

BCM-MEDICAL GENETICS LABORATORIES

PHONE: 800-411-GENE | FAX: 713-798-2787 | www.bcmgeneticlabs.org

NEW YORK STATE APPROVED TEST LIST

DIVISION	TEST CODE	TITLE	METHODOLOGY	SAMPLE TYPE	STATUS
BIO	4300	Acylcarnitine Analysis - Plasma	Tandem Mass Spectroscopy	Plasma	Approved
BIO	4310	Carnitine Determination - Plasma	Tandem Mass Spectroscopy	Plasma	Approved
BIO	4130, 4260	Creatine and Guanidinoacetate Determination	Tandem Mass Spectroscopy	Plasma; Urine	Approved
BIO	4200	Organic Acid Screen - Urine	Tandem Mass Spectroscopy	Urine	Approved
BIO	4210	Orotic Acid/Orotidine Determination - Urine	Tandem Mass Spectroscopy	Urine	Approved
BIO	4509	Adenosine Deaminase Enzyme Analysis	Radiometric	Red Blood Cells (RBC)	Grandfather Clause
BIO	4510	Adenosine Deaminase Enzyme Analysis	Radiometric	Skin Fibroblast Culture	Grandfather Clause
BIO	4511	Adenosine Deaminase Enzyme Analysis	Radiometric	White Blood Cells (WBC)	Grandfather Clause
BIO	4507	Adenosine Deaminase Enzyme Analysis (Prenatal Diagnosis - Amniocytes)	Radiometric	Chorionic Villi Sample (CVS); Amniotic Fluid	Grandfather Clause
BIO	4517	Alpha-Galactosidase A Enzyme Analysis	Fluorimetric	White Blood Cells (WBC)	Grandfather Clause
BIO	4576	Alpha-L-iduronidase Enzyme Analysis	Fluorimetric	White Blood Cells (WBC)	Grandfather Clause
BIO	4160	Amino Acid Analysis - Cerebrospinal Fluid	HPLC with Ninhydrin Reaction	Cerebrospinal fluid	Grandfather Clause
BIO	4100	Amino Acid Analysis - Plasma	HPLC with Ninhydrin Reaction	Plasma	Grandfather Clause
BIO	4240	Amino Acid Analysis - Urine	HPLC with Ninhydrin Reaction	Urine	Grandfather Clause
BIO	4536	Arginase Enzyme Analysis	Spectrophotometric	Red Blood Cells (RBC)	Grandfather Clause
BIO	4524	Argininosuccinate Lyase Enzyme Analysis	Radioisotopic	Red Blood Cells (RBC)	Grandfather Clause
BIO	4525	Argininosuccinate Lyase Enzyme Analysis	Radioisotopic	Skin Fibroblast Culture	Grandfather Clause
BIO	4545	Argininosuccinate Synthetase Enzyme Analysis	Radiometric	Skin Fibroblast Culture	Grandfather Clause
BIO	4538	Arylsulfatase A Enzyme Analysis	Spectrophotometric	White Blood Cells (WBC)	Grandfather Clause
BIO	4549	Beta-Galactosidase Enzyme Analysis	Fluorimetric	White Blood Cells (WBC)	Grandfather Clause
BIO	4554	Beta-Glucosidase Enzyme Analysis	Fluorimetric	White Blood Cells (WBC)	Grandfather Clause
BIO	4555	Biotinidase Enzyme Analysis	Colorimetric	Serum	Grandfather Clause
BIO	4561	Carbamoyl Phosphate Synthetase Enzyme Analysis	Colorimetric	Liver	Grandfather Clause
BIO	4566	Galactocerebrosidase Enzyme Analysis	Radiometric	White Blood Cells (WBC)	Grandfather Clause
BIO	4569	Hexosaminidase A & B Enzyme Analysis	Fluorimetric	Serum	Grandfather Clause
BIO	4140	Homocysteine Determination - Plasma	Tandem Mass Spectroscopy	Plasma	Grandfather Clause
BIO	4572	Hypoxanthine-Guanine Phosphoribosyltransferase Enzyme Analysis	Radiometric	Red Blood Cells (RBC)	Grandfather Clause
BIO	4504	Lysosomal Acid Lipase Analysis	Radiometric	White Blood Cells (WBC)	Grandfather Clause

