Autism and Intellectual Disability Comprehensive Panel

Autism spectrum disorder (ASD) and intellectual disability (ID) represent a neurodevelopmental continuum with significant comorbidity and overlapping etiologies. ASD is classified by varying degrees of social impairment, communication limitations, repetitive behaviors, and/or restricted interests. Symptoms typically present by age 3. ID is classified by broad impairment in cognitive and adaptive functioning, typically with an IQ below 70, and presents before age 18. A global developmental delay (DD) diagnosis often precedes a diagnosis of ID, as cognitive skill or IQ cannot be reliably assessed prior to age 6. Those with severe DD diagnosed before age 6 are most likely to develop ID.

ASD and IDs are often comorbid and may be a feature in various genetic syndromes associated with chromosomal copy number variants (CNVs), fragile X syndrome, and inborn errors of metabolism (IEMs). Literature suggests ~10-15% of individuals with ASD and/or ID have detectable chromosomal abnormalities, CNVs, and fragile X syndrome.

Disease Overview

Indications for Ordering (Testing)
- Comprehensive evaluation of an individual with ASD and/or ID (with or without comorbidities)
- Assist with decisions about treatment and management of an individual with ASD and/or ID
- Confirmation of a clinical diagnosis of ASD and/or ID

Heritability
- ASD: ~90%
- ID: ~50%

Prevalence
- ASD: ~1/68 (1/42 males, 1/189 females)
- ID: ~1/100
- ASD and ID combined incidence: ~1/250

Test Limitations

Autism and intellectual disability panel testing limitations:
- Other etiologies of ASD and ID, such as single gene disorders, may not be identified
- Uninformative results may occur due to variants of uncertain clinical significance
- Results may not predict disorder severity

Cytogenomic SNP microarray limitations:
- The genome-wide resolution is approximately 25-50 kb for copy number changes and approximately 3 Mb for regions of homozygosity
- Testing does not detect
  - CNVs below the limit of resolution of this platform
  - Sequence-level variants (mutations) including point mutations and indels
  - Low-level mosaicism (generally, less than 20-30%)
  - Balanced chromosomal rearrangements (translocations, inversions, and insertions)
  - Genomic imbalance in repetitive DNA regions (centromeres, telomeres, segmental duplications, and acrocentric chromosome short arms)
  - Mitochondrial DNA alterations
- For further review, see Additional Technical Information for Cytogenomic SNP Microarray 2003414

Featured ARUP Testing

Autism and Intellectual Disability Comprehensive Panel 2014314
- Comprehensive genetic evaluation of individuals with ASD and/or ID
- Includes cytogenomic microarray, fragile X methylation analysis, and comprehensive metabolic disorders analysis

Test Components

May be ordered separately

Cytogenomic SNP Microarray 2003414
Method: Genomic Microarray (Oligo-SNP Array)
- Preferred first-tier test for DD, ID, ASD, and multiple anomalies
- Testing is performed on peripheral blood
- CytoScan HD platform (Thermo Fisher Scientific) to detect copy number changes, including deletions and duplications as well as copy-neutral regions of homozygosity

Cytogenomic SNP Microarray Buccal Swab 2006267
Method: Genomic Microarray (Oligo-SNP Array)
- Preferred first-tier test for DD, ID, ASD, and multiple anomalies
- Testing is performed on buccal sample

Fragile X (FMR1) with Reflex to Methylation Analysis 2009033
Method: Polymerase Chain Reaction/Capillary Electrophoresis
- Preferred test to diagnose fragile X syndrome and carrier screening in individuals with a positive family history
- If fragile X testing detects a CGG repeat of 100 or greater by polymerase chain reaction and capillary electrophoresis, methylation analysis will be added

Autism and Intellectual Disability Metabolic Panel 2014312
- Comprehensive genetic evaluation of individuals with ASD and/or ID
- Includes cytogenomic microarray, fragile X methylation analysis, and comprehensive metabolic disorders analysis
Evaluate for ASD, DD, and/or ID in individuals who have had negative FMR1 (fragile X) and cytogenomic SNP microarray testing

Fragile X testing limitations:

- Estimated size is not provided for full mutations with >200 repeats
- Rare mutations in FMR1 unrelated to trinucleotide expansion will not be detected
- Diagnostic errors can occur due to rare sequence variations
- For further review, see Additional Technical Information for Fragile X (FMR1) with Reflex to Methylation Analysis 2009033

Additional Resources


Related Information

**Alpha-Iduronidase Enzyme Activity - Mucopolysaccharidosis Type I**

**Creatine Disorders Panel Testing**

**Laboratory Testing for Developmental Delay, Intellectual Disability, and Autism Spectrum Disorder**

**Testing for Genetic Syndromes Related to Developmental Delay (DD), Intellectual Disability (ID), and Autism Spectrum Disorder (ASD)**

© 2023 ARUP Laboratories. All Rights Reserved. Client Services - (800) 522-2787