltase enzyme deficiency which causes CSID.
Enzyme Biotinidase Deficiency, Serum	402362	Diagnosis of biotinidase deficiency. This test is appropriate for the confirmation of newborn screen-positive biotinidase deficiency results.
Factor II (Prothrombin), DNA Analysis	511162	Mutation detection in factor II gene causing increased risk of thrombosis.
Factor V_{Leiden} Mutation Analysis	511154	Detection of Leiden mutation in factor V gene; the Leiden mutation causes 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 patients who are heterozygous for Factor V_{Leiden}. Factor V R2 further increases risk for venous thrombosis in these patients.
Factor V R2 DNA Analysis	503940	Detection of the Factor V R2 polymorphism, which further increases risk for venous thrombosis in individuals who have one copy of the Factor V_{Leiden} mutation.
α-Galactosidase Deficiency, Leukocytes	402388	Diagnose patients with Fabry disease.
β-Galactosidase Deficiency, Leukocytes	402370	Aids in the diagnosis of patients with isolated β-galactosidase deficiency, Morquio disease type B (MPS IVb), and combined β-galactosidase/neuraminidase deficiency (galactosialidosis).
Hereditary Hemochromatosis, DNA Analysis	511345	Follow-up evaluation in individuals with elevated saturated transferrin; identification of carrier or affected individuals for three mutations associated with hereditary hemochromatosis.
Infertility—Male, Y Deletion Analysis (DAZ)	512053	Determine the genetic basis for oligospermia or azoöspermia. Male infertility may also be associated with Klinefelter syndrome or cystic fibrosis mutations, primarily the 5T allele.
Methylenetetrahydrofolate Reductase (MTHFR) Thermolabile Variant, DNA Analysis	511238	Evaluation appropriate for patients with hyperhomocysteinemia and/or venous thrombosis.
Organic Acid Analysis, Urine	716720	Useful in the diagnosis of inborn errors of organic acid metabolism, amino acid metabolism, fatty acid oxidation disorders, urea cycle disorders, and defects of the mitochondrial respiratory chain.

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Sex Determination (SRY), DNA Analysis*	510222	Resolution of unexplained sex reversal or infertility; can help rule out XY cells in individuals with Turner syndrome.
SHOX-DHPLC	500110	Identifies mutations causing short stature related to SHOX deficiency. SHOX deficiency is an indication for somatotropin (Humatrope®).
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), DNA Analysis*	470054	Establishes the chromosome parent of origin to rule out syndromes that result from single-parent inheritance of a specific chromosome pair.
Uniparental Disomy of Chromosome 14 (UPD 14)*	470060	Methylation-specific PCR is used to amplify divergent lengths of the methylated and unmethylated MEG3 DMR region on chromosome 14q32 in a single reaction and accurately identified normal, maternal UPD14, and paternal UPD14. This test is not offered prenatally.
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.

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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.
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.
Carrier and Diagnostic Testing (Includes testing related to ethnicity and prenatal diagnosis)		
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.
α_1-Antitrypsin Deficiency, DNA Analysis*	511881	DNA-based determination of common alleles for hereditary α_1-antitrypsin deficiency, which is associated with chronic obstructive pulmonary disease (COPD) and childhood-onset liver disease. This analysis can be used for prenatal diagnosis and can differentiate homozygotes and heterozygotes when the null allele is present.
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.
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.
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).
Bloom Syndrome, DNA Analysis*	512145	Identification of carrier and affected individuals by testing for the 2281del6ins7 mutation associated with Bloom syndrome in the Ashkenazi Jewish population.
Canavan Disease, DNA Analysis*	511147	Identification of carrier and affected individuals by testing for four point mutations associated with Canavan disease in the Ashkenazi Jewish population.
Chromosome Analysis, High Resolution and Fragile X Syndrome	511058	Accurate identification of the genetic cause of fragile X syndrome or other chromosomally caused mental retardation syndromes. Recommended when there is no family history of fragile X syndrome.
Cystic Fibrosis (CF) Profile, 97 Mutations, CF*plus*®	450020	Determine affected or carrier status for 97 CF gene mutations. This assay may be used for individuals whose family history or ethnicity requires testing for less common mutations. Also available for routine screening of pregnant couples. Discriminates between the ΔF508 mutation and the following polymorphisms: F508C, I506V, and I507V.

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Cystic Fibrosis (CF) Profile, 97 Mutations, CF*plus*®, Fetal Analysis*	480819	An expanded mutation profile of 97 mutations for cystic fibrosis for prenatal testing, diagnostic testing, and for testing in those individuals whose family history or ethnicity requires testing for less common mutations.
Cystic Fibrosis (CF) Profile, DNA Analysis	480533	Determine affected or carrier status for the 32 most common CF mutations. Routine screening for pregnant couples.
Cystic Fibrosis (CF) Profile, DNA Analysis and 5T Allele Genotyping	480555	Useful in work-up for congenital absence of the vas deferens and/or chronic pancreatitis.
Cystic Fibrosis (CF) Profile, 32 Mutations, Fetal Analysis*	480541	Used to detect fetal status for the 32 most common CF mutations.
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.
Familial Dysautonomia, DNA Analysis*	511352	Identification of carrier and affected individuals by testing for two mutations 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 by testing for two mutations associated with Fanconi anemia C in the Ashkenazi Jewish population.
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, Reflex to Southern Blot	510234	Identifies carriers and/or affected individuals with fragile X. Recommended without cytogenetics when an affected individual has already been identified within the family.
Gaucher Disease, DNA Analysis*	511048	Identifies carriers and affected individuals using eight mutations associated with Gaucher disease in the Ashkenazi Jewish population. DNA testing may be used to confirm affected status.
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 mutations associated with GSD1a in the Ashkenazi Jewish population.
Hemoglobin, Sickle Cell, Prenatal, DNA*	451391	DNA analysis to detect mutations known to cause sickle cell anemia.
Joubert Syndrome Type II, DNA Analysis*	511490	Detect the presence of the R12L mutation (also called R73L) in the *TMEM216* gene.
informaSeq℠ Prenatal Test	550476	Prenatal aneuploidy assay used for screening chromosomes 13, 18, and 21. Test is validated for singleton and twin pregnancies with gestational ages of at least 10 weeks.
informaSeq℠ Prenatal Test With X, Y Analysis	550716	Prenatal aneuploidy assay used for screening chromosomes 13, 18, 21, X, and Y. The test is validated for singleton pregnancies with gestational ages of at least 10 weeks. This test is not intended to be used for fetal diagnostic purposes or as a stand-alone diagnostic test without confirmation by another medically established diagnostic product or procedure. Test results that suggest high risk for fetal trisomy should prompt consideration for genetic counseling. Results should be considered in the context of other clinical criteria.

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informaSeq℠ Prenatal Test With Y Analysis	550757	Prenatal aneuploidy assay used for screening chromosomes 13, 18, and 21. The X chromosome is not assessed by this assay. The test is validated for singleton and twin pregnancies with gestational ages of at least 10 weeks. This test is not intended to be used for fetal diagnostic purposes or as a stand-alone diagnostic test without confirmation by another medically established diagnostic product or procedure. Test results that suggest high risk for fetal trisomy should prompt consideration for genetic counseling. Results should be considered in the context of other clinical criteria.
Maple Syrup Urine Disease (MSUD) 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 mutations 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.
Mucolipidosis Type IV Mutation Detection*	511386	Carrier testing for two mutations associated with mucolipidosis type IV in the Ashkenazi Jewish population. DNA testing may be used to confirm affected status.
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.
Niemann-Pick Disease, DNA Analysis*	511329	Identifies carrier and affected individuals using four mutations associated with Niemann-Pick disease, types A and B, in the Ashkenazi Jewish population.
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, Serum	510412	Determines Tay-Sachs carrier and affected status in all ethnic groups. (This serum assay should **not** be performed on women who are pregnant or who are taking oral contraceptives.)
Tay-Sachs Disease, DNA Analysis*	510404	Identifies Tay-Sachs disease carriers and affected individuals in specific ethnic groups. The test identifies three mutations associated with the Ashkenazi Jewish population, one mutation associated with the French Canadian population, one associated with non-Jewish Caucasians, and two pseudodeficiency mutations.
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.
Walker-Warburg Syndrome*	511480	Detection of the c.1167insA mutation on the *FKTN* gene, which accounts for approximately 99% of Walker-Warburg carriers in the Ashkenazi Jewish population.

