



● *Biochemical Genetics*

- Acetylcholinesterase
- Acylcarnitine
- Alpha-1-Antitrypsin Phenotype¹
- Alpha-1-Antitrypsin Quantitation
- Alpha-1-Antitrypsin Quantitation & Phenotype¹
- Alpha-Fetoprotein, Amniotic Fluid with Reflex (x2)
(includes reflex to acetylcholinesterase and fetal hemoglobin)
- Amino Acid Analysis, Qualitative, Plasma
- Amino Acid Analysis, Qualitative, Urine
- Amino Acid Analysis, Quantitative, 1 Amino Acid, CSF
- Amino Acid Analysis, Quantitative, 2-5 Amino Acids, CSF
- Amino Acid Analysis, Quantitative, Full Panel, CSF
- Amino Acid Analysis, Quantitative, 1 Amino Acid, Plasma
- Amino Acid Analysis, Quantitative, 2-5 Amino Acids, Plasma
- Amino Acid Analysis, Quantitative, Full Panel, Plasma
- Amino Acid Analysis, Quantitative, 1 Amino Acid, Urine
- Amino Acid Analysis, Quantitative, 2-5 Amino Acids, Urine
- Amino Acid Analysis, Quantitative, Full Panel, Urine
- Carnitine
- Cholinesterase with Dibucaine Inhibition, Serum
- Cystine, Qualitative, Urine
- Cystine, Quantitative, 24-Hour Urine
- Cystine, Quantitative, Random Urine
- Delta Aminolevulinic Acid, 24-Hour Urine
- Delta Aminolevulinic Acid, Random Urine
- Fetal Hemoglobin, Amniotic Fluid
- Glucose-6-Phosphate Dehydrogenase, Quantitative
- Hemoglobinopathy Evaluation
(includes RBC, hemoglobin, hematocrit, MCV, MCH, RDW, hemoglobin A1, fetal hemoglobin, hemoglobin A2, and abnormal hemoglobins)
- Homocysteine (Cardiovascular), FPIA, Serum
- Homocysteine (Nutritional & Congenital), FPIA, Serum
- Homocysteine, Total, Urine
- Inborn Errors of Metabolism, Qualitative
(includes amino acids and organic acids)
- Inborn Errors of Metabolism, Quantitative
(includes amino acids, organic acids, and carnitine)
- Invasive Trophoblast Antigen (ITA) (Tumor)¹
- Invasive Trophoblast Antigen (ITA) (Maternal Screen), Urine¹
- Invasive Trophoblast Antigen (ITA) (Pregnancy)¹
- Maternal Serum 1 (AFP)
- Maternal Serum 3 (AFP, hCG, uE3)
- Maternal Serum 4 (AFP, hCG, uE3, Inhibin A)
- Methylmalonic Acid, Serum
- Methylmalonic Acid, Urine
- Organic Acids, Qualitative, Urine
- Organic Acids, Quantitative, Full Panel, Urine
- Phenylalanine
- Phenylalanine and Tyrosine
- Porphobilinogen, Quantitative, 24-Hour Urine
- Porphobilinogen, Quantitative, Random Urine
- Porphyrins, Fractionated, Plasma
- Porphyrins, Fractionated, Quantitative, 24-Hour Feces
- Porphyrins, Fractionated, Quantitative, 24-Hour Urine
- Porphyrins, Fractionated, Quantitative, Random Urine
- Porphyrins, Fractionated, Quantitative and Porphobilinogen, 24-Hour Urine
- Porphyrins, Total, Plasma
- Sickle Cell Screen
- Tyrosine

● *Cytogenetics*

- Cell Culture for Possible Additional Prenatal Studies
- Chromosome Analysis & AFP w/Reflex to Acetylcholinesterase & Fetal Hemoglobin, Amniotic Fluid
- Chromosome Analysis, Amniotic Fluid
- Chromosome Analysis, Blood
- Chromosome Analysis, Breakage
- Chromosome Analysis, Chorionic Villus Sample
- Chromosome Analysis, Follow-up
- Chromosome Analysis, High Resolution
- Chromosome Analysis, Mosaicism
- Chromosome Analysis, Sister Chromatid Exchange
- Chromosome Analysis, Tissue
- FISH, Acrocentric p-arm¹
- FISH, Angelman¹
- FISH, Chromosome-Specific Probe¹
(Choose one of the following: Chromosome-Specific [1-22, X and Y] Centromere or Chromosome-Specific [1-22, X and Y] Painting)
- FISH, Cri du chat¹
- FISH, DiGeorge, Velocardiofacial (VCFS)¹
- FISH, Kallmann¹
- FISH, Marker Chromosome 15¹
- FISH, Microdeletion Syndromes Panel¹
- FISH, Miller-Dieker¹
- FISH, Neonatal Screen
- FISH, Prader Willi¹
- FISH, Prenatal Screen, AneuVysion[®]
- FISH, SKY™ Marker Chromosome¹
- FISH, Smith-Magenis¹
- FISH, SRY/X Centromere¹
- FISH, Subtelomere Screen¹
- FISH, Williams¹
- FISH, Wolf-Hirschhorn¹
- FISH, X-Linked Ichthyosis Steroid Sulfatase Deficiency¹

