



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	Gas Chromatography/Mass Spectrometry	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	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	4555	Biotinidase Enzyme Analysis	Colorimetric	Serum	Grandfather Clause
BIO	4561	Carbamoyl Phosphate Synthetase Enzyme Analysis	Colorimetric	Liver	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	4504	Lysosomal Acid Lipase Analysis	Radiometric	White Blood Cells (WBC)	Grandfather Clause
BIO	4582	Ornithine Transcarbamylase Enzyme Analysis	Colorimetric	Liver	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



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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	Approved
CMA	8665, 8639	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), Amniotic fluid	Approved
CYTO	8483	FISH Analysis - Williams Syndrome	FISH	Whole Blood; Amniotic Fluid Sample; Chorionic Villus Sample (CVS)	Approved
CYTO	8484	FISH Analysis - Wolf-Hirschhorn Syndrome	FISH	Whole Blood	Approved
CYTO	8500, 8530, 8501, 8550	Chromosome Analysis - Prenatal - Amniotic Fluid with AFP	Standard methodology is used for G-banded chromosome Analysis	Amniotic Fluid	Grandfather Clause
DNA	6000	Achondroplasia Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6067, 6074	ARX Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Approved
DNA	6180, 6185, 6190	ASS1 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved



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DNA	6012, 6535, 6536, 6537	Bloom Syndrome Ashkenazic Mutation Panel	MALDI-TOF; also offer whole gene sequencing	Chorionic Villus Sample (CVS); Amniotic Fluid; Whole Blood	Approved
DNA	6017	CFTR - 5T Variant Analysis	MALDI-TOF	Whole Blood	Approved
DNA	6085	Duchenne-Becker muscular dystrophy	Sequencing	Whole Blood; EDTA	Approved
DNA	6028	Factor V Leiden Mutation Panel	MALDI-TOF	Whole Blood	Approved
DNA	6570, 6571, 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, 6636, 6637	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	6019, 6078	GJB2 Sequence Analysis	Sequence analysis is performed in both forward and reverse directions	Whole Blood	Approved
DNA	6063, 6136, 6135	GLA 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
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, 6435, 6440	LIPA	Sequencing	Whole Blood	Approved
DNA	6039, 6102, 6101	Lowie 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	6127, 6128, 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



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DNA	6065, 6107, 6106	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	6060	SRY Molecular Analysis	PCR-amplification followed by gel electrophoresis	Whole Blood	Approved
DNA	6006	Angelman Syndrome Methylation Analysis	Southern Blot	Whole Blood	Grandfather Clause
DNA	6350	DMD Deletion/Duplication Analysis	High resolution array CGH	Whole Blood	Conditionally Approved
DNA	6062	Thrombophilia Mutation Panel	Allele-specific genotyping by MALDI-TOF mass spectrometry	Whole Blood	Grandfather Clause
DNA	6007, 6073	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
Mito	21400, 21402	Noonan Spectrum Disorders Panel	Next Generation Sequencing	Whole Blood, Saliva, Tissue: Prenatal, extracted DNA, Chorionic villus sample (CVS)	Conditionally Approved
DNA	60101, 60201, 60206, 60301, 60306, 60401, 60406, 60105, 60205, 60305, 60405	Carrier Tests	NGS, TaqMan, PCR, MLPA	Whole Blood, Saliva	Conditionally Approved
DNA	1500	Whole Exome Sequencing	NGS	Whole Blood: EDTA, cultured cells: fibroblasts, purified DNA	Approved
DNA	1600, 1390, 1722, 1622	Whole Exome Sequencing (Trio, Critical, Prenatal, and Total BluePrint Panel)	NGS	Whole Blood: EDTA, cultured cells: fibroblasts, purified DNA	Conditionally Approved
DNA	23000, 22350	Hereditary High Risk Breast Cancer/ BRCA1 and BRCA2 Panel	NGS	Whole Blood, Saliva	Conditionally Approved
Mito	21200	PreSeek Non-invasive Prenatal Gene Sequencing Screen	NGS	Whole Blood, Saliva	Approved
DNA	60897	Cystic Fibrosis (CFTR gene)	NGS	Whole Blood, Saliva, fibroblasts	Conditionally Approved
DNA	6573	Fragile X (FMR1 gene)	PCR	Whole Blood, Saliva	Conditionally Approved



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DNA	6059	Spinal Muscular Atrophy (SMA) (SMN1 and SMN2 genes)	MLPA	Whole Blood	Conditionally Approved
DNA	60290, 60295	GeneAware v3 (274) Carrier screening	NGS	Whole Blood, extracted DNA, saliva	Conditionally Approved
DNA	62890, 62895	GeneAware v3 (289) Carrier screening	NGS	Whole Blood, extracted DNA, saliva	Conditionally Approved
DNA	24001	Expanded Common Hereditary Cancer Panel	NGS	Whole Blood, extracted DNA, saliva, cultured cells, buccal swab	Conditionally Approved
DNA	60140, 60145, 64000, 64005	Comprehensive GeneAware v3 (421 genes) carrier screening	NGS	Whole Blood, extracted DNA, saliva, buccal swab	Conditionally Approved