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Gene Sequencing (Full Sequencing and Known Mutation)		
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSC2* (Full Gene Sequencing)	252380	Confirm a clinical diagnosis of ARVD/C; identify presymptomatic family members, guiding prophylactic measures.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSC2* (Known Mutation)	252630	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.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSG2* (Full Gene Sequencing)	252383	Confirm a clinical diagnosis of ARVD/C; identify presymptomatic family members, guiding prophylactic measures.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSG2* (Known Mutation)	252633	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.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSP* (Full Gene Sequencing)	252376	Confirm a clinical diagnosis of ARVD/C; identify presymptomatic family members, guiding prophylactic measures.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSP* (Known Mutation)	252626	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.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): Five-gene Profile (*PKP2, DSP, DSC2, DSG2, TMEM43*) (Full Gene Sequencing)	252370	Confirm a clinical diagnosis of ARVD/C; identify presymptomatic family members, guiding prophylactic measures.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *PKP2* (Full Gene Sequencing)	252373	
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *PKP2* (Known Mutation)	252623	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.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *TMEM43* (Full Gene Sequencing)	252386	Confirm a clinical diagnosis of ARVD/C; identify presymptomatic family members, guiding prophylactic measures.
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *TMEM43* (Known Mutation)	252637	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.
Atrial Septal Defect (ASD) With Atrioventricular Block (AVB): *NKX2.5* (Full Gene Sequencing)	252405	Identify NKX2.5 mutations as the cause of familial ASD, indicating high risk of AVB and allowing early diagnosis in family members.
Atrial Septal Defect (ASD) With Atrioventricular Block (AVB): *NKX2.5* (Known Mutation)	252651	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.
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.
Autoimmune Polyglandular Syndrome Type 1 (APS1/APECED): *AIRE* (Known Mutation)	252737	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.
β-Thalassemia: *HBB* (Full Gene Sequencing)	252823	Confirm a clinical diagnosis of β-thalassemia; detect carriers; help to establish a prognosis.

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β-Thalassemia: *HBB* (Known Mutation)	252827	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.
β-Thalassemia: *HBB* Prenatal Test (Full 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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.
Bardet-Biedl Syndrome (BBS): *BBS1* (Full Gene Sequencing)	252549	Confirm a clinical diagnosis of BBS; detect carriers; allow early diagnosis in family members.
Bardet-Biedl Syndrome (BBS): *BBS1* (Known Mutation)	252753	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.
Bardet-Biedl Syndrome (BBS): *BBS2* (Full Gene Sequencing)	252553	Confirm a clinical diagnosis of BBS; detect carriers; allow early diagnosis in family members.
Bardet-Biedl Syndrome (BBS): *BBS2* (Known Mutation)	252756	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.
Bardet-Biedl Syndrome (BBS): Two-gene Profile (*BBS1, BBS2*) (Full Gene Sequencing)	252556	Confirm a clinical diagnosis of BBS; detect carriers; allow early diagnosis in family members.
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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.
Congenital Bilateral Absence of the Vas Deferens (CBAVD): *CFTR* (Full Gene Sequencing)	252766	Confirm a clinical diagnosis of CBAVD; predict risk of CF in blood relatives.
Congenital Bilateral Absence of the Vas Deferens (CBAVD): *CFTR* (Known Mutation)	252770	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.
Cystic Fibrosis (CF): *CFTR* (Full Gene Sequencing)	252763	Confirm a clinical diagnosis of CF; predict risk of CF in blood relatives.
Cystic Fibrosis (CF): *CFTR* (Known Mutation)	252760	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.
Dilated Cardiomyopathy (DCM): *ACTC* (Full Gene Sequencing)	252364	Confirm a clinical diagnosis of DCM; identify presymptomatic family members, guiding prophylactic measures.
Dilated Cardiomyopathy (DCM): *ACTC* (Known Mutation)	252615	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.
Dilated Cardiomyopathy (DCM): *LMNA* (Full Gene Sequencing)	252367	Confirm a clinical diagnosis of DCM; identify presymptomatic family members, guiding prophylactic measures.
Dilated Cardiomyopathy (DCM): *LMNA* (Known Mutation)	252620	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.

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Dilated Cardiomyopathy (DCM): *MYBPC3* (Full Gene Sequencing)	252357	Confirm a clinical diagnosis of DCM; identify presymptomatic family members, guiding prophylactic measures.
Dilated Cardiomyopathy (DCM): *MYBPC3* (Known Mutation)	252609	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.
Dilated Cardiomyopathy (DCM): *MYH7* (Full Gene Sequencing)	252360	Confirm a clinical diagnosis of DCM; identify presymptomatic family members, guiding prophylactic measures.
Dilated Cardiomyopathy (DCM): *MYH7* (Known Mutation)	252612	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.
Dilated Cardiomyopathy (DCM): Six-gene Profile (*TNNT2, TPM1, MYH7, MYBPC3, ACTC, LMNA*) (Full Gene Sequencing)	252343	Confirm a clinical diagnosis of DCM; identify presymptomatic family members, guiding prophylactic measures.
Dilated Cardiomyopathy (DCM): *TNNI3* (Full Gene Sequencing)	252350	
Dilated Cardiomyopathy (DCM): *TNNI3* (Known Mutation)	252603	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.
Dilated Cardiomyopathy (DCM): *TNNT2* (Full Gene Sequencing)	252347	Confirm a clinical diagnosis of DCM; identify presymptomatic family members, guiding prophylactic measures.
Dilated Cardiomyopathy (DCM): *TNNT2* (Known Mutation)	252599	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.
Dilated Cardiomyopathy (DCM): *TPM1* (Full Gene Sequencing)	252354	Confirm a clinical diagnosis of DCM; identify presymptomatic family members, guiding prophylactic measures.
Dilated Cardiomyopathy (DCM): *TPM1* (Known Mutation)	252606	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.
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *APOB* (Single Exon Sequencing)	252392	Confirm a clinical diagnosis of familial hypercholesterolemia; allow early diagnosis in family members, guiding use of pharmacological treatment in children.
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *APOB* (Known Mutation)	252644	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.
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *LDLR* (Full Gene Sequencing)	252388	Confirm a clinical diagnosis of familial hypercholesterolemia; allow early diagnosis in family members, guiding use of pharmacological treatment in children.
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *LDLR* (Known Mutation)	252640	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.
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *PCSK9* (Full Gene Sequencing)	252873	Confirm a clinical diagnosis of familial hypercholesterolemia and allow early diagnosis in family members, thus promoting early intervention, which may prevent or repair atherosclerotic damage and lower the risk of CHD.
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *PCSK9* (Known Mutation)	252877	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.

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Test Name	Test N°	Use
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: Three-gene Profile (*LDLR, APOB, PCSK9*) (*LDLR/PCSK9*-Full Gene Sequencing, *APOB*-Single Exon Sequencing)	252880	Confirm a clinical diagnosis of familial hypercholesterolemia and allow early diagnosis in family members, thus promoting early intervention, which may prevent or repair atherosclerotic damage and lower the risk of CHD.
Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: Two-gene Profile (*LDLR, APOB*) (*LDLR*-Full Gene Sequencing, *APOB*-Single Exon Sequencing)	252396	Confirm a clinical diagnosis of familial hypercholesterolemia; allow early diagnosis in family members, guiding use of pharmacological treatment in children.
Familial Mediterranean Fever: *MEFV* (Full Gene Sequencing)	252797	Can confirm a clinical diagnosis of FMF, detect carriers, and allow early diagnosis in family members, guiding prophylactic measures.
Familial Mediterranean Fever: *MEFV* (Known Mutation)	252800	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.
Galactosemia: *GALT* (Full Gene Sequencing)	252816	Confirm a clinical diagnosis of galactosemia; detect carriers; help to establish a prognosis.
Galactosemia: *GALT* (Known Mutation)	252820	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.
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	
GeneSeq®: Cardio Familial Arrhythmia Profile	451412	
GeneSeq®: Cardio Familial Cardiomyopathy Profile	451422	
GeneSeq®: Cardio Familial Congenital Heart Disease Profile	451402	
GeneSeq®: Cardio Noonan Syndrome and Related Conditions Profile	451441	Confirm a clinical diagnosis of Noonan syndrome and identify presymptomatic family members, guiding prophylatic measures.
GJB2 Sequencing, Full Gene Sequencing*	511405	Detects mutations in the coding region and noncoding first exon of the connexin 26 (*GJB2*) gene associated with nonsyndromic sensorineural hearing loss (NSHL)
GJB2 Sequencing, Family-targeted (Single Exon Sequencing–Known Mutation)*	511414	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.
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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.

