



GeneDx Announces American Academy of Pediatrics Recommends Exome and Genome Sequencing as First-Tier Tests for Children with Global Developmental Delay or Intellectual Disability

June 23, 2025

-- A milestone in pediatric medicine, general pediatricians are now expected to play a more active role in initiating genetic testing

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-- Earlier use of exome and genome testing delivers better clinical outcomes and reduces costs across the U.S. healthcare system¹ --

-- GeneDx will lead education and adoption efforts to end the diagnostic odyssey --

GAITHERSBURG, Md.--(BUSINESS WIRE)--Jun. 23, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, announces the American Academy of Pediatrics (AAP) issued [updated guidance](#) to now recommend exome and genome sequencing as first-tier tests for children with global developmental delay (GDD) or intellectual disability (ID) in most circumstances because of superior diagnostic yield and higher cost-effectiveness if pursued earlier in the diagnostic process². For more than 60,000 pediatricians, this milestone marks a turning point in pediatric healthcare, ushering in earlier diagnosis, interventions, and improved outcomes for millions of children.

"The AAP's updated guidance is a long-overdue validation of what many of us have known – earlier access to genomic testing leads to faster, more accurate diagnoses and better outcomes – and represents an important milestone in bringing genomics to the front line of pediatric medicine as we seek to protect children from unnecessary disease progression," said Katherine Stueland, President and CEO of GeneDx. "Exome and genome testing are no longer tests of last resort; they are now the first step, and there is no one better equipped than GeneDx to support pediatricians through this next chapter. We're ready to slash the average five-year diagnostic odyssey to a fraction of that time, drive down healthcare costs, and, most importantly, change children's lives sooner. Having driven innovation and advanced patient care through genomic testing, working in partnership with geneticists and pediatric specialists, GeneDx is best positioned to empower general pediatricians to take a leading role in early diagnosis and precision medicine."

The new diagnostic approach underscores the growing recognition that pediatricians are the front line of genetic medicine and marks a major advancement by equipping these clinicians with actionable genomic insights to accelerate targeted treatments and end the burdensome diagnostic waiting period faced by too many families today. Previous guidance from AAP³, published in 2014, recommended the use of narrow testing such as Chromosomal Microarray Analysis (CMA) and FMR1, both of which can be considered antiquated recommendations for these patient populations considering the advancements made with exome and genome testing, including the superior diagnostic yield, cost-effectiveness and speed to diagnosis.

GeneDx is the leader in advancing genomic technologies, providing clinicians and patients with access to comprehensive exome and genome testing. With unparalleled expertise from sequencing more than 800,000 exomes and genomes – alongside millions of phenotypic data points – GeneDx has assembled one of the largest, rare disease datasets. GeneDx's standard exome and genome results are available in as little as three weeks, and insurance coverage for this testing continues to expand across the country, supported by coverage from major commercial payors and Medicaid programs.

"Given general pediatricians depend on clinical guidance to provide the best possible care for our patients, this updated report will empower pediatricians to offer families the most current, evidence-based genetic testing — helping to shorten the often long and difficult diagnostic journey," said Dr. Deborah Ondrasik, General Pediatrician, Southcoast Health. "I'm thrilled to see the AAP update these recommendations to reflect just how far genetics have come in the past decade. This is a meaningful step forward for our patients and their families."

With its recently launched brand campaign, [Diagnosis is Power](#), GeneDx is educating clinicians and parents on the need for earlier genetic testing to accelerate answers and improve outcomes for more patients and families. Core to its mission, [GeneDx believes](#) in the power of diagnosis and envisions a world where any genetic disorder is diagnosed quickly to prevent disease progression and ensure long and healthy lives for all.

About GeneDx

At GeneDx (Nasdaq: WGS), we believe that everyone deserves personalized, targeted medical care—and that it all begins with a genetic diagnosis. Fueled by one of the world's largest, rare disease data sets, our industry-leading exome and genome tests translate complex genomic data into clinical answers that unlock personalized health plans, accelerate drug discovery, and improve health system efficiencies. For more information, please visit [genedx.com](https://www.genedx.com) and connect with us on [LinkedIn](#), [Facebook](#), and [Instagram](#).

Forward Looking Statements

This press release may contain “forward-looking statements” within the meaning of Section 21E of the Securities Exchange Act of 1934, as amended, and the U.S. Private Securities Litigation Reform Act of 1995. These forward-looking statements generally are identified by the words “believe,” “project,” “expect,” “anticipate,” “estimate,” “intend,” “strategy,” “future,” “opportunity,” “plan,” “may,” “should,” “will,” “would,” “will be,” “will continue,” “will likely result,” and similar expressions. Forward-looking statements are predictions, projections and other statements about future events that are based on current expectations and assumptions and, as a result, are subject to risks and uncertainties. Many factors could cause actual future events to differ materially from the forward-looking statements in this press release, including but not limited to: (i) our ability to implement business combinations, plans, goals and forecasts, and identify and realize additional opportunities, (ii) the risk of downturns and a changing regulatory landscape in the highly competitive healthcare industry, (iii) the size and growth of the market in which we operate, (iv) our ability to pursue our new strategic direction, and (v) our ability to enhance our artificial intelligence tools that we use in our clinical interpretation platform. The foregoing list of factors is not exhaustive. A further list and description of risks, uncertainties and other matters can be found in the “Risk Factors” section of our Annual Report on Form 10-K for the fiscal year ended December 31, 2024, and other documents filed by us from time to time with the SEC. These filings identify and address other important risks and uncertainties that could cause actual events and results to differ materially from those contained in the forward-looking statements. Forward-looking statements speak only as of the date they are made. Readers are cautioned not to put undue reliance on forward-looking statements, and we assume no obligation and do not intend to update or revise these forward-looking statements, whether as a result of new information, future events, or otherwise. We do not give any assurance that we will achieve our expectations.

1. Lavelle TA, Feng X, Keisler M, et al. Cost-effectiveness of exome and genome sequencing for children with rare and undiagnosed conditions. *Genet Med.* 2022;24(6):1349-1361. doi: 10.1016/j.gim.2022.03.005.
2. Rodan LH, et al. Genetic Evaluation of the Child With Intellectual Disability or Global Developmental Delay: Clinical Report. *Pediatrics.* 2025 Jun 23:e2025072219. doi: 10.1542/peds.2025-072219. Epub ahead of print.
3. Moeschler JB, Shevell M; Committee on Genetics. Comprehensive evaluation of the child with intellectual disability or global developmental delays. *Pediatrics.* 2014 Sep;134(3):e903-18. doi: 10.1542/peds.2014-1839.

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