



Genetics

Biochemical Genetics

Acetylcholinesterase
 Acetylcholinesterase and Fetal Hemoglobin
 Acylcarnitine, Plasma
 Alpha-1-Antitrypsin Quantitation
 Alpha-Fetoprotein, Amniotic Fluid with Reflex to AchE and Fetal Hgb
 Amino Acid Analysis for MSUD, LC/MS, Plasma
 Amino Acid Analysis, LC/MS, CSF
 Amino Acid Analysis, LC/MS, Plasma
 Amino Acid Analysis, LC/MS, Urine
 Amino Acid Analysis, Limited, LC/MS, Plasma
 Arylsulfatase A
 CAH Panel 1 (21-Hydroxylase vs 11Beta-Hydroxylase Deficiency)
 CAH Panel 3 (Aldosterone Synthase Deficiency)
 CAH Panel 4 (17-Hydroxylase Deficiency in Females)
 CAH Panel 6 (StAR Deficiency)
 CAH Panel 6B (Comprehensive Screen)
 CAH Panel 7 (21-Hydroxylase Deficiency Therapeutic Monitoring)
 CAH Panel 8 (17-Hydroxylase Deficiency in Males)
 CAH Panel 9 (3Beta-Hydroxysteroid Dehydrogenase Deficiency)
 CAH Panel 11, Neonatal Random Urine
 Carnitine, LC/MS/MS
 Carnitine, LC/MS/MS and Acylcarnitine
 Cholinesterase, RBC & Plasma
 Cholinesterase, Serum
 Cholinesterase, Serum, with Dibucaine Inhibition
 Cystine, Quantitative, 24-Hour Urine
 Cystine, Quantitative, Random Urine
 Erythrocyte Protoporphyrin (EP)
 Fetal Hemoglobin, Amniotic Fluid
 Fetal Hemoglobin, Whole Blood
 First Trimester Screen, hCG¹
 First Trimester Screen, Hyperglycosylated hCG (h-hCG)¹
 Glucose-6-Phosphate Dehydrogenase, Quantitative (G-6-PD)
 Hemoglobin A2, Quantitative
 Hemoglobin S, Quantitative
 Hemoglobinopathy Evaluation
Includes RBC, hemoglobin, hematocrit, MCV, MCH, RDW, hemoglobin A1, fetal hemoglobin, hemoglobin A2, and abnormal hemoglobins.
 Homocysteine (Cardiovascular), Serum
 Homocysteine (Nutritional & Congenital)
 Homocysteine, Total, Urine
 Integrated Screen, Part 1¹
 Integrated Screen, Part 2¹
 Maternal Serum AFP
 Methylmalonic Acid, Serum
 Methylmalonic Acid, Urine
 Methylmalonic Acid and Homocysteine (Nutritional & Congenital)
 Organic Acids, Qualitative, Urine
 Organic Acids, Quantitative, Full Panel, Urine
Includes minimum of 76 organic acids.

Penta Screen
 Phenylalanine
 Phenylalanine and Tyrosine
 Porphobilinogen, Quantitative, 24-Hour Urine
 Porphobilinogen, Quantitative, Random Urine
 Porphyrins, Fractionated, Plasma
 Porphyrins, Fractionated, Quantitative, 24-Hour Urine
 Porphyrins, Fractionated, Quantitative, Random Urine
 Porphyrins, Fractionated, Quantitative and Porphobilinogen, 24-Hour Urine
 Porphyrins, Total, Plasma
 Quad Screen
 Sequential Integrated Screen, Part 1¹
 Sequential Integrated Screen, Part 2¹
 Serum Integrated Screen, Part 1¹
 Serum Integrated Screen, Part 2¹
 Sickle Cell Screen
 Sickle Cell Screen with Reflex to Hemoglobinopathy Evaluation
 Triple Screen
 Tryptophan, LC/MS
 Tyrosine

Cytogenetics

Cell Culture for Possible Additional Prenatal Studies
 Chromosome Analysis and AFP w/Reflex to AchE & Fetal Hgb, Amniotic Fluid
 Chromosome Analysis, Amniotic Fluid
 Chromosome Analysis, Blood
 Chromosome Analysis, Blood with Reflex to Postnatal ClariSure™ CGH²
 Chromosome Analysis, Chorionic Villus Sample
 Chromosome Analysis, Follow-up
 Chromosome Analysis, High Resolution
 Chromosome Analysis, High Resolution with Reflex to Postnatal ClariSure™ CGH²
 Chromosome Analysis, Mosaicism
 Chromosome Analysis, Sister Chromatid Exchange
 Chromosome Analysis, Tissue
 Chromosomes, DEB Assay for Fanconi Anemia
 ER/PR/DNA/HER2 w/Reflex to HER2 FISH (Photomicrograph), Paraffin Block³
 ER/PR/DNA/HER2 w/Reflex to HER2 FISH, Paraffin Block³
 ER/PR/HER2 w/Reflex to HER2 FISH, Paraffin Block³
 FISH, Angelman³
 FISH, Chromosome-Specific Probe³
Choose one of the following: chromosome-specific (1-22, X and Y) centromere or chromosome-specific (1-22, X and Y) painting.
 FISH, Chromosome-Specific Probe (x1), ClariSure™ CGH Follow-up³
 FISH, Cri du chat³
 FISH, DiGeorge, Velocardiofacial (VCFS)³
 FISH, Kallmann³