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Test Name	Test N°	Use
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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.
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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation 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.
Hyper-IgM Syndrome (HIGM): Three-gene Profile (*AICDA, UNG, CD40*) (Full Gene Sequencing)	252442	Confirm a clinical diagnosis of HIGM; detect carriers; allow early diagnosis in family members, guiding prophylactic measures.
Hyper-IgM Syndrome (HIGM): Two-gene Profile (*AICDA, UNG*) (Full Gene Sequencing)	252439	
Hypertrophic Cardiomyopathy (HCM): *ACTC* (Full Gene Sequencing)	252332	
Hypertrophic Cardiomyopathy (HCM): *ACTC* (Known Mutation)	252589	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.
Hypertrophic Cardiomyopathy (HCM): Eight-gene Profile (*TNNT2, TNNI3, TPM1, MYBPC3, MYH7, MYL2, MYL3, ACTC*) (Full Gene Sequencing)	252300	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): Five-gene Minor Profile (*TNNI3, TPM1, MYL2, MYL3, ACTC*) (Full Gene Sequencing)	252297	
Hypertrophic Cardiomyopathy (HCM): *LAMP2* (Full Gene Sequencing)	252340	
Hypertrophic Cardiomyopathy (HCM): *LAMP2* (Known Mutation)	252596	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.
Hypertrophic Cardiomyopathy (HCM): Metabolic HCM Profile (*PRKAG2, LAMP2*) (Full Gene Sequencing)	252303	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): *MYBPC3* (Full Gene Sequencing)	252321	

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Test Name	Test N°	Use
Hypertrophic Cardiomyopathy (HCM): *MYBPC3* (Known Mutation)	252575	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.
Hypertrophic Cardiomyopathy (HCM): *MYH7* (Full Gene Sequencing)	252324	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): *MYH7* (Known Mutation)	252579	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.
Hypertrophic Cardiomyopathy (HCM): *MYL2* (Full Gene Sequencing)	252327	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): *MYL2* (Known Mutation)	252582	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.
Hypertrophic Cardiomyopathy (HCM): *MYL3* (Full Gene Sequencing)	252329	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): *MYL3* (Known Mutation)	252586	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.
Hypertrophic Cardiomyopathy (HCM): *PRKAG2* (Full Gene Sequencing)	252335	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): *PRKAG2* (Known Mutation)	252592	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.
Hypertrophic Cardiomyopathy (HCM): Reflex Profile (*TNNT2, MYH7, MYBPC3 TPM1, TNNI3, MYL2, MYL3, ACTC*) (Full Gene Sequencing)	252307	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): Three-gene Major Profile (*TNNT2, MYH7, MYBPC3*) (Full Gene Sequencing)	252293	
Hypertrophic Cardiomyopathy (HCM): *TNNI3* (Full Gene Sequencing)	252314	
Hypertrophic Cardiomyopathy (HCM): *TNNI3* (Known Mutation)	252568	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.
Hypertrophic Cardiomyopathy (HCM): *TNNT2* (Full Gene Sequencing)	252310	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): *TNNT2* (Known Mutation)	252565	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.
Hypertrophic Cardiomyopathy (HCM): *TPM1* (Full Gene Sequencing)	252317	Confirm a clinical diagnosis of HCM; identify presymptomatic family members, guiding prophylactic measures.
Hypertrophic Cardiomyopathy (HCM): *TPM1* (Known Mutation)	252572	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.
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.

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Test Name	Test N°	Use
Hypohidrotic Ectodermal Dysplasia With Immune Deficiency (HED-ID): *IKBKG* (NEMO) (Known Mutation)	252744	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.
Interferon-γ Receptor Deficiency: *IFNGR1* (Full Gene Sequencing)	252519	Confirm a clinical diagnosis of IFNGR1; guide therapy; detect carriers; allow early diagnosis in family members.
Interferon-γ Receptor Deficiency: *IFNGR1* (Known Mutation)	252727	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.
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 mutation is known and can be documented by the ordering physician. If the mutation 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.
Loeys-Dietz Syndrome (LDS): *TGFBR1* (Full Gene Sequencing)	252413	Confirm a clinical diagnosis of LDS; identify presymptomatic family members, guiding prophylactic measures.
Loeys-Dietz Syndrome (LDS): *TGFBR1* (Known Mutation)	252657	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.
Loeys-Dietz Syndrome (LDS): *TGFBR2* (Full Gene Sequencing)	252416	Confirm a clinical diagnosis of LDS; identify presymptomatic family members, guiding prophylactic measures.
Loeys-Dietz Syndrome (LDS): *TGFBR2* (Known Mutation)	252660	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.
Loeys-Dietz Syndrome (LDS): Two-gene Profile (*TGFBR1, TGFBR2*) (Full Gene Sequencing)	252419	Confirm a clinical diagnosis of LDS; identify presymptomatic family members, guiding prophylactic measures.
Marfan Syndrome (MFS): *FBN1* (Full Gene Sequencing)	252406	Confirm a clinical diagnosis of MFS; identify presymptomatic family members, guiding prophylactic measures.
Marfan Syndrome (MFS): *FBN1* (Known Mutation)	252654	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.
Marfan Syndrome to Loeys-Dietz Syndrome Reflex Profile (MFS LDS): *FBN1 TGFBR1, TGFBR2* (Full Gene Sequencing)	252409	Confirm a clinical diagnosis of MFS or LDS; identify presymptomatic family members, guiding prophylactic measures.
Pulmonic Stenosis: *PTPN11* (Full Gene Sequencing)	252399	Confirm a clinical or biochemical diagnosis of pulmonic stenosis; allow early diagnosis in family members.
Pulmonic Stenosis: *PTPN11* (Known Mutation)	252647	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.
SCN1A Sequencing, Full Gene	511236	Detects severe myoclonic epilepsy of infancy (SMEI), known as Dravet syndrome.
SCN1A Family-targeted Sequencing	511274	

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Test Name	Test N°	Use
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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.
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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.

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Test Name	Test N°	Use
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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation 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.
Severe Combined Immunodeficiency (SCID): *IL7R* (Known Mutation)	252710	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.
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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.
Spinal Muscular Atrophy (SMA) Carrier Testing	450010	Determine carrier status and prenatal diagnosis for spinal muscular atrophy
Thoracic Aortic Aneurysms and Dissections (TAAD): Three-gene Profile (*FBN1, TGFBR1, TGFBR2*) (Full Gene Sequencing)	252422	Identify MFS or LDS as the genetic cause of syndromic TAAD; allow identification of presymptomatic family members, guiding prophylactic measures.
Transthyretin Amyloidosis: *TTR* (Full Gene Sequencing)	252810	Confirm a clinical diagnosis of transthyretin amyloidosis in a patient and predict risk of transthyretin amyloidosis in blood relatives.
Transthyretin Amyloidosis: *TTR* (Known Mutation)	252813	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.
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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.

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Test Name	Test N°	Use
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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation 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 mutation is known and can be documented by the ordering physician. If the mutation cannot be documented, please order full gene sequencing.
Cytogenetic Analysis (Prenatal and Postnatal Diagnosis)		
Chromosome Analysis and AFP, Amniotic Fluid*	510032	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)*	510255	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.
Chromosome Analysis, Amniotic Fluid*	052040	Determine fetal karyotype. The test allows prenatal detection of a chromosomal abnormality in pregnant women at increased risk for aneuploidy, translocation, etc. While chromosome analysis is being performed, additional biochemical or molecular analysis can be done.
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. The microarray test allows prenatal detection of of clinically relevant alterations below the resolution of chromosome analysis. The genotyping portion of the SNP Microarray will also screen for UPD for all chromosomes and estimate identity by descent.
Chromosome Analysis, Blood (Constitutional)	511035	Provides karyotype on individuals with congenital malformations, mental retardation, growth retardation, infertility, cryptorchidism, hypogonadism, amenorrhea (primary), abnormal/ambiguous genitalia, recurrent miscarriage, Turner syndrome, Klinefelter syndrome, Down syndrome, or other suspected chromosomal disorders.
Chromosome Analysis, Chorionic Villi Biopsy*	510988	Determine fetal karyotype using chorionic villi. Allows prenatal detection of a chromosomal abnormality in at-risk pregnancies.
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. The microarray test allows prenatal detection of of clinically relevant alterations below the resolution of chromosome analysis. The genotyping portion of the SNP Microarray will also screen for UPD for all chromosomes and estimate identity by descent.
Chromosome Analysis, High Resolution	052215	Detects small chromosome abnormalities not detectable using routine methods; precise identification of abnormal chromosomes previously detected by routine methods.

