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 deficits, 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%).

Emory Genetics Laboratory’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, AP1S2, ARX, ATRX, BCKDK, BRAF, CACNA1C, CASK, CDH8, CDKL5, CD7, CNTNAP2, CREBBP, DHCR7, DMD, EHMT1, FGD1, FMR1, FOLR1, FOXP1, FOXP2, HPRT1, KDM5C, L1CAM, MAGEL2, MBDS, MECP2, MED12, MEF2C, MID1, NHS, NIPBL, NLGN3, NLGN4X, NR1I3, NRXN1, NSD1, OPHN1, PAFAH1B1, PCDH19, PHF6, PNKP, POGP1, PTCHD1, PTPN11, RAB39B, RAI1, RELN, SCN1A, SLCA21, SLCA9A6, 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.

Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

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
Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of 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.