FISH, Microdeletion Syndromes Panel³
Includes FISH markers for DiGeorge, Kallmann, Prader-Willi/Angelman, Smith-Magenis, and Williams syndromes.

FISH, Miller-Dieker³

FISH, Neonatal Screen

FISH, Prader Willi³

FISH, Prenatal Screen

FISH, Product of Conception (POC) Panel³
Includes FISH probes for centromeres X, Y, and 18; and probes for 13q14, 16q11.2, 21q11.2-q22.2, and 22q11.2.

FISH, SKY[®] Marker Chromosome³

FISH, Smith-Magenis³

FISH, SRY/X Centromere³

FISH, Subtelomere Screen³

FISH, Williams³

FISH, Wolf-Hirschhorn³

FISH, X-Linked Ichthyosis Steroid Sulfatase Deficiency³

Male Infertility Genetic Analysis³

Molecular Genetics

Achondroplasia Mutation Analysis³

Alpha-1 Antitrypsin (AAT) Mutation Analysis³

Alpha-Globin Complete²

Alpha-Globin Gene Deletion or Duplication²

Alpha-Thalassemia DNA Mutation Analysis²

Angiotensin Converting Enzyme (ACE) Polymorphism (Insertion/Deletion)³

Angiotensin II Type 1 Receptor (*AGTR1*) Gene 1166A→C Polymorphism³

Ashkenazi Jewish Panel³
Includes mutations associated with cystic fibrosis, Canavan disease, Gaucher disease, Fanconi anemia, Bloom syndrome, Tay-Sachs disease, familial dysautonomia, and Niemann-Pick disease.