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Test Name	Test N°	Use
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, 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, High Resolution, 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 2.65 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; used to rule out tetraploidy and rearrangements not detected by microarray, such as balanced translocation and inversions. This test provides detection of uniparental disomy of any chromosome, the percentage and location of homozygosity, including the degree of identity by descent. Abbreviated chromosome analysis detects balanced rearrangements.
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.
Inheritest® Carrier Screen	451381	This analysis provides carrier testing by analyzing 434 mutations associated with more than 90 diseases. Mutations are selected for relatively high frequency in the general population or in specific ethnic populations; therefore, the clinical sensitivity and specificity varies for each disease and for each ethnic group.
Inheritest® Select Carrier Screen	451394	This analysis provides carrier testing by analyzing 147 mutations associated with more than 18 diseases. Mutations are selected for relatively high frequency in the general population or in specific ethnic populations; therefore, the clinical sensitivity and specificity varies for each disease and for each ethnic group.
Fluorescence in situ Hybridization (FISH), Microdeletion Syndromes*	510770	Confirmation/identification of targeted deletions below the resolution of cytogenetics. (Call the laboratory for a list of available probes.)
Fluorescence in situ Hybridization (FISH), Multiprobe, Subtelomere-specific	510350	Primarily used for detection of cryptic chromosome rearrangements in adults with a vamily history of newborns with idiopathic congenital syndromes.
Fluorescence in situ Hybridization (FISH), Paraffin Block	510825	For specific FISH probe analysis of tissue specimens.
Fluorescence in situ Hybridization (FISH), Prenatal Aneuploid Evaluation, Amniotic Fluid*	510365	Rapid identification of common prenatal aneuploidy (specific for X, Y, 13, 18, and 21), using FDA-approved probes and protocol.
Fluorescence in situ Hybridization (FISH), Prenatal Aneuploid Evaluation, Chorionic Villus Sampling*	510960	Rapid identification of common prenatal aneuploidy (specific for X, Y, 13, 18, and 21).
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.

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Test Name	Test N°	Use
SNP Microarray– Prenatal (Reveal®)*	510100	This test will detect chromosomal imbalance that could be associated with developmental delay/congenital anomalies. Provides detection of uniparental disomy of any chromosome, the percentage and location of homozygosity including the degree of identity by descent.
SNP Microarray – Products of Conception (POC)/Tissue (Reveal®)	510110	This test will detect chromosomal imbalance that may be associated with fetal loss and is ideal for detection of complete and partial moles.
Cancer Genetics		
1P,19Q Oncology Fluorescence in situ Hybridization (FISH)	510360	Confirmation/identification of cancer-related alterations (associated with oligoglioma).
Acute Lymphocytic Leukemia (ALL), FISH	510762	Diagnostic and prognostic test for acute lymphocytic leukemia; detection rate is improved from 50% with a chromosome study to about 90% with fluorescence in situ hybridization (FISH).
Acute Lymphocytic Leukemia/Lymphoma (ALL), FISH	511077	Confirmation/identification of chromosome abnormalities below the resolution of cytogenetics (call for list of available probes). Leukemia/lymphoma monitoring of residual disease, transplants, and indolent clones.
Aggressive B-Cell Lymphoma Profile, FISH	510344	Diagnostic test for aggressive non-Hodgkin's lymphoma. Detects genetic changes associated with rearrangements of *MYC, BCL2,* and *BCL6*.
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 B-cell 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 B-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.
BCL2-IGH Gene Rearrangement	480566	The translocation t(14;18)(q32;q21) is found in 80% to 90% of follicular lymphomas, 30% of large diffuse lymphomas, and 50% of undifferentiated lymphomas.
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.
BCR-ABL1, Transcript Detection for Chronic Myelogenous Leukemia and Acute Lymphocytic Leukemia, Quantitative	480481	Measures *BCR-ABL1* transcript levels of the b2a2/b3a2 (p210) and e1a2 (p190) fusion transcripts. These *BCR-ABL1* fusion transcripts are found in patients with CML and Philadelphia chromosome-positive ALL. This quantitative test is used to monitor the response of patients to imatinib mesylate or other therapies.

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Test Name	Test N°	Use
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.
Bladder Cancer FISH, PhD Read	130090	
BRAF Gene Mutation Assay, Melanoma	480450	Approximately 50% of melanomas have an activating mutation in the BRAF oncogene. The majority of these genetic alterations are the V600E (1799 T>A) mutation. Recent studies have shown that the drug vemurafenib (Zelboraf™) has clinical efficacy in tumors with the mutation. The cobas® 4800 BRAF V600 Mutation test is approved by the FDA for the qualitative detection of the BRAF V600E mutation in DNA extracted from formalin-fixed, paraffin-embedded human melanoma tissue.
BRAF Gene Mutation Detection	480340	Metastatic colorectal cancer patients with BRAF mutation do not have a strong response to anti-EGFR therapies such as cetuximab and panitumumab. This assay detects the *V600E* mutation in the BRAF gene, allowing identification of patients who are likely to benefit from such treatment.
BRCA1 Targeted Analysis (BRCA*ssure*℠)	252235	Once a mutation is identified in an index patient, family members can be tested for the presence of that specific mutation (single-amplicon testing by sequencing or deletion/duplication testing by multiplex ligation-dependent probe amplification [MLPA]).
BRCA2 Targeted Analysis (BRCA*ssure*℠)	252250	
BRCA1/2 Ashkenazi Jewish Profile (BRCA*ssure*℠)	252970	Screens for three founder mutations in *BRCA*1 (c.68_69delAG and c.5266dupC) and *BRCA*2 (c.5946delT) genes in the Ashkenazi Jewish population. These mutations are also known by their previous nomenlcature, namely 187delAG and 5382insC for the *BRCA1* and 6174delT for the *BRCA2* gene.
BRCA1/2 Comprehensive Analysis (BRCA*ssure*℠)	252911	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*℠)	252888	If no mutation or inconclusive results are reported after sequence analysis, testing for deletions or complex alleles in *BRCA1* and/or *BRCA2* may be considered.
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 Fluorescence in situ Hybridization (FISH)	510349	Confirmation/identification of cancer-related alterations (associated with myxoid liposarcomas/round liposarcomas).

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Test Name	Test N°	Use
Chromosome 18q Allelic Loss, Paraffin Block	481101	Identifies tumors with chromosome 18q allelic loss (loss of heterozygosity), which is a prognostic marker in colorectal cancer.
Chromosome 18q Allelic Loss, Frozen Tissue	480459	
Chromosome Analysis, Leukemia/ Lymphoma	510999	Identifies chromosomal abnormalities associated with leukemias or lymphomas.
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, Fluorescence in situ Hybridization (FISH)	510594	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.
Chronic Myelogenous Leukemia (CML) Profile: Chromosome Analysis and *BCR-ABL*, Fluorescence in situ Hybridization (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 Mutation Analysis, and MPL Mutation Analysis	511595	Confirm the diagnosis of CML; establish the chronic phase karyotype for comparison with blast crisis alterations; monitor residual disease
EGFR Oncology Fluorescence in situ Hybridization (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) Gene Mutation Analysis, Non–Small-cell Lung Cancer (cobas®)	489489	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.
Epidermal Growth Factor Receptor (EGFR) Mutation Analysis for Non–Small-cell Lung Cancer	489360	
EWSR1 Oncology Fluorescence in situ Hybridization (FISH)	510379	Confirmation/identification of cancer-related alterations (Ewing sarcoma).
FKHR Oncology Fluorescence in situ Hybridization (FISH)	510371	Confirmation/identification of cancer-related alterations (alveolar rhabomyosarcoma).
Fluorescence in situ Hybridization ALK (FISH), Non–Small-cell Lung Cancer	510950	Confirmation/identification of non–small-cell lung cancer.
Fluorescence in situ Hybridization (FISH), Oncology	510669	Confirmation/identification of cancer-related alterations. (Call the laboratory for a list of available probes.)

