Autism Spectrum Disorders: Tier 2 Deletion/Duplication Panel

**Test Code:** MD021  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81161 x1, 81323 x1, 81403 x1, 81404 x1, 81405 x1, 81406 x1

**Condition Description**

Genetics of Autism Spectrum Disorders

Autism spectrum disorders (ASDs) are a group of neurodevelopmental disorders which include autism, pervasive developmental delay—not otherwise specified (PDD-NOS), and Asperger syndrome. ASDs are characterized by impairments in social relationships, variable degrees of language and communication deficiencies, and repetitive behaviors and/or a narrow range of interests. The age of onset is prior to age 3 with a variable clinical presentation, ranging in severity both amongst individuals as well as amongst the various subtypes of ASDs. Additional clinical features may also be observed in individuals with an ASD, such as intellectual disability (up to ~50%) and seizures (~25%).

Known genetic causes of autism include cytogenetically visible chromosome abnormalities (3-5%), copy number variants – which include submicroscopic deletions and duplications (~6-7%), and single gene disorders (~5%).

EGL Genetics’s integrated testing strategy allows for a comprehensive cytogenetics, metabolic, and molecular analysis of ASD in your patient. For a summary of autism testing at EGL, please click [here](#).

*Please note that some genes on this panel are associated with additional phenotypes.*

**References:**


**Genes**

ADSL, AFF2, APIS2, ARX, ATRX, BCKDK, BRAF, CACNA1C, CASK, CDH8, CDKL5, CHD7, CNTNAP2, CREBBP, DHCR7, DMD, EHMT1, EGD1, FMR1, FOXL1, FOXG1, FOXP1, FOXP2, HPRT1, KDM5C, L1CAM, MAGEL2, MBDS, MECP2, MED12, MEF2C, MID1, NHS, NIPBL, NLGN3, NLGN4X, NR1I3, NRXN1, NSD1, OPHN1, PAFAH1B1, PCDH19, PHF6, PINK1, POCBP1, PTCHD1, PSEN1, PTEN, RAB39B, RA1, RELN, SCN1A, SLC2A1, SLC9A6, SMARCB1, SMC1A, TCF4, UBE2A, UBE3A, VPS13B, ZEB2

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of autism or an autism spectrum disorder.
- Carrier testing in adults with a family history of autism or an autism spectrum disorder.

**Methodology**

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

**Detection**

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient’s clinical and/or biochemical phenotype.

**Specimen Requirements**

**Type:** Whole Blood (EDTA)

**Specimen Requirements:**

- EDTA (Purple Top)
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml
- Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

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**Type:** DNA, Isolated

**Specimen Requirements:**
- Microtainer
- 3 µg

Isolation using the Perkin Elmer™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**
- Autism Spectrum Disorders Panel: Tier 1 Cytogenetics and Molecular.
- Autism Spectrum Disorders Panel: Tier 1 Biochemical.

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