Baylor Miraca Genetics Laboratories

Genetic Testing for Autism Spectrum Disorders

BIOCHEMICAL, MOLECULAR, CYTOGENETIC, MITOCHONDRIAL

Thorough genetic evaluation of patients with Autism Spectrum Disorders (ASD) is critical for appropriate medical management and family counseling. The American College of Medical Genetics has approved a systematic Practice Guideline to aid clinicians with this complex diagnostic schema. Based on these recommendations, Baylor Miraca Genetics Laboratories (BMGL) have developed a panel of tests to provide the physician with a comprehensive genetic workup for appropriate patients with ASD to obtain information about the etiology and to provide appropriate medical management as well as genetic counseling to the family.

The BMGL has the unique ability to offer metabolic, molecular and cytogenetic analyses, which encompass the multitude of tests recommended in the ACMG guideline. BMGL also offers a uniquely comprehensive evaluation of mitochondrial disorders, which may contribute to susceptibility for ASD. Our new ASD-Panels are designed to reflect the ACMG clinical guideline for female and male patients. Please note, any test in the panel may be ordered individually to meet the needs of each patient.

Genetic testing for autism requires biochemical, molecular (fragile X and gene sequencing), cytogenetic (microarray), and sometimes mitochondrial studies. See Schaefer, Mendelsohn, ACMG Practice Guidelines Genet Med 10:301-305, 2008 (PMID 18414214).

Available ASD Testing	Chromosomal Microarray	Fragile X Testing	Biochemistry	MECP2 Sequencing & Deletion/Duplication
Comprehensive Autism Panel	+	+	**Autism 8-Plex Panel**	**Females Only**

Test codes 8100 for males, 8110 for females

REQUIREMENTS:

- **8100** MALE SPECIFIC COMPREHENSIVE AUTISM PANEL: Chromosomal Microarray Analysis - HR+SNP Screen (Comprehensive); FMR1-Related Disorders Repeat Expansion Analysis, and Biochemistry 8-Plex.
- **8110** FEMALE SPECIFIC COMPREHENSIVE AUTISM PANEL: Chromosomal Microarray Analysis - HR+SNP Screen (Comprehensive); FMR1-Related Disorders Repeat Expansion Analysis, and Biochemistry 8-Plex; MECP2-Related Disorders Sequencing and Deletion/Duplication Analysis

Special Note: *Tests will be reported individually and no comprehensive summary of the testing is provided.*

SPECIMEN REQUIREMENTS	
Test Requisition:	**Autism Testing**
Specimen Type:	Blood - EDTA [along with Blood - Heparin, Plasma, & Urine]
Requirements:	Draw blood in an EDTA (purple-top) tube(s) and send 10 cc
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier to arrive within 36 hours of collection. Do not heat or freeze.
Specimen Type:	Blood - Heparin [along with Blood - EDTA, Plasma, & Urine]
Requirements:	Draw blood in a Heparin (green-top) tube(s) and send 3-5 cc
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier to arrive within 36 hours of collection. Do not heat or freeze.
Specimen Type:	Plasma [along with Blood - Heparin, Blood - EDTA, & Urine]
Requirements:	Draw blood in a Heparin (green-top) tube(s) and separate as soon as possible. Send 2 cc of plasma. Store the specimen frozen at -20C.
Shipping Conditions:	Ship frozen on dry ice in an insulated container by overnight courier.
Specimen Type:	Urine [along with Blood - Heparin, Blood - EDTA, & Plasma]
Requirements:	Send 3-5 cc of a random urine. Do not add preservatives. Store the specimen frozen at -20C.
Shipping Conditions:	Ship frozen on dry ice in an insulated container by overnight courier.

Ship specimens at ambient temperature in an insulated container to arrive within 36 hours of collection.

SHIPPING AND HANDLING:
2450 Holcombe, Grand Blvd. Receiving Dock, Houston, Tx 77021-2024

Prepaid shipping kits are available upon request: Please call us at 1-800-411-GENE (4363).
• Test Requisition: Autism Testing • Turnaround Time: 3-15 days

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INDIVIDUAL TESTS AVAILABLE FOR PATIENTS REQUIRING A MORE TAILORED GENETIC EVALUATION

High Resolution Cytogenetic Analysis

- 8665 Chromosomal Microarray Analysis (CMA) - HR + SNP *(Comprehensive)*
- 8600 Chromosome analysis, blood

Fragile X Testing

- 6032 FMR-1 Related Disorders Repeat Expansion Analysis

Biochemistry 8-Plex - Test Code 4000

- 4100 Plasma Amino Acid Analysis
- 4200 Urine Organic Acid Profile
- 4130 Plasma Creatine/Guanidinoacetate Analysis
- 4135 Urine Carnitine Biosynthesis Panel
- 4145 Plasma Carnitine Biosynthesis Panel
- 4260 Urine Creatine/Guanidinoacetate Analysis
- 4300 Plasma Acylcarnitine Profile
- 4220 Urine Purine Analysis
- 4215 Urine Pyrimidine Analysis

MECP2 Sequencing and Deletion Test

- 6068 MECP2 Related Disorders Sequencing
- 6069 MECP2 Related Disorders Deletion/ Duplication analysis

Autism-related Gene Sequencing

- 6126 CDKL5-Related Atypical Rett syndrome Sequencing
- 6540 SLC9A6 X-linked Angelman-like syndrome Sequencing
- 6505 PTEN-Related Disorders Sequencing
- 6007 Angelman syndrome - UBE3A Sequencing
- 6067 ARX Related Disorders Sequencing

Other Gene Sequencing

- 6127 PLP1-Related Disorders Sequencing
- 6121 RECQL4-Related Disorders - Sequencing
- 6165 CHARGE syndrome - CHD7 Sequencing
- 6065 Noonan syndrome - PTPN11 Sequencing
- 6460 Noonan syndrome - SOS1 Sequencing
- 6445 Noonan syndrome - KRAS Sequencing
- 6475 Noonan syndrome - RAF1 Sequencing
- 6240 Lesch-Nyhan syndrome - HPRT1 Sequencing;
- 2510 TMLHE Deficiency (TMLHE) Sequencing *(only indicated if enzyme deficiency first documented)*

Mitochondrial Testing to be Considered in Autism Evaluation

(Testing is specimen specific based on clinical findings. Please refer to mitochondrial algorithm on our web site: www.BMGL.com)

- 3000 Mitochondrial DNA screening panel
- 2130 Mitochondrial DNA depletion/integrity panel
- 3500 MitoMet oligonucleotide array CGH Analysis/MitoMet oligo aCGH, blood
- 3700 Mitochondrial DNA content (qPCR) Analysis, skeletal/muscle
- 3200 Electron Transport Chain Analysis, skeletal/muscle
- 2055 Comprehensive Mitochondrial DNA Analysis by massively parallel sequencing, (Mito NGS)

2450 Holcombe Boulevard, Suite 0-100, Houston, Texas 77030
713-798-6555 • 1-800-411-gene (4363) • www.BMGL.com

3/2015