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Test Name	Test N°	Use
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.)
Glutathione-S-Transferase (*GST-P1*) and Adenomatous Polyposis Coli (*APC*) Gene Promoter Methylation Assay	489320	Hypermethylation of the promoter regions of the *GSTP1* and *APC* genes occurs at a significantly higher frequency in prostate cancer samples than in benign conditions of the prostate gland. Gene methylation assays may be used as an adjunct to histopathology for patients in whom prostate disease is suspected.
HER-2/CEP17, Fluorescence in situ Hybridization (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.
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): *MLH1* (Known Mutation)	511635	Identify presymptomatic mutation carriers within a family who are at high risk of developing the familial disease.
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	
JAK2 Exon 12 Mutation Analysis	489212	JAK2 exon 12 mutation status is associated with erythrocytosis and atypical bone marrow morphology; mutation analysis may be used to differentiate reactive conditions from malignant erythrocytosis. JAK2 exon 12 mutations appear to be specific to polycythemia vera (PV) or idiopathic erythrocytosis. Patients who test negative for JAK2^{V617F} mutation should be tested for JAK2 exon 12 mutation to classify further into PV.
JAK2^{V617F} Mutation Detection, 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.
JAK2^{V617F} Mutation Analysis, Qualitative, With Reflex to *CALR* Mutation Analysis, JAK2 Exon 12 Mutation Analysis, and MPL Mutation Analysis	489395	This test will ssess for the JAK2^{V617F} (exon 14) mutation first and will reflex to *CALR* mutation analysis, JAK2 exon 12 mutation analysis, and MPL mutation analysis when the JAK2^{V617F} mutation is negative.
JAK2^{V617F} Mutation Analysis, Quantitative	489470	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). TheJAK2^{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. *V617F* is an acquired mutation that alters a highly conserved valine present in the negative regulatory JH2 domain of the JAK2 protein and is predicted to dysregulate kinase activity.
***K-ras* Gene Mutation Detection**	480090	This assay detects K-ras mutations in codon 12 and 13. If a K-ras mutation is detected, then patients with metastatic colorectal carcinoma should not receive anti-EGFR antibody therapy as part of their treatment.
***K-ras* Gene Mutation Detection With Reflex to *BRAF* Gene Mutation Detection**	480360	Negative results of K-ras gene mutation will automatically reflex to BRAF gene mutation detection. Please see K-ras Gene Mutation Detection (489200) and BRAF Gene Mutation Detection (480340).

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Test Name	Test N°	Use
Microsatellite Instability	511311	Identify colorectal tumors with high microsatellite instability associated with a less aggressive clinical course; identify individuals with hereditary nonpolyposis colorectal cancer (HNPCC).
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®).
MLH1 Comprehensive Analysis	511615	Confirm a clinical diagnosis of hereditary nonpolyposis colorectal cancer (HNPCC) and allow early diagnosis in family members, guiding preventive measures.
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	MPL (myeloproliferative leukemia virus oncogene homology) belongs to the hematopoietin superfamily and enables its ligand thrombopoietin to facilitate both global hematopoiesis and megakaryocyte growth and differentiation. MPL 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.
MPN/CML, FISH	511425	Confirmation/identification of chromosome abnormalities in interphase nuclei. Leukemia monitoring of residual disease.
MPN With Hypereosinophilia, FISH	511444	
MSH2 Comprehensive Analysis	511632	Confirm a clinical diagnosis of hereditary nonpolyposis colorectal cancer (HNPCC) and allow early diagnosis in family members, guiding preventive measures.
MSH2 Deletion/Duplication Analysis	511705	
MSH6 Comprehensive Analysis	511636	
MSH6 Deletion/Duplication Analysis	511720	
Multiple Myeloma Profile, Fluorescence in situ Hybridization (FISH)	510830	Diagnostic test for multiple myeloma: detection of 13q deletion improved from 15% to 20% to 38% to 54% in newly diagnosed patients; profile improves overall cytogenetic detection rate from 30% to about 80%; IgH translocations detected in 47% of MGUS patients, 60% to 70% of intermedullary MM, and 80% of patients with PCL.
MYCN Oncology Fluorescence in situ Hybridization (FISH)	510945	Confirmation/identification of cancer-related alterations. (Call the laboratory for a list of available probes.)
Myelodysplastic Syndrome, Fluorescence in situ Hybridization (FISH)	510599	Diagnostic test for myelodysplastic syndrome. Principle 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.

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Test Name	Test N°	Use
Non–Small-cell Lung Cancer (NSCLC) Therapeutic Profile: EGFR Mutation Analysis and ALK FISH Analysis	511550	Non–small-cell lung cancer (NSCLC) is the leading cause of death from cancer in both men and women in the United States. A subgroup of NSCLC patients have shown clinical responsiveness to the EGFR inhibitors gefitinib (Iressa®) and erlotinib (Tarceva®). Responsiveness to these drugs is characteristic of distinct subgroups of patients, including those who have never smoked, patients with adenocarcinoma, and individuals of Asian ethnicity. 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 response to gefitinib and erlotinib and is strongly predictive of prolonged survival in NSCLC patients.
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.
***PMS2* Comprehensive Analysis**	511630	Confirm a clinical diagnosis of hereditary nonpolyposis colorectal cancer (HNPCC) and allow early diagnosis in family members, guiding preventive measures.
***PMS2* Deletion/Duplication Analysis**	511725	
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.
P53 Oncology Fluorescence in situ Hybridization (FISH)	510940	Confirmation/identification of cancer-related alterations. (Call the laboratory for a list of available probes.)
***PIK3CA* Oncogene Mutation Detection**	480880	Somatic mutations in the *PIK3CA* oncogene are frequently found in human cancers. They are common in liver cancer, breast cancer, colorectal cancer, and ovarian cancer. These mutations may indicate prognosis and drug response. This assay detects four *PIK3CA* mutations (*H1047R, E542K, E545D, E545K*) in exon 9 and 20, allowing determination of whether there is a correlation between *PIK3CA* mutation status and drug response.
***PML-RARA* Transcript Detection for Acute Promyelocytic Leukemia, Quantitative**	510840	The translocation t(15;17) (q22;q21) is the prototype arrangement 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 PML gene 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 of APL. In vitro studies have indicated that this assay has an analytical sensitivity that allows for the detection of 10 or more copies of the *PML-RARA* fusion transcript.
RB1 Oncology Fluorescence in situ Hybridization (FISH)	510374	Confirmation/identification of cancer-related alterations. (Call the laboratory for a list of available probes.)
RET Oncology Fluorescence in situ Hybridization (FISH)	510315	Confirmation/identification of cancer-related alterations (for lung cancer).

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Test Name	Test N°	Use
RET Proto-oncogene Mutation Detection	489270	The RET (rearranged during transfection) gene encodes a transmembrane tyrosine kinase that functions as the receptor for growth factors of the glial-derived neurotrophic factor (GDNF) family. Mutations in RET are associated with multiple endocrine neoplasias (types 2A and 2B), medullary thyroid cancers (MTC) and Hirschsprung disease. Medullary thyroid cancer (MTC) accounts for about 10% of all diagnosed thyroid cancers and occurs in virtually all multiple endocrine neoplasia (MEN) patients. The M918T mutation occurs in >80% of MTC. This mutation activates RET dimerization leading to VEGFR and EGFR activation downstream, which triggers angiogenesis. It also increases calcitonin secretory cell (C-cell) proliferation and correlates with poor prognosis. RET mutation testing is recommended preoperatively for all MTC patients.
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 Fluorescence in situ Hybridization (FISH)	510384	Confirmation/identification of cancer-related alterations. (Call the laboratory for a list of available probes.)
T and B Gene Rearrangement, PCR	480860 (combines 480708 and 480716)	See 480708 and 480716.
T-Cell Receptor Gene Rearrangements Profile, γ and β	481080 (combines 480985 and 480708)	See 480985 and 480708.
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		
Clopidogrel *CYP2C19* Genotyping	511710	Detects poor metabolizer *CYP2C19* alleles *2, *3, as well as the ultrametabolizer allele, *17. Other rare alleles are not detected by this assay.
Cytochrome P450 *2C9* Genotyping	511270	Detects the genetic polymorphisms, *CYP2C9* *2 and *3, that are associated with poor metabolism of certain drugs with an increased risk for adverse drug reactions.
Cytochrome P450 *2C19* Genotyping	511320	Detects the genetic polymorphisms, *CYP2C19* *2 through *8, that are associated with poor metabolism of certain drugs with an increased risk for adverse drug reactions. The ultrametabolizer allele *17 is also detected.
Cytochrome P450 *2D6* Genotyping	511675	Detects genetic polymorphisms of the *CYP2D6* gene that are associated with poor, intermediate, and ultrarapid metabolism of certain drugs that could lead to an increased risk for adverse drug reactions.