Beta-Globin Complete³

Bloom Syndrome DNA Mutation Analysis³

CAH (21-Hydroxylase Deficiency) Common Mutations³

CAH (21-Hydroxylase Deficiency) Rare Mutations³

Canavan Disease Mutation Analysis³

Central Diabetes Insipidus (CDI) Mutations³

CFTR Intron 8 Poly-T Analysis³

Chromosome Analysis, Blood with Reflex to Postnatal ClariSure™ CGH²

Chromosome Analysis, High Resolution with Reflex to Postnatal ClariSure™ CGH²

CKR-5 Gene, DNA Mutation Analysis³

CYP1B1 Mutation Analysis³

Cystic Fibrosis Complete Rare Mutation Analysis, Entire Gene Sequence³

Cystic Fibrosis D1152H Mutation Analysis³

Cystic Fibrosis DNA Analysis, Fetus³

Cystic Fibrosis Gene Deletion or Duplication³

Cystic Fibrosis Rare Mutation Analysis, One Exon³

Cystic Fibrosis Rare Mutation Analysis, Two Exon³

Cystic Fibrosis Screen³

Cytochrome P450 2C19 Genotyping³

Cytochrome P450 2C9 and *VKORC1* Mutation Analysis²

Cytochrome P450 2C9 Genotype³

Cytochrome P450 2D6 Genotype³

Cytochrome P450 2D6/2C19 Genotyping

Dihydropyrimidine Dehydrogenase (DPD) Gene Mutation Analysis³

Factor V (Leiden) Mutation Analysis³

Factor V (Leiden) Mutation Analysis w/Reflex to HR2 Mutation Analysis³

Factor V HR2 Allele DNA Mutation Analysis³

Factor XI Mutation Analysis (Ashkenazi Jewish)³

Familial Dysautonomia Mutation Analysis³

Fanconi's Anemia DNA Mutation Analysis³

Fragile X DNA Analysis, Fetus³

Gaucher Disease, DNA Mutation Analysis³

Genomic Alterations, Postnatal, ClariSure CGH²

Glycogen Storage Disease Type Ia Mutation Analysis (Ashkenazi Jewish)³

Hereditary Hemochromatosis DNA Mutation Analysis³

Huntington Disease Mutation Analysis³

Long Chain Acyl-CoA Dehydrogenase (LCHAD) Mutation Analysis³

Male Infertility Genetic Analysis³

Maple Syrup Disease (MSUD) Mutation Analysis (Ashkenazi Jewish)³

Maternal Cell Contamination Study, STR Analysis¹

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Mutation Analysis³

Methylenetetrahydrofolate Reductase (*MTHFR*), DNA Mutation Analysis³

Mucopolidosis Type IV Mutation Analysis³

Nephrogenic Diabetes Insipidus (Autosomal) Mutations³

Nephrogenic Diabetes Insipidus (X-Linked) Mutations³

Niemann-Pick Disease Mutation Analysis³

Plasminogen Activator Inhibitor-1 (PAI-1) 4G/5G Polymorphism³

Prader-Willi/Angelman Syndrome³

Prothrombin (Factor II) 20210G→A Mutation Analysis³

Resistance to Thyroid Hormone (RTH) Mutation Analysis³

Rett Syndrome Mutation Analysis³

Sickle Cell Anemia, DNA Probe Analysis, Fetus³

Tay-Sachs Disease Mutation Analysis (Ashkenazi Jewish)³

Tay-Sachs Mutation Analysis (non-Ashkenazi Jewish)³

Thrombophilia DNA Mutation Analysis³
Includes factor V (Leiden) and prothrombin (factor II) 20210G→A mutation analyses.

Thrombophilia Mutation Analysis with Reflex to HR2 Mutation Analysis³
Includes factor V (Leiden) and prothrombin (factor II) 20210G→A mutation analyses with reflex to HR2 mutation analysis

Thrombophilia Screen, Inherited³
Includes antithrombin III activity, factor V (Leiden) mutation with reflex to factor V HR2 mutation, protein C activity, free protein S, and prothrombin (factor II) 20210G→A mutation.

TPMT Genotype³

Tremor/Ataxia Syndrome (FXTAS)³

Twin Zygosity¹

Venous Thrombosis Hypercoagulability Panel³
Includes activated protein C resistance (APCR), antithrombin III activity, cardiolipin antibody (IgG, IgM), factor V (Leiden) mutation with reflex to factor V HR2 mutation, homocysteine, lupus anticoagulant dRVVT confirmation, lupus anticoagulant-hexagonal phospholipid neutralization, protein C activity with reflex to protein C antigen, and free protein S.

von Willebrand Disease Mutation Analysis²

XSense,™ Fragile X with Reflex²

XSense,™ Fragile X with Reflex and Chromosome Analysis, Blood²

Y Chromosome Microdeletion, DNA Analysis¹

Oncology-related Genetics

Lymphoid Malignancies

B-Cell Gene Rearrangement, Minimal Residual Disease (MRD)³

B-Cell Gene Rearrangement, PCR²

B-Cell Gene Rearrangement, Qualitative PCR, Plasma-based, Leumeta™³

B-Cell Gene Rearrangement, Quantitative PCR, Plasma-based, Leumeta™³

BCL-2, IHC³

Chromosome Analysis, CLL/LPD

Chromosome Analysis, Hematologic Malignancy

Chromosome Analysis, Lymph Node

Chronic Lymphocytic Leukemia, *IgV_H* Mutation Status, Cell-based³

Chronic Lymphocytic Leukemia, *IgV_H* Mutation Status, Leumeta™³

FISH, ALCL, *ALK*, 2p23 Rearrangements³

FISH, ALL, +4, +10, +17³

FISH, ALL, Extended Panel³

Includes probes for t(12;21), MLL (11q23) rearrangement, t(9;22), MYC (8q24) rearrangement, CDKN2A (p16; 9p21) rearrangement, E2A (TCF3; 19p13.3) rearrangement, IGH (14q32) rearrangement, and enumeration of chromosomes 4, 10 and 17.

FISH, ALL, Pre-B Panel³

Includes probes for t(12;21), MLL rearrangement, t(9;22), and enumeration of chromosomes 4, 10, 17.

FISH, ALL, *TEL/AML1* Translocation 12;21²

FISH, ALL/NHL, *MYC*-BA, 8q24 Rearrangement²

FISH, Alveolar Rhabdomyosarcoma, *FKHR*, *rea* 13q14³

FISH, B-Cell Chronic Lymphocytic Leukemia (B-CLL) Panel³

Includes FISH probes for 11q22.3, 12 cen, 13q14.3, and 17p13 as well as 13q34 (control probe).