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BIO	4150	Methylmalonic Acid - Plasma	Tandem Mass Spectroscopy	Plasma	Grandfather Clause
BIO	4582	Ornithine Transcarbamylase Enzyme Analysis	Colorimetric	Liver	Grandfather Clause
BIO	4120	Phenylalanine Determination - Blood Spot	Tandem Mass Spectroscopy	Blood spot	Grandfather Clause
BIO	4110	Phenylalanine Determination - Plasma	HPLC with Ninhydrin Reaction	Plasma	Grandfather Clause
BIO	4585	Phosphatidylinositol Bisphosphate Phosphatase Enzyme Analysis	Radiometric	Skin Fibroblast Culture	Grandfather Clause
BIO	4592	Purine Nucleoside Phosphorylase Enzyme Analysis	Radiometric	Red Blood Cells (RBC)	Grandfather Clause
BIO	4594	Purine Nucleoside Phosphorylase Enzyme Analysis	Radiometric	White Blood Cells (WBC)	Grandfather Clause
BIO	4220	Purine Panel - Urine	Tandem Mass Spectroscopy	Urine	Grandfather Clause
BIO	4603	Sialidase Enzyme Analysis	Fluorimetric	Skin Fibroblast Culture	Grandfather Clause
BIO	4608	Sphingomyelinase Enzyme Assay	Radiometric	White Blood Cells (WBC)	Grandfather Clause
BIO	4615	Steroid Sulfatase Enzyme Analysis	Radiometric	White Blood Cells (WBC)	Grandfather Clause
BIO	4630	Steroid Sulfatase Enzyme Analysis (Prenatal Diagnosis)	Radiometric	Amniotic Fluid Cultured Cells; Cultured Chorionic Villus Cells	Grandfather Clause
BIO	4250	Succinylacetone Determination - Urine	Gas Chromatography/Mass Spectrometry	Urine	Grandfather Clause
BIO	4620	Tay-Sachs Carrier Testing	Fluorimetric	White Blood Cells (WBC)	Conditionally Approved
BIO	4627	White Blood Cell Cystine	Tandem Mass Spectroscopy	White Blood Cells (WBC)	Grandfather Clause
CMA	8655	Chromosomal Microarray Analysis - HR	Chromosome Analysis	Whole Blood; Product of Conception; Skin Biopsy	Approved
CMA	8665	Chromosomal Microarray Analysis - HR + SNP	Chromosome Analysis	Whole Blood; Product of Conception; Skin Biopsy	Conditionally Approved
CMA	8670, 8671	Expanded Prenatal Chromosomal Microarray Analysis	Chromosome Analysis	Chorionic Villus Sample (CVS); Amniotic Fluid Sample	Conditionally Approved
CYTO	8600	Chromosome Analysis - Blood	Standard methodology is used for G-banded chromosome Analysis	Whole Blood	Approved
CYTO	8700	Chromosome Analysis - Prenatal - CVS	Standard methodology is used for G-banded chromosome Analysis	Chorionic Villus Sample (CVS)	Approved
CYTO	8464	FISH Analysis - Cri-Du-Chat Syndrome	FISH	Whole Blood	Approved
CYTO	8486	FISH Analysis - DiGeorge/Velocardiofacial Syndrome Type 1 (22q)	FISH	Whole Blood	Approved
CYTO	8476	FISH Analysis - Prader-Willi Syndrome Panel	FISH	Whole Blood	Approved
CYTO	8410	FISH Analysis - Prenatal Aneuploidy	FISH	Chorionic Villus Sample (CVS)	Approved
CYTO	8483	FISH Analysis - Williams Syndrome	FISH	Whole Blood	Approved
CYTO	8484	FISH Analysis - Wolf-Hirschhorn Syndrome	FISH	Whole Blood	Approved

All of the tests listed above (approved or grandfather clause) do not require restricted permit for the test order.

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CYTO	8500	Chromosome Analysis - Prenatal - Amniotic Fluid with AFP	Standard methodology is used for G-banded chromosome Analysis	Amniotic Fluid	Grandfather Clause
CYTO	8458	FISH Analysis - Alagille Syndrome, JAG1 Related	FISH	Whole Blood	Grandfather Clause
DNA	6000	Achondroplasia Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6067	ARX Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Approved
DNA	6180, 6190	ASS1 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6012, 6537	Bloom Syndrome Ashkenazic Mutation Panel	MALDI-TOF; also offer whole gene sequencing	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6070	Canavan Disease Ashkenazic Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6017	CFTR - 5T Variant Analysis	MALDI-TOF	Whole Blood	Approved
DNA	7600, 7601	CFTR- Related Disorders Mutation Panel	BioMark_Fluidigm	Whole Blood; Cultured cells (amniotic or CV); Amniotic Fluid; Chorionic Villus	Approved
DNA	6027	DRPLA Repeat Expansion Analysis	PCR-Amplification followed by gel electrophoresis	Whole Blood	Approved
DNA	6084, 6085	Duchenne-Becker muscular dystrophy	Sequencing	Whole Blood	Approved
DNA	6028	Factor V Leiden Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6030	Familial Dysautonomia	MALDI-TOF	Whole Blood	Approved
DNA	6570, 6572	FMR1 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6635	FOXP1 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6031	Friedreich Ataxia Repeat Expansion Analysis	Southern Blot, PCR	Whole Blood	Approved
DNA	6033	Gaucher Disease Ashkenazic Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6019	GJB2 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Approved
DNA	6063, 6135	GLA Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6123, 6115	GPR143 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6035	HFE Mutation Panel	MALDI-TOF	Whole Blood	Approved

