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

Laboratory testing should be considered as part of a comprehensive evaluation for individuals with suspected autism spectrum disorder and/or developmental delay/intellectual disability.

ARUP Laboratories provides high-quality, accurate results to assist you with decisions about treatment and management. Get the answers you need for your patients and their families.

COMBINED MICROARRAY, FRAGILE X, AND BIOCHEMICAL TESTING PANEL

Genetic and biochemical evaluation to support the clinical diagnosis of autism spectrum disorder and/or intellectual disability and to aid in treatment decision-making

- 2014314 Autism and Intellectual Disability Comprehensive Panel
- Includes recommended first-tier testing for autism spectrum disorder and intellectual disability (genomic microarray, fragile X testing with reflex to methylation analysis, and metabolic disorder testing)









CYTOGENOMIC MICROARRAY

Preferred first-tier test for developmental delay, intellectual disability, and autism spectrum disorder

2003414	Cytogenomic SNP Microarray
2006267	Cytogenomic SNP Microarray Buccal Swab

• Detects copy number changes (deletions and duplications) and copy-neutral regions of homozygosity

GENOME AND EXOME SEQUENCING

Consider as a first- or second-tier test in the evaluation of developmental delay and intellectual disability

- 3016583 Exome Sequencing
- 3016493 Whole Genome Sequencing

OTHER TESTS

2009033 Fragile X (*FMR1*) with Reflex to Methylation Analysis

• Reflexes to methylation analysis if a CGG repeat of 100 or greater is detected by polymerase chain reaction and capillary electrophoresis

2014312 Autism and Intellectual Disability Metabolic Panel

• Includes testing for acylcarnitine, mucopolysaccharides, organic acids, creatine disorders, and amino acids