FISH, B-Cell Malignancy, *IGH*, 14q32 Rearrangement³

FISH, Burkitt's/NHL/ALL, *IGH/MYC*, t(8;14)³

FISH, EGFR³

FISH, Follicular Lymphoma, *IGH/BCL2*, t(14;18)³

FISH, MALT Lymphoma, *API2/MALT1*, t(11;18)³

FISH, MALT Lymphoma, *MALT1*, 18q21 Rearrangement³

FISH, MALT Lymphoma, *MALT1*, *rea*18q21 with Reflex to *API2/MALT1*, t(11;18)³

FISH, Mantle Cell Lymphoma, *IGH/CCND1*, t(11;14)³

FISH, MLL (11q23) Gene Rearrangement³

FISH, Multiple Myeloma, 13q-, 17p-, *rea* 14q32³

FISH, Multiple Myeloma, Chromosomes 5,9,15³

FISH, Myeloma, 13q,14q, 17p with Reflex to 5,9,15³

FISH, Synovial Sarcoma, *SYT*, *rea* 18q11.2³

FISH, X/Y, Post Bone Marrow Transplant

Follicular Lymphoma, *BCL2/JH* t(14;18), Real-time PCR, Cell-based³

Follicular Lymphoma, *BCL2/JH* t(14;18), Real-time PCR, Leumeta™³

Mantle Cell Lymphoma, *BCL1/JH* t(11;14), Real-time PCR, Cell-based³

Mantle Cell Lymphoma, *BCL1/JH* t(11;14), Real-time PCR, Leumeta™³

T-Cell Receptor (TCR) Gene Rearrangement, Qualitative PCR, Cell-based³

T-Cell Receptor (TCR) Gene Rearrangement, Qualitative PCR, Leumeta™³

T-Cell Receptor (TCR) Gene Rearrangement, Quantitative PCR, Leumeta™³

Myeloid Malignancies

AML1/ETO t(8;21) Quantitative Real-Time PCR³

BCR/ABL Gene Rearrangement, Quantitative PCR, Cell-based³

BCR/ABL Gene Rearrangement, Quantitative PCR, Plasma-based, Leumeta™³

BCR/ABL Gene Rearrangement, Quantitative PCR with Reflex to Subtype³

BCR/ABL Protein Quantitation (Total and Phosphorylated), Leumeta™³

CBFB/MYH11 inv(16), Quantitative Real-Time PCR³

Chromosome Analysis, Hematologic Malignancy

FISH, AML M3, *PML/RARA*, Translocation 15;17³

FISH, AML, *AML1/ETO* Translocation 8;21³

FISH, AML, *CBFB/MYH11*, Inversion 16³

FISH, Chromosome 20q Deletion³

FISH, CML/ALL, *BCR/ABL* Translocation 9;22³

FISH, MDS/Myeloid Panel, -5/5q-, -7/7q-, +8,20q³

FISH, MLL (11q23) Gene Rearrangement³

FISH, Myeloid Disorders Profile³

Includes FISH probes for 5q31, 7q31, 8 centromere, and 20q12 as well as 5p15.2 and 7 centromere (control probes).

FISH, X/Y, Post Bone Marrow Transplant

FLT3 Mutations (ITD and D835)³

PML/RARA t(15;17), Quantitative PCR³

Solid Tumor Malignancies

Chromosome Analysis, Solid Tumor

Colorectal Cancer (CRC) Pharmacogenomic Panel³

Epidermal Growth Factor Receptor (*EGFR*) Mutation Analysis (TK Domain)³

FISH, Bladder Cancer, Bladder Washing

FISH, EGFR³

FISH, Ewing/PNET, *EWSR1*, 22q12 Rearrangements³

FISH, *HER-2/neu*, Paraffin Block

FISH, Lung Cancer³

FISH, *N-myc* Amplification, Neuroblastoma³

FISH, Oligodendroglioma, 1p/19q³

FISH, Prostate Cancer³

FISH, Vysis® UroVysion™, Bladder Cancer

HER2 (HerceptTest®), IHC with Reflex to FISH

MEN2 and *FMTC* Mutations, Exons 10, 11, 13-16³

Microsatellite Instability (MSI), HNPCC³

MLH1 and *MSH2* Mutations (Deletion and Duplication), HNPCC³

MLH1 and *MSH2* Mutations, HNPCC³

MLH1 Mutation, One Exon, HNPCC³

MSH2 Mutation, One Exon, HNPCC³

MSH6 Mutation, HNPCC³

MSH6 Mutation, One Exon, HNPCC³

UGT1A1 Gene Polymorphism (TA Repeat)³

¹This test is performed using a kit that has not been approved or cleared by the FDA. The analytical performance characteristics of this test have been determined by Quest Diagnostics Nichols Institute. This test should not be used for diagnosis without confirmation by other medically established means.

²This test was developed and its performance characteristics have been determined by Quest Diagnostics Nichols Institute. Performance characteristics refer to the analytical performance of the test.

³This test was developed and its performance characteristics determined by Quest Diagnostics Nichols Institute. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. Performance characteristics refer to the analytical performance of the test.

Reflex tests are performed at an additional charge.

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