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DNA	6034, 6099	Huntington Disease Repeat Expansion Analysis	PCR and Southern Blot	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6036, 6100	Incontinentia Pigmenti Common Deletion Analysis	Southern Blot	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6037	Kennedy Disease Repeat Expansion Analysis	PCR	Whole Blood	Approved
DNA	6430, 6440	LIPA	Sequencing	Whole Blood	Approved
DNA	6039, 6101	Lowe syndrome	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6045	MTHFR Thermolabile Variant Analysis	MALDI-TOF	Whole Blood	Approved
DNA	6041, 6105	Myotonic Dystrophy Type 1 Repeat Expansion Analysis	PCR-amplification followed by Southern Blot with densitometry	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6047	Niemann-Pick Disease Type A Ashkenazic Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6127, 6133	PLP1 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6050	Prader-Willi Syndrome - Methylation Analysis	Southern Blot	Whole Blood	Approved
DNA	6048	Prothrombin Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6065	PTPN11 Noonan Syndrome	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6053	Sickle Cell Disease Mutation Analysis	MALDI-TOF	Whole Blood	Approved
DNA	6059	SMN1 Deletion Analysis	PCR-amplification followed by gel electrophoresis	Whole Blood	Approved
DNA	6054	Spinocerebellar Ataxia Type 1 Repeat Expansion Analysis	PCR-amplification followed by gel electrophoresis, Southern analysis	Whole Blood	Approved
DNA	6055	Spinocerebellar Ataxia Type 10 Repeat Expansion Analysis	PCR-amplification followed by gel electrophoresis, Southern analysis	Whole Blood	Approved
DNA	6060	SRY Molecular Analysis	PCR-amplification followed by gel electrophoresis	Whole Blood	Approved
DNA	6066	Tay-Sachs Disease Ashkenazic Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6006	Angelman Syndrome Methylation Analysis	Southern Blot	Whole Blood	Grandfather Clause
DNA	6001	Ashkenazic Genetic Disease Panel	Allele-specific genotyping by MALDI-TOF mass spectrometry	Whole Blood	Grandfather Clause
DNA	6360	ASL Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause

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DNA	6126	CDKL5 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause
DNA	6370	CFTR Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause
DNA	6870	CYP2C19 Genotyping	Sequence analysis is performed in both forward and reverse directions	Blood, Saliva	Conditionally Approved
DNA	6084	DMD Sequence Analysis	DNA amplification by multiplex PCR, allele specific primer extension, microarray hybridization and Fluorescence signal detection	Whole Blood	Grandfather Clause
DNA	6350	DMD Deletion/Duplication Analysis	High resolution array CGH	Whole Blood	Conditionally Approved
DNA	6355	GJB6 Deletion/Duplication Analysis	PCR amplification and electrophoresis	Whole Blood	Grandfather Clause
DNA	6740	LDLR Comprehensive - Sequence & Deletion/Duplication Analysis	Sequence analysis is performed in both forward and reverse directions and Multiplex Ligation-dependent Probe Amplification (MLPA) analysis is performed to detect gross deletion and duplications	Whole Blood	Grandfather Clause
DNA	6068	MECP2 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause
DNA	6706	MLH1 Deletion/Duplication Analysis	Multiplex Ligation-dependent Probe Amplification (MLPA) analysis is performed to detect gross deletion and duplications	Whole Blood	Grandfather Clause
DNA	6095	MLH1 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause
DNA	6096	MSH2 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause
DNA	6113	Spinocerebellar Ataxia Type 1 Repeat Expansion Analysis (Prenatal Diagnosis)	PCR-amplification followed by gel electrophoresis, Southern analysis	Chorionic Villi Sample (CVS); Amniotic Fluid	Grandfather Clause
DNA	6062	Thrombophilia Mutation Panel	Allele-specific genotyping by MALDI-TOF mass spectrometry	Whole Blood	Grandfather Clause
DNA	6118	TP53 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause
DNA	6007	UBE3A Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Grandfather Clause
DNA	6072	UBE3A Sequence Analysis (Prenatal Diagnosis)	Sequencing analysis of the region(s) containing the familial alteration(s) is performed in the forward and reverse directions	Chorionic Villi Sample (CVS); Amniotic Fluid	Grandfather Clause