aruplab.com/
testing/autism

References

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2. Centers for Disease Control. Autism Spectrum Disorder (ASD). **https://www.cdc.gov/ncbddd/autism/data/index.html**

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keyword: AUTISM

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Autism and Intellectual Disability

testing at ARUP Laboratories

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There is no one cause of autism, just as there is no one type of autism. Over the last five years, scientists have identified a number of rare genetic changes or mutations associated with autism. **Research has identified more than 100 autism risk genes.** In approximately 15% of cases, a specific genetic cause of autism can be identified. However, most cases involve a complex and variable combination of genetic risk and environmental factors that influence early brain development. In other words, in the presence of a genetic predisposition to autism, a number of nongenetic or environmental influences further increase a child's risk.[1]

Importance of Testing

The earlier a child is diagnosed, the earlier he/she can begin participating in behavioral intervention. Early intervention helps improve learning, communication, and social skills, which can give these children a chance at a better life.

Laboratory Testing

ARUP offers three primary tests to make ordering testing for autism and intellectual disabilities easier and more intuitive.

Autism and Intellectual Disability Metabolic Panel (2014312)
Preferred first-tier test to determine if a patient's symptoms of autism or intellectual disability are of a metabolic nature.

Autism and Intellectual Disability Comprehensive Panel (2014314)
Preferred comprehensive test to determine the etiology of autism or intellectual disability in a patient.

Exome Sequencing, Trio (2006332)
This test may shorten the diagnostic odyssey when other testing does not explain a patient's symptoms.

ARUP has a well-rounded menu for autism and intellectual disabilities. The following assays are the individual testing components for a more targeted assessment:

Amino Acids Quantitative by LC-MS/MS, Plasma (2009389)
Diagnose and monitor aminoacidopathies (e.g., PKU, MSUD).

Acylcarnitine Quantitative Profile, Plasma (0040033)
Diagnose and monitor fatty acid oxidation disorders and organic acidemias. Use in conjunction with urine organic acids and acylglycines testing.

Creatine Disorders Panel, Serum or Plasma (2002328)
Initial test to diagnose or rule out creatine deficiency syndromes following clinical presentation. Order Creatine Disorders Panel, Urine (2002333) simultaneously for proper result interpretation.

Creatine Disorders Panel, Urine (2002333)
Initial test to diagnose or rule out creatine deficiency syndromes following clinical presentation. Order Creatine Disorders Panel, Serum or Plasma (2002328) simultaneously for proper result interpretation.

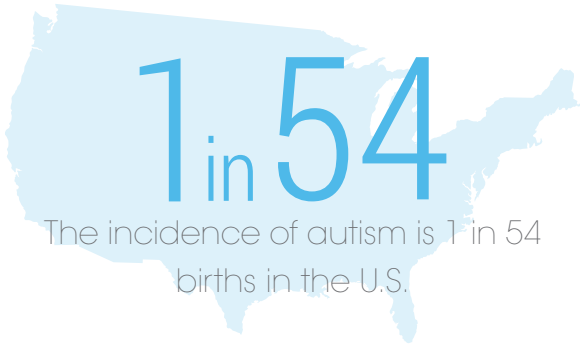
Organic Acids, Urine (0098389)
Diagnostic evaluation of patients with possible organic acidemias, fatty acid oxidation disorders, and other conditions.

Mucopolysaccharides Screen - Electrophoresis and Quantitation, Urine (0081352)
Use to evaluate symptomatic patients for mucopolysaccharidoses (MPS).

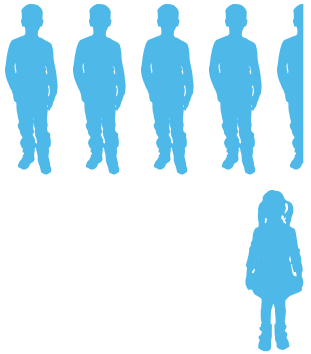
Cytogenomic SNP Microarray (2003414)
Preferred first-tier test for developmental delay, multiple anomalies, and autism-spectrum disorders. Testing is performed on peripheral blood.

Fragile X (*FMR1*) with Reflex to Methylation Analysis (2009033)
Preferred test to diagnose fragile X syndrome and carrier screening in individuals with a positive family history.

For additional information, educational opportunities, and ARUP's complete autism and intellectual disabilities test menu, visit: **aruplab.com/topics/autism**.



The incidence of autism is 1 in 54 births in the U.S.



ASD is 4.3 times more common in males, with a prevalence of 29.7 per 1,000 than females, who have a rate of 6.9 per 1,000 children.[2]

About 1 in 6 children in the United States was diagnosed with a developmental disability from 2009 to 2017, ranging from mild disabilities, such as speech and language impairments, to serious developmental disabilities, such as intellectual disabilities, cerebral palsy, and autism.[2]