Whole Genome Sequencing

Connect Patients With Answers

ARUP LABORATORIES

ARUP Laboratories' robust whole genome sequencing (WGS) and rapid whole genome sequencing (rWGS) testing suite provides comprehensive genomic analysis that supports efficient and cost-effective diagnosis.

- **Supported by expertise**: Every genome sequencing case is reviewed by our highly experienced team of medical directors and clinical variant scientists to ensure accurate and relevant interpretation.
- **Correlated with clinical findings**: Our interpretation is correlated with the clinical picture to pinpoint relevant variants and provide clear answers.
- **Driven by efficiency**: Our competitive turnaround times mean prompt results.
- **Committed to patients**: ARUP offers reanalysis of WGS or rWGS data for any samples initially tested at ARUP.

ARUP Tests

Test Number	Test Name	Turnaround Time
3019943	Genome Sequencing	14–21 days
3019947	Rapid Genome Sequencing	5–7 days
3005939	Whole Genome Reanalysis	14–21 days

WGS and rWGS assays include copy number variants, mitochondrial variants, and *SMN1* deletions for spinal muscular atrophy.

Elevate patient care with ARUP.

aruplab.com/genome

Clinical Benefits of Whole Genome Sequencing

When used as a first-tier testing strategy, WGS is more likely to provide a precise diagnosis.

Increased diagnostic yield: WGS has demonstrated superior diagnostic yield over whole exome sequencing and targeted gene assays.[1]

Decreased overall cost: Using WGS as an initial, comprehensive testing strategy decreases the overall cost of care by reducing the need for sequential or follow-up testing and inpatient stays.[2,3]

Decreased time to diagnosis: WGS significantly reduces the time needed to reach a precise diagnosis by eliminating the need for sequential testing. A study to evaluate diagnostic yield and time to precise genetic diagnosis found that the average time to diagnosis decreased from 289 to 13 days when WGS or WES was used as first-line testing.[4]

Recommendations From Professional Organizations

- The American College of Medical Genetics and Genomics (ACMG) and the American Academy of Pediatrics (AAP) recommend whole exome and genome sequencing as first-tier approaches to investigate developmental delay and intellectual disability.[5]
- The National Society of Genetic Counselors (NSGC) recommends WGS for unexplained epilepsy in individuals of all ages.

1. ARUP Laboratories. Genomic sequencing: an evolving standard in molecular genetic diagnosis. Published Sep 2025.
2. Moore C, Arenchild M, Waldman B, et al. Rapid whole-genome sequencing as a first-line test is likely to significantly reduce the cost of acute care in a private payer system. *J Appl Lab Med.* 2025;10(4):833-842. https://pubmed.ncbi.nlm.nih.gov/40248916/
3. Dimmock D, Caylor S, Waldman B, et al. Project Baby Bear: rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care. *Am J Hum Genet.* 2021;108(7):1231-1238. https://pubmed.ncbi.nlm.nih.gov/34089648/
4. Keefe AC, Scott AA, Kruidenier L, et al. Implementation of first-line rapid genome sequencing in non-critical care pediatric wards. *J Pediatr.* Published online Jun 2025. https://pubmed.ncbi.nlm.nih.gov/40562302/
5. Rodan L, Stoler J, Chen E, et al. Genetic evaluation of the child with intellectual disability or global developmental delay: clinical report. *Pediatrics*. 2025;156(1):e2025072219. https://pubmed.ncbi.nlm.nih.gov/40545261/

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