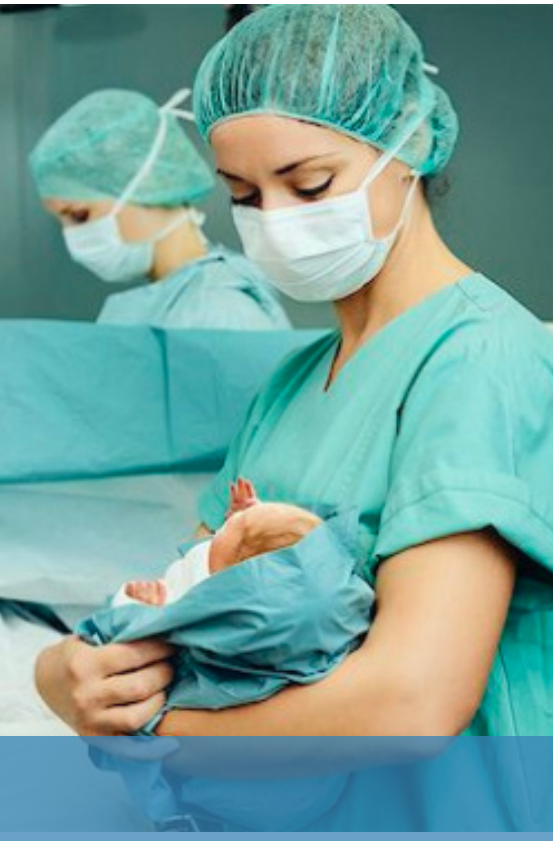
NewbornDx™

Advanced Sequencing Evaluation

When every minute counts...



Athena Diagnostics offers:

- A comprehensive test menu
- Access to a team of genetic counselors and other medical experts
- More than 30 years of neurology and genetics expertise
- Easy-to-read, comprehensive reports

AthenaDiagnostics.com
1-800-394-4493

Rapid results can bring earlier diagnosis and treatment for the most fragile patients

When every minute counts, the NewbornDx™ Advanced Sequencing Evaluation from Athena Diagnostics delivers rapid, 5-7 day results on a targeted 1,722 gene panel—helping to improve outcomes in critically ill newborns by expediting an accurate diagnosis and effective treatment.

The average NICU stay can cost $76,164 and typically lasts 13.2 days.[1] Shortening that stay by even one day can have significant economic and emotional benefits for families. With its extensive gene panel, the NewbornDx™ Advanced Sequencing Evaluation can help you diagnose a majority of the genetic causes found through an exome or genome panel in less than half the time, and for a fraction of the cost.[2]

NewbornDx™ can help improve outcomes for your patients by:

- A targeted, clinically actionable panel of 1,722 genes, specifically designed for conditions that are often present in NICU/PICU patients
- Rapid delivery of results, within 5-7 days, and verbal results available when report is signed off
- Patient-friendly, non-invasive blood spot collection
- Proband, duo, or trio testing options to decrease the number of variants of unknown significance and reduce the need for follow-up testing
- A mean coverage of 330X and 99.4% of bases covered at greater than 20X
- Whole blood, blood spot, or saliva options for parental samples

In comparison to published exome and genome studies, **NewbornDx™ includes 80%-100% of disease causing genes identified by these methods**—the highest diagnostic yield of all commercially available panels for NICU patients.[2-10]

athena diagnostics®

Athena Insight™: Finding Color in a World of Gray Results

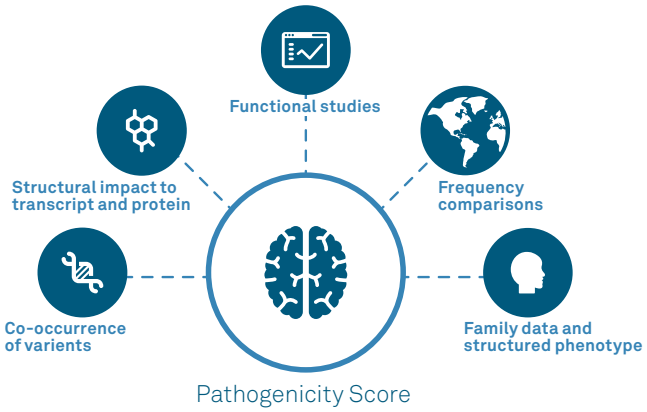
You hear them every day—the questions parents ask. What's causing my baby's symptoms? Why did this happen? What are the chances that I can pass this onto other children? The answers aren't always clear. When it comes to a genetic diagnosis, you want the most reliable, up-to-date, understandable information.

We understand your challenge. We understand your quest for insight. When a variant of unknown significance (VUS) is found, we know that the interpretation is not always straightforward. That's why we've developed Athena Insight™, a process for determining the significance of genetic variants.

