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.


<table>
<thead>
<tr>
<th>Test codes</th>
<th>8100 for males, 8110 for females</th>
</tr>
</thead>
</table>

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

**SPECIMEN REQUIREMENTS**

**Test Requisition:** Autism Testing

**Test Requisition:** Autism Testing

<table>
<thead>
<tr>
<th>Test Requisition:</th>
<th>Autism Testing</th>
</tr>
</thead>
</table>

**Shipping and Handling:**

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
Genetic Testing for Autism Spectrum Disorders

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)