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Test Name	Test N°	Use
Cytochrome P450 *2D6/2C19* Genotyping	511905	Both the *CYP2D6* and *CYP2C19* genes encode members of the cytochrome P450 superfamily of enzymes known to contribute to the metabolism of many clinically relevant drugs in several therapeutic areas, ie, cardiovascular, gastroenterology, neurology, and psychiatry. An individual's specific genotype can result in distinct drug metabolizing phenotypes. Information about *CYP2D6* and *CYP2C19* genotype may be used as an aid to clinicians in determining therapeutic strategy and treatment doses for therapeutics that are metabolized by the *CYP2D6* and *CYP2C19* gene product.
DPD 5-Fluorouracil Toxicity	511176	The *2A allele (IVS14+1 G>A) in the DPD (dihydropyrimidine dehydrogenase) gene causes DPD deficiency. DPD deficiency is associated with severe 5-FU (5-fluorouracil) toxicity, which has been shown to be fatal in some *2A homozygotes as well as some heterozygotes.
Interleukin 28B (*IL28B*) Polymorphism (rs12979860)	480630	Detects the *IL28B* genotype CC, which is associated with a twofold to threefold greater rate of sustained viral response in hepatitis C virus genotype 1 chronically infected individuals treated with combination pegylated interferon/ribavirin therapy. The CC genotype has also been associated with a threefold increase in the rate of spontaneous clearance of HCV.
Opioid *CYP2D6* Genotyping	511380	Detects genetic polymorphisms of the *CYP2D6* gene that are associated with poor, intermediate, and ultrarapid metabolism of opioid analgesic medications.
Tamoxifen P450 *2D6* Genotyping	511280	Detects genetic polymorphisms of the *CYP2D6* gene that are associated with poor, intermediate, and ultrarapid metabolism of tamoxifen, a commonly prescribed medication for the treatment of breast cancer.
Thiopurine Methyltransferase (TPMT), Enzyme Activity, Erythrocytes	510750	Determination of TPMT enzyme levels that may be associated with toxicity of anticancer and anti-inflammatory drugs. (This test is offered under license from the Mayo Foundation for Medical Education and Research.)
***UGT1A1* Irinotecan Toxicity**	511200	Irinotecan is used or is 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 (ie, grade 4 neutropenia) is commonly observed in cancer patients receiving irinotecan who carry the *UGT1A1* *28 allele, also called $(TA)_7$. This test provides valuable information to physicians prior to initiating or modifying treatment or supplementing treatment with additional drugs.
Warfarin (P450 *2C9* and *VKORC1*)	511460	Detects the genetic polymorphisms, *CYP2C9* *2 and *3, and the *VKORC1* variant 1173 C>T that is in complete linkage disequilibrium with the -1639 G>A causal variant. *CYP2C9* *2 and *3 and *VKORC1* -1639 G>A are each associated with abnormal metabolism of warfarin.

*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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Comprehensive List of Assays

1P,19Q Oncology Fluorescence in situ Hybridization (FISH) (510360)

Acetylcholinesterase (AChE), Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)* (510354)

Acute Lymphocytic Leukemia (ALL), (FISH) (510762)

Acute Lymphocytic Leukemia/Lymphoma (ALL), FISH (511077)

Aggressive B-Cell Lymphoma Profile, FISH (510344)

α_1-Antitrypsin Deficiency, DNA Analysis* (511881)

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)

α-Galactosidase Deficiency, Leukocytes (402388)

α-Thalassemia, DNA Analysis* (511172)

Amino Acid Profile, Quantitative, Cerebrospinal Fluid (700180)

Amino Acid Profile, Quantitative, Plasma (700068)

Amino Acid Profile, Quantitative, Random Urine (700140)

Angelman and Prader-Willi Syndromes, DNA Analysis* (511210)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSC2* (Full Gene Sequencing) (252380)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSC2* (Known Mutation) (252630)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSG2* (Full Gene Sequencing) (252383)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSG2* (Known Mutation) (252633)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSP* (Full Gene Sequencing) (252376)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *DSP* (Known Mutation) (252626)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): Five-gene Profile (*PKP2, DSP, DSC2, DSG2, TMEM43*) (Full Gene Sequencing) (252370)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *PKP2* (Full Gene Sequencing) (252373)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *PKP2* (Known Mutation) (252623)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *TMEM43* (Full Gene Sequencing) (252386)

Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C): *TMEM43* (Known Mutation) (252637)

Arylsulfatase A Deficiency, Leukocytes (402396)

Ashkenazi Jewish Carrier Profile (333561)

Ashkenazi Jewish Carrier Profile Plus (332859)

Atrial Septal Defect (ASD) With Atrioventricular Block (AVB): *NKX2.5* (Full Gene Sequencing) (252405)

Atrial Septal Defect (ASD) With Atrioventricular Block (AVB): *NKX2.5* (Known Mutation) (252651)

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)

BCL2-IgH Gene Rearrangement (480566)

BCR-ABL1 Kinase Domain Mutation Analysis (480510)

BCR-ABL1, Transcript Detection for Chronic Myelogenous Leukemia and Acute Lymphocytic Leukemia, Quantitative (480481)

β-Galactosidase Deficiency, Leukocytes (402370)

β-Thalassemia: *HBB* (Full Gene Sequencing) (252823)

β-Thalassemia: *HBB* (Known Mutation) (252827)

β-Thalassemia: *HBB* Prenatal Test (Full Sequencing) (252867)

β-Thalassemia: *HBB* Prenatal Test (Known Mutation) (252870)

Bardet-Biedl Syndrome (BBS): *BBS1* (Full Gene Sequencing) (252549)

Bardet-Biedl Syndrome (BBS): *BBS1* (Known Mutation) (252753)

Bardet-Biedl Syndrome (BBS): *BBS2* (Full Gene Sequencing) (252553)

Bardet-Biedl Syndrome (BBS): *BBS2* (Known Mutation) (252756)

Bardet-Biedl Syndrome (BBS): Two-gene Profile (*BBS1, BBS2*) (Full Gene Sequencing) (252556)

Bladder Cancer FISH, Pathologist Review (130080)

Bladder Cancer FISH, PhD Read (130090)

Bloom Syndrome, DNA Analysis* (512145)

BRAF Gene Mutation Assay, Melanoma (480450)

BRAF Gene Mutation Detection (480340)

BRCA1 Targeted Analysis (BRCA*ssure*SM) (252235)

BRCA2 Targeted Analysis (BRCA*ssure*SM) (252250)

BRCA1/2 Ashkenazi Jewish Profile (BRCA*ssure*SM) (252970)

BRCA1/2 Comprehensive Analysis (BRCA*ssure*SM) (252911)

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BRCA1/2 Deletion/Duplication Analysis (BRCA*ssure*SM) (252888)

Calreticulin (*CALR*) Mutation Analysis (489450)

Canavan Disease, DNA Analysis* (511147)

Carnitine, Total and Free (706500)

CEBPA Mutation Analysis (489170)

CHOP Oncology Fluorescence in situ Hybridization (FISH) (510349)

Chromosome 18q Allelic Loss, Paraffin Block (48110)

Chromosome 18q Allelic Loss, Frozen Tissue (480459)

Chromosome Analysis and AFP, Amniotic Fluid* (510032)

Chromosome Analysis, AFP, AChE, Amniotic Fluid With Reflex to Fetal Hemoglobin (Hb F)* (510255)

Chromosome Analysis, Amniotic Fluid* (052040)

Chromosome Analysis, Amniotic Fluid With Reflex to SNP Microarray (Reveal®)* (052104)

Chromosome Analysis, Blood (Constitutional) (511035)

Chromosome Analysis, Chorionic Villi Biopsy* (510988)

Chromosome Analysis, Chorionic Villi Biopsy With Reflex to SNP Microarray (Reveal®)* (511033)

Chromosome Analysis, High Resolution (052215)

Chromosome Analysis, High Resolution and Fragile X Syndrome (511058)

Chromosome Analysis, Instability Syndrome (511045)

Chromosome Analysis, Leukemia/Lymphoma (510999)

Chromosome Analysis, Prenatal Cordocentesis and Fetal Hemoglobin (511025)

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®), 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, Fluorescence in situ Hybridization (FISH) (510594)