● Molecular Genetics

- Alpha-Thalassemia DNA Mutation Analysis
- Angiotensin Converting Enzyme (ACE) Polymorphism (Insertion/Deletion)¹
- Angiotensin II Type 1 Receptor (ATR1) Gene 1166A→C Polymorphism¹
- Ashkenazi Jewish Panel¹
(includes mutations associated with cystic fibrosis, Canavan disease, Gaucher disease, Fanconi anemia, Bloom syndrome, Tay-Sachs disease, familial dysautonomia, and Niemann-Pick disease)
- Bloom Syndrome DNA Mutation Analysis¹
- Canavan Disease Mutation Analysis¹
- CKR-5 Gene, DNA Mutation Analysis
- Cystic Fibrosis DNA Analysis¹
- Cystic Fibrosis DNA Analysis, Fetus¹
- Cystic Fibrosis Carrier Screen¹
- Cystic Fibrosis Rare Mutation Analysis, One Exon¹
- Cystic Fibrosis Rare Mutation Analysis, Two Exons¹
- Cystic Fibrosis Complete™ Rare Mutation Analysis, Entire Gene Sequence¹
- Cytochrome P450 2C19 Genotype¹
- Cytochrome P450 2C9 Genotype¹
- Cytochrome P450 2D6 Genotype¹
- Factor V (Leiden) Mutation Analysis¹
- Factor V HR2 Allele DNA Mutation Analysis¹
- Factor V (Leiden) Mutation Analysis w/Reflex to HR2 Mutation Analysis¹
- Familial Dysautonomia Mutation Analysis¹
- Fanconi Anemia DNA Mutation Analysis¹
- Fragile X, DNA Probe Analysis¹
- Fragile X, DNA Probe Analysis¹ and Chromosome Analysis, Blood
- Fragile X, DNA Probe Analysis, Fetus¹
- Gaucher Disease, DNA Mutation Analysis¹
- Hereditary Hemochromatosis DNA Mutation Analysis¹
- Kennedy Disease Mutation Analysis¹
- Long Chain Acyl-CoA Dehydrogenase (LCHAD) Mutation Analysis¹
- Maternal Cell Contamination (MCC) Study, STR Analysis²
- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Mutation Analysis¹
- Methylene tetrahydrofolate Reductase (MTHFR) DNA Mutation Analysis¹
- Mucopolidiosis Type IV Mutation Analysis¹
- Niemann-Pick Disease Mutation Analysis¹
- Plasminogen Activator Inhibitor-1 (PAI-1) 4G/5G Polymorphism³
- Prader-Willi/Angelman Syndrome DNA Methylation Analysis¹
- Prothrombin (Factor II) 20210G→A Mutation Analysis¹
- Sickle Cell Anemia, DNA Probe Analysis
- Tay Sachs Disease Mutation Analysis¹
- Thrombophilia DNA Mutation Analysis¹
[Includes factor V (Leiden) and prothrombin (factor II) 20210G→A mutation analyses]
- Thrombophilia Mutation Analysis with Reflex to HR2 Mutation Analysis¹
[Includes factor V (Leiden) and prothrombin (factor II) 20210G→A mutation analyses with reflex to HR2 mutation analysis]
- Twin Zygosity²

● Oncology Related Genetics

Lymphoid Malignancies

- B-Cell Gene Rearrangement, PCR, Blood, Bone Marrow, or Frozen Tissue¹
- B-Cell Gene Rearrangement, PCR, Paraffin Block¹
- *bcl-1*, t(11;14) Translocation, PCR¹
- *bcl-2* Gene Translocation (*mbr*), Quantitative PCR with Reflex to (*mcr*)¹
- *bcl-2* Gene Translocation (*mbr*), Quantitative PCR¹
- *bcl-2*, IHC¹
- Chromosome Analysis, CLL/LPD
- Chromosome Analysis, Hematologic Malignancy
- Chromosome Analysis, Lymph Node
- FISH, ALL, TEL/AML1 Translocation 12,21¹
- FISH, B-Cell Malignancy Panel¹
(includes [+12], del(13)(q14), *rea*(14)(q32), and [17p-])
- FISH, MLL (11q23) Gene Rearrangement¹
- FISH, Trisomy 12 and 13q Deletion for CLL Diagnosis¹
- FISH, X/Y, Post Bone Marrow Transplant
- T-cell Receptor Gene Rearrangement, PCR/TTGE¹

Myeloid Malignancies

- *bcr/abl* Gene Rearrangement, Qualitative PCR with Reflex to Translocation Type¹
- *bcr/abl* Gene Rearrangement, Qualitative PCR¹
- Chromosome Analysis, Hematologic Malignancy
- Chromosome Analysis, Hematologic Malignancy & *bcr/abl* Gene Rearrangement, Qualitative PCR¹
- FISH, AML M3, PML/RARA, Translocation 15,17¹
- FISH, AML, AML1/ETO Translocation 8,21¹
- FISH, AML, CBFβ/MYH11, Inversion 16¹
- FISH, Chromosome 20q Deletion¹
- FISH, Chronic Myelogenous Leukemia (CML) Blast Crisis¹
- FISH, CML/ALL, *bcr/abl* Translocation 9,22¹
- FISH, *c-myc* Amplification¹
- FISH, MLL (11q23) Gene Rearrangement¹
- FISH, Myeloid Disorders Panel¹
- FISH, X/Y, Post Bone Marrow Transplant
- Retinoic Acid Receptor alpha (RAR-alpha) PML t(15;17) Translocation, PCR¹

Solid Tumor Malignancies

- Chromosome Analysis, Solid Tumor
- FISH, N-*myc* Amplification, Neuroblastoma¹
- FISH, PathVysion® HER-2/*neu*, Paraffin Block
- FISH, Vysis UroVysion™, Bladder Cancer Recurrence
- HER2 (HercepTest™), IHC with Reflex to HER2, FISH
- MEN2 and FMTC Mutations, Exons 10, 11, 13-16¹

1. This 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.

2. This 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.

3. This 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. Performance characteristics refer to the analytical performance of the test.

Polymerase chain reaction (PCR) is performed pursuant to a license agreement with Roche Molecular Systems, Inc.

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