Athena Insight™ is powered by a team of highly trained scientists whose primary focus is to assess the pathogenicity of genetic variants identified in patients. To achieve this goal, we conduct a thorough investigation of published research which is then integrated with data from internal and external databases, research collaborations, and clinician-provided phenotypes. A comprehensive and objective assessment of the data is made by utilizing a standardized, rules-based algorithm resulting in a final pathogenicity assessment score. Close collaboration among our scientists, genetic counselors, and clinical laboratory directors ensures that all the relevant information has been collected and systematically analyzed in order to provide the most clinically-informative result.

We understand your goals, and we work with you. Our high-touch approach optimizes the value of modern science and human insight. We look for color in a world of gray results.

For more information on the variant assessment process, please visit AthenaDiagnostics.com/AthenaInsight



Test Specifications

Test Code	Test Name	CPT Codes	Preferred Specimen	Sample Specifications	Turnaround Time	Specimen Stability
2052	NewbornDx™ Advanced Sequencing Evaluation, Proband	81443	Dried blood spot (DBS) card, Whole Blood in lavender-top (EDTA) tubes	One DBS card with 5 spots (2 spot min) 2 mL whole blood (1 mL min)	5–7 days	**Dried Blood Spot:** Room Temperature: 2 days Refrigerated: 2 years Frozen: Unacceptable **Whole Blood:** Room Temperature: 10 days Refrigerated: 10 years Frozen: Unacceptable
2053	NewbornDx™ Advanced Sequencing Evaluation, Family Member	Not Applicable	Dried blood spot (DBS) card, Whole Blood in lavender-top (EDTA) tubes, Saliva	One DBS card with 5 spots (2 spot min) 2 mL whole blood (1 mL min) One Oragene OGD 500 kit	Not Applicable	**Dried Blood Spot:** Room Temperature: 2 days Refrigerated: 2 years Frozen: Unacceptable **Whole Blood:** Room Temperature: 10 days Refrigerated: 10 years Frozen: Unacceptable **Saliva:** Room Temperature: 1 year Refrigerated: Unacceptable Frozen: Unacceptable

The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party.
Please contact one of our genetic counselors regarding specific acceptance policies and specimen requirements for prenatal testing at 800-394-4493.

Athena Diagnostics offers a comprehensive genetic test menu.
Customers in the US and Canada please call toll free **1.800.394.4493**
or visit us online at **AthenaDiagnostics.com/NewbornDx**

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References
1. March of Dimes. National Perinatal Information Center: Quality Analytic Services. Special Care Nursey Admissions. https://www.marchofdimes.org/peristats/pdfdocs/nicu_summary_final.pdf. Accessed April 18, 2019. 2. Meng L, Pammi M, Saronwala A, et al. Use of exome sequencing for infants in intensive care units: ascertainment of severe single-gene disorders and effect on medical management. *JAMA Pediatr.* 2017;171(12):e173438. doi: 10.1001/jamapediatrics.2017.3438 3. Petrikin JE, Cakici JA, Clark MM, et al. The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. *NPJ Genom Med.* 2018;3:6. doi: 10.1038/s41525-018-0045-8 4. Quinlan-Jones E, Lord J, Williams D, et al. Molecular autopsy by trio exome sequencing (ES) and postmortem examination in fetuses and neonates with prenatally identified structural anomalies. *Genet Med.* 2018. doi: 10.1038/s41436-018-0298-8 5. Soden SE, Saunders CJ, Willig LK, et al. Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. *Sci Transl Med.* 2014;6(265):265ra168. doi: 10.1126/ scitranslmed.3010076 6. Stark Z, Tan TY, Chong B, et al. A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. *Genet Med.* 2016;18(11):1090-1096. doi: 10.1038/gim.2016.1 7. Trujillano D, Bertoli-Avella AM, Kumar Kandaswamy K, et al. Clinical exome sequencing: results from 2819 samples reflecting 1000 families. *Eur J Hum Genet.* 2017;25(2):176-182. doi: 10.1038/ejhg.2016.146 8. Vissers LELM, van Nimwegen KJM, Schieving JH, et al. A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. *Genet Med.* 2017;19(9):1055-1063. doi: 10.1038/gim.2017.1 9. Willig LK, Petrikin JE, Smith LD, et al. Whole-genome sequencing for identification of Mendelian disorders in critically ill infants: a retrospective analysis of diagnostic and clinical findings. *Lancet Respir Med.* 2015; 3(5):377–387. doi: 10.1016/S2213-2600(15)00139-3 10. Yang Y, Muzny DM, Xia F, et al. Molecular findings among patients referred for clinical whole-exome sequencing. *JAMA.* 2014;312(18):1870-1879. doi: 10.1001/jama.2014.14601

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