Chronic Myelogenous Leukemia (CML) Profile: Chromosome Analysis and *BCR-ABL*, Fluorescence in situ Hybridization (FISH) (150500)

c-KIT Mutation Analysis in Tumors of Hematopoietic Tissue (480940)

Clopidogrel *CYP2C19* Genotyping (511710)

CML FISH Reflex to JAK2^{V617F} Mutation Analysis, Qualitative, With Reflex to *CALR* Mutation Analysis, JAK2 Exon 12 Mutation Analysis, and MPL Mutation Analysis (511595)

Comparative Genomic Hybridization (CGH) Chip Array, Constitutional (510010)

Congenital Adrenal Hyperplasia (CAH) 21-Hydroxylase (CYP21) Mutation (500768)

Congenital Bilateral Absence of the Vas Deferens (CBAVD): *CFTR* (Full Gene Sequencing)(252766)

Congenital Bilateral Absence of the Vas Deferens (CBAVD): *CFTR* (Known Mutation) (252770)

Congenital Sucrase-Isomaltase Deficiency (CSID) (511570)

Cystic Fibrosis (CF): *CFTR* (Full Gene Sequencing) (252763)

Cystic Fibrosis (CF): *CFTR* (Known Mutation) (252760)

Cystic Fibrosis (CF) Profile, 97 Mutations, CFplus® (450020)

Cystic Fibrosis (CF) Profile, 97 Mutations, CFplus®, Fetal Analysis* (480819)

Cystic Fibrosis (CF) Profile, DNA Analysis and 5T Allele Genotyping (480555)

Cystic Fibrosis (CF) Profile, DNA Analysis (480533)

Cystic Fibrosis (CF) Profile, 32 Mutations, Fetal Analysis* (480541)

Cytochrome P450 *2C19* Genotyping (511675)

Cytochrome P450 *2C9* Genotyping (511270)

Cytochrome P450 *2D6* Genotyping(511230)

Cytochrome P450 *2D6/2C19* Genotyping (511905)

Dihydrolipoamide Dehydrogenase (DLD)* (450080)

Dilated Cardiomyopathy (DCM): *ACTC* (Full Gene Sequencing) (252364)

Dilated Cardiomyopathy (DCM): *ACTC* (Known Mutation) (252615)

Dilated Cardiomyopathy (DCM): *LMNA* (Full Gene Sequencing) (252367)

Dilated Cardiomyopathy (DCM): *LMNA* (Known Mutation) (252620)

Dilated Cardiomyopathy (DCM): *MYBPC3* (Full Gene Sequencing) (252357)

Dilated Cardiomyopathy (DCM): *MYBPC3* (Known Mutation) (252609)

Dilated Cardiomyopathy (DCM): *MYH7* (Full Gene Sequencing) (252360)

Dilated Cardiomyopathy (DCM): *MYH7* (Known Mutation) (252612)

Dilated Cardiomyopathy (DCM): Six-gene Profile (*TNNT2, TPM1, MYH7, MYBPC3, ACTC, LMNA*) (Full Gene Sequencing) (252343)

Dilated Cardiomyopathy (DCM): *TNNI3* (Full Gene Sequencing) (252350)

Dilated Cardiomyopathy (DCM): *TNNI3* (Known Mutation) (252603)

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Dilated Cardiomyopathy (DCM): *TNNT2* (Full Gene Sequencing) (252347)

Dilated Cardiomyopathy (DCM): *TNNT2* (Known Mutation) (252599)

Dilated Cardiomyopathy (DCM): *TPM1* (Full Gene Sequencing) (252354)

Dilated Cardiomyopathy (DCM): *TPM1* (Known Mutation) (252606)

DPD 5-Fluorouracil Toxicity (511176)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *APOB* (Single Exon Sequencing) (252392)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *APOB* (Known Mutation) (252644)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *LDLR* (Full Gene Sequencing) (252388)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *LDLR* (Known Mutation) (252640)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *PCSK9* (Full Gene Sequencing) (252873)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: *PCSK9* (Known Mutation) (252877)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: Three-gene Profile (*LDLR, APOB, PCSK9*) (*LDLR/PCSK9*-Full Gene Sequencing, *APOB*-Single Exon Sequencing) (252880)

Early-onset Coronary Heart Disease/Familial Hypercholesterolemia: Two-gene Profile (*LDLR, APOB*) (*LDLR*-Full Gene Sequencing, *APOB*-Single Exon Sequencing)

EGFR Oncology Fluorescence in situ Hybridization (FISH) (510355)

Enzyme Biotinidase Deficiency, Serum (402362)

EPCAM Deletion/Duplication Analysis (511654)

Epidermal Growth Factor Receptor (EGFR) Gene Mutation Analysis, Non–Small-cell Lung Cancer (cobas®) (489489)

Epidermal Growth Factor Receptor (EGFR) Mutation Analysis for Non–Small-cell Lung Cancer (489240)

EWSR1 Oncology Fluorescence in situ Hybridization (FISH) (510379)

Factor II (Prothrombin), DNA Analysis (511162)

Factor V_{Leiden} Mutation Analysis (511154)

Factor V_{Leiden} With Reflex to R2 DNA Analysis (503853)

Factor V R2 DNA Analysis (503940)

Familial Dysautonomia, DNA Analysis* (511352)

Familial Hyperinsulinism (FHI)* (450070)

Familial Mediterranean Fever: *MEFV* (Full Gene Sequencing) (252797)

Familial Mediterranean Fever: *MEFV* (Known Mutation) (252800)

Fanconi Anemia (Type C), DNA Analysis* (511212)

First Trimester Screen With Nuchal Translucency (017500)

FKHR Oncology Fluorescence in situ Hybridization (FISH) (510371)

Fluorescence in situ Hybridization ALK (FISH), Non–Small-cell Lung Cancer (510950)

Fluorescence in situ Hybridization (FISH), Microdeletion Syndromes* (510770)

Fluorescence in situ Hybridization (FISH), Multiprobe, Subtelomere-specific (510350)

Fluorescence in situ Hybridization (FISH), Oncology (510669)

Fluorescence in situ Hybridization (FISH), Paraffin Block (510825)

Fluorescence in situ Hybridization (FISH), Prenatal Aneuploid Evaluation, Amniotic Fluid* (510365)

Fluorescence in situ Hybridization (FISH), Prenatal Aneuploid Evaluation, Chorionic Villus Sampling* (510960)

Fluorescence in situ Hybridization (FISH), TELO-SCAN, Multiprobe, Subtelomere-specific (510552)

Fragile X Syndrome, PCR, Reflex to Southern Blot (510234)

Fragile X Syndrome, DNA Analysis, Prenatal with Southern Blot Analysis* (510300)

Galactosemia: *GALT* (Full Gene Sequencing) (252816)

Galactosemia: *GALT* (Known Mutation) (252820)

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 Noonan Syndrome and Related Conditions Profile (451441)

GJB2 Sequencing, Full Gene Sequencing* (511405)

GJB2 Sequencing, Family-targeted (Single Exon Sequencing–Known Mutation)* (511414)

Glutathione-S-Transferase (*GST-P1*) and Adenomatous Polyposis Coli (*APC*) Gene Promoter Methylation Assay (489320)

Glycogen Storage Disease 1a* (511290)

Hemoglobin S and C, Fetal DNA* (480530)

Hemoglobin, Sickle Cell, Prenatal, DNA* (451391)

HER-2/CEP17, Fluorescence in situ Hybridization (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)

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Hereditary Nonpolyposis Colorectal Cancer (HNPCC): *MSH6* (Known Mutation) (511765)

Hereditary Nonpolyposis Colorectal Cancer (HNPCC): *PMS2* (Known Mutation) (511776)

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)

Hypertrophic Cardiomyopathy (HCM): *ACTC* (Full Gene Sequencing) (252332)

Hypertrophic Cardiomyopathy (HCM): *ACTC* (Known Mutation) (252589)

Hypertrophic Cardiomyopathy (HCM): Eight-gene Profile (*TNNT2, TNNI3, TPM1, MYBPC3, MYH7, MYL2, MYL3, ACTC*) (Full Gene Sequencing) (252300)

Hypertrophic Cardiomyopathy (HCM): Five-gene Minor Profile (*TNNI3, TPM1, MYL2, MYL3, ACTC*) (Full Gene Sequencing) (252297)

Hypertrophic Cardiomyopathy (HCM): *LAMP2* (Full Gene Sequencing) (252340)

Hypertrophic Cardiomyopathy (HCM): *LAMP2* (Known Mutation) (252596)

Hypertrophic Cardiomyopathy (HCM): Metabolic HCM Profile (*PRKAG2, LAMP2*) (Full Gene Sequencing) (252303)

Hypertrophic Cardiomyopathy (HCM): *MYBPC3* (Full Gene Sequencing) (252321)

Hypertrophic Cardiomyopathy (HCM): *MYBPC3* (Known Mutation) (252575)

Hypertrophic Cardiomyopathy (HCM): *MYH7* (Full Gene Sequencing) (252324)

Hypertrophic Cardiomyopathy (HCM): *MYH7* (Known Mutation) (252579)

Hypertrophic Cardiomyopathy (HCM): *MYL2* (Full Gene Sequencing) (252327)

Hypertrophic Cardiomyopathy (HCM): *MYL2* (Known Mutation) (252582)

Hypertrophic Cardiomyopathy (HCM): *MYL3* (Full Gene Sequencing) (252329)

Hypertrophic Cardiomyopathy (HCM): *MYL3* (Known Mutation) (252586)

Hypertrophic Cardiomyopathy (HCM): *PRKAG2* (Full Gene Sequencing) (252335)

Hypertrophic Cardiomyopathy (HCM): *PRKAG2* (Known Mutation) (252592)

Hypertrophic Cardiomyopathy (HCM): Reflex Profile (*TNNT2, MYH7, MYBPC3 TPM1, TNNI3, MYL2, MYL3, ACTC*) (Full Gene Sequencing) (252307)

Hypertrophic Cardiomyopathy (HCM): Three-gene Major Profile (*TNNT2, MYH7, MYBPC3*) (Full Gene Sequencing) (252293)

Hypertrophic Cardiomyopathy (HCM): *TNNI3* (Full Gene Sequencing) (252314)

Hypertrophic Cardiomyopathy (HCM): *TNNI3* (Known Mutation) (252568)

Hypertrophic Cardiomyopathy (HCM): *TNNT2* (Full Gene Sequencing) (252310)

Hypertrophic Cardiomyopathy (HCM): *TNNT2* (Known Mutation) (252565)

Hypertrophic Cardiomyopathy (HCM): *TPM1* (Full Gene Sequencing) (252317)

Hypertrophic Cardiomyopathy (HCM): *TPM1* (Known Mutation) (252572)

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 (DAZ) (512053)

informaSeq℠ Prenatal Test (550746)

informaSeq℠ Prenatal Test With X, Y Analysis (550716)

informaSeq℠ Prenatal Test With Y Analysis (550757)

Inheritest® Carrier Screen (451381)

Inheritest® Select Carrier Screen (451394)

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)

Interleukin 28B (*IL28B*) Polymorphism (rs12979860) (480630)

JAK2 Exon 12 Mutation Analysis (489212)

JAK2^{V617F} Mutation Analysis, Quantitative (489470)

JAK2^{V617F} Mutation Detection (489200)

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JAK2^{V617F} Mutation Analysis, Qualitative, With Reflex to *CALR* Mutation Analysis, JAK2 Exon 12 Mutation Analysis, and MPL Mutation Analysis (489395)

Jaubert Syndrome Type II, DNA Analysis* (511490)

K-ras Gene Mutation Detection (480090)

K-ras Gene Mutation Detection With Reflex to *BRAF* Gene Mutation Detection (480360)

Loeys-Dietz Syndrome (LDS): *TGFBR1* (Full Gene Sequencing) (252413)

Loeys-Dietz Syndrome (LDS): *TGFBR1* (Known Mutation) (252657)

Loeys-Dietz Syndrome (LDS): *TGFBR2* (Full Gene Sequencing)(252416)

Loeys-Dietz Syndrome (LDS): *TGFBR2* (Known Mutation) (252660)

Loeys-Dietz Syndrome (LDS): Two-gene Profile (*TGFBR1*, *TGFBR2*) (Full Gene Sequencing) (252419)

Lung Cancer (489220 [FFPE specimen]; 489240 [fresh-frozen tissue])

Maple Syrup Urine Disease (MSUD) Carrier Test, DNA* (511310)

Marfan Syndrome (MFS): *FBN1* (Full Gene Sequencing) (252406)

Marfan Syndrome (MFS): *FBN1* (Known Mutation) (252654)

Marfan Syndrome to Loeys-Dietz Syndrome Reflex Profile (MFS LDS): *FBN1 TGFBR1*, *TGFBR2* (Full Gene Sequencing) (252409)

Maternal Cell Contamination* (511402)

Methylenetetrahydrofolate Reductase (MTHFR) Thermolabile Variant, DNA Analysis (511238)

Microsatellite Instability (511311)

MGMT (O^6-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)

MSH2 Comprehensive Analysis (511632)

MPL Mutation Analysis (489150)

MPN/CML, FISH (511425)

MPN With Hypereosinophilia, FISH (511444)

MSH2 Deletion/Duplication Analysis (511705)

MSH6 Comprehensive Analysis (511636)

MSH6 Deletion/Duplication Analysis (511720)

Mucolipidosis Type IV Mutation Detection* (511386)

Multiple Myeloma Profile, Fluorescence in situ Hybridization (FISH) (510830)

MYCN Oncology Fluorescence in situ Hybridization (FISH) (510945)

Myelodysplastic Syndrome, Fluorescence in situ Hybridization (FISH) (510599)

Nemaline Myopathy* (450040)

Niemann-Pick Disease, DNA Analysis* (511329)

Non-Small-cell Lung Cancer (NSCLC) Therapeutic Profile: EGFR Mutation Analysis and ALK FISH Analysis (511550)

NPM1 Mutation Analysis (489140)

Opioid CYP2D6 Genotyping (511380)

Organic Acid Analysis, Urine (716720)

P53 Oncology Fluorescence in situ Hybridization (FISH) (510940)

PIK3CA Oncogene Mutation Detection (480880)

PML-RARA Transcript Detection for Acute Promyelocytic Leukemia, Qualitative (480491)

PMS2 Comprehensive Analysis (511630)

PMS2 Deletion/Duplication Analysis (511725)

Prostate Cancer Gene 3 (PCA3) (489160)

Pulmonic Stenosis: *PTPN11* (Full Gene Sequencing) (252399)

Pulmonic Stenosis: *PTPN11* (Known Mutation) (252647)

RB1 Oncology Fluorescence in situ Hybridization (FISH) (510374)

RET Oncology Fluorescence in situ Hybridization (FISH) (510315)

RET Proto-oncogene Mutation Detection (489270)

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)

*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.

For additional information, including specimen requirement, CPT coding, and RUO/IUO status, consult the online Test Menu at www.LabCorp.com.

Test information is included here for quick reference. More detailed descriptions are published in the *Directory of Services and Interpretive Guide* (DoS) in book and electronic form. For the most recent edition of the paper form, contact your LabCorp service representative; to consult the electronic version, visit www.LabCorp.com.

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)

Severe Combined Immunodeficiency (SCID): *ZAP70* (Known Mutation) (252720)

Sex Determination (SRY), DNA Analysis* (510222)

SHOX-DHPLC (500110)

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) Carrier Testing (450010)

SYT Oncology Fluorescence in situ Hybridization (FISH) (510384)

T and B Gene Rearrangement, PCR (480860 [combines 480708 and 480716])

Tamoxifen P450 2D6 Genotyping (511280)

Tay-Sachs Disease, Biochemical, Leukocytes (511246)

Tay-Sachs Disease, Biochemical, Serum (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 480708])

Thiopurine Methyltransferase (TPMT), Enzyme Activity Erythrocytes (510750)

Thoracic Aortic Aneurysms and Dissections (TAAD): Three-gene Profile (*FBN1*, *TGFBR1*, *TGFBR2*) (Full Gene Sequencing) (252422)

Thrombotic Risk Profile, DNA Analysis (512103)

Transthyretin Amyloidosis: *TTR* (Full Gene Sequencing) (252810)

Transthyretin Amyloidosis: *TTR* (Known Mutation) (252813)

UGT1A1 Irinotecan Toxicity (511200)

Uniparental Disomy (UPD), DNA Analysis* (470054)

Uniparental Disomy of Chromosome 14 (UPD 14)* (470060)

Usher Syndrome Type IF*(450060)

Usher Syndrome Type III* (450050)

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)

Warfarin (P450 *2C9* and *VKORC1*) (511460)

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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