



GeneDx to Launch Autism Partnership Program to Expand Access to Exome and Genome Testing, including Dedicated SHANK3 Program

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Jaguar Gene Therapy joins as founding partner to initiate a genetic testing program for SHANK3-related autism spectrum disorder and Phelan-McDermid syndrome

GAITHERSBURG, Md.--(BUSINESS WIRE)--Oct. 9, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced the Autism Partnership Program to expand access to its industry leading exome and genome testing – the only autism testing program committed to delivering answers to families in weeks. Jaguar Gene Therapy joins the program as the founding partner, initiating a genetic testing program for individuals clinically suspected of having SHANK3-related autism spectrum disorder (ASD) and Phelan-McDermid syndrome. A single pathogenic variant or deletion in the SHANK3 gene is a leading single-gene cause of ASD.

By uniting healthcare providers, advocacy groups, biopharmaceutical companies and payers, the Autism Partnership Program aims to eliminate barriers to testing and deliver clinically actionable insights earlier in the diagnostic process. Although clinical guidelines strongly endorse genetic testing for those with ASD, many families still encounter significant barriers, leading to delays in receiving an accurate genetic diagnosis, sometimes for years. The Autism Partnership Program is dedicated to driving more equitable care by increasing availability and easing financial burdens. As part of the program, individuals with moderate to severe development delay, intellectual disability, ASD, or autistic-like behavior with clinical suspicion of Phelan-McDermid syndrome will have access to exome and genome sequencing from GeneDx, with costs covered by Jaguar Gene Therapy when patients have Medicare / Medicare Advantage and when commercial insurance coverage is unavailable. The genomic data generated through the SHANK3 program will help advance Jaguar's mission to accelerate the development of treatments and therapies for patients living with severe genetic disorders.

"Behind every autism diagnosis is a family searching for answers. Genetic testing can illuminate the 'why' - transforming confusion into clarity and opening doors to the right care," said Lisa Gurry, Chief Business Officer of GeneDx. "Far too many families still wait too long or never gain access at all to the answers that can change lives. Our Autism Partnership Program is designed to change that - helping reduce inequities and deliver insights that can improve outcomes. The collaboration with Jaguar Gene Therapy is a powerful example of how we're working with biopharma partners to expand access and translate the answers we uncover into tomorrow's breakthroughs."

Studies demonstrate that 16% of children with autism have an identifiable genetic diagnosis – with over 800 genes associated with autism – and clinical guidelines recommend genetic testing, including exome and genome sequencing, as part of the diagnostic workup.^{1, 2} Yet despite these recommendations, access to testing remains uneven especially for underserved populations. By expanding access to exome and genome sequencing, families will have answers sooner. Additionally, insights can be generated that help researchers and biopharma partners better understand the genetic underpinnings of autism, paving the way for more targeted therapies.

A single mutation or deletion in SHANK3 is the underlying cause of Phelan-McDermid syndrome (also known as 22q13.3 deletion syndrome), a rare genetic disorder with an estimated prevalence of 1 in 10,000.^{3,4,5} Genetic sequencing studies have revealed that SHANK3 pathogenic variants may be present in approximately 0.5%-0.69% of patients with ASD, equating to around 46,000 patients in the United States, including approximately 10,000 pediatric patients under the age of 18. Despite this prevalence, clinical experts believe SHANK3-related diagnoses remain underreported due to limited access and low adoption of genetic testing in the diagnostic journey of ASD.^{6,7,8,9,10}

"We're proud to partner with GeneDx to expand access to exome and genome testing for individuals with clinical suspicion of SHANK3-related autism spectrum disorder and Phelan-McDermid syndrome with the aim of shortening the diagnostic journey," said Joe Nolan, CEO of Jaguar Gene Therapy. "The SHANK3 gene's role in autism underscores the urgent need to broaden the use of genomic sequencing and for those with Phelan-McDermid syndrome to enroll in natural history studies to better understand the condition and support the development of targeted therapies."

"For individuals with autism, genetic testing can be truly life-changing – providing answers that open doors to community, care guidelines, and emerging treatment options. Yet, far too many children with genetic conditions like Phelan-McDermid syndrome never receive a diagnosis simply because they lack access to testing," said Geraldine Bliss, President of CureSHANK. "The Autism Partnership Program is an important step toward reducing disparities in access and ensuring more equitable opportunities for families to benefit from precision medicine. We're deeply grateful to Jaguar Gene Therapy and GeneDx for their leadership in establishing the first sponsored genetic testing program for autism."

To be eligible for the Autism Partnership Program patients must meet certain criteria, including the following:

- Patient must reside in the United States;
- Patient's ordering provider must be authorized under applicable law to order genetic testing in the United States;
- Patient must present with moderate to severe development delay, intellectual disability, ASD, or autistic like behavior with clinical suspicion of Phelan-McDermid syndrome;
- Additional eligibility criteria apply.

To learn more about the Autism Partnership Program, including additional opportunities for biopharmaceutical companies, contact us at genedx.com/biopharma.

About GeneDx

GeneDx (Nasdaq: WGS) is the global leader in rare disease diagnosis, transforming the way medicine is practiced by making genomics the starting point for health, not the last resort. We bring together unmatched clinical expertise, advanced technology, and the power of GeneDx Infinity™ – the largest rare disease dataset – built over 25 years from millions of genomic tests and deep clinical insights. This unparalleled foundation powers our ExomeDx™ and GenomeDx™ tests, giving clinicians the highest likelihood of delivering a timely, accurate diagnosis. GeneDx is shaping the future of healthcare by moving the standard of care from sick care to proactive healthcare. While our roots are in rare disease diagnosis, our commitment extends beyond – growing with the families we serve – as a trusted partner at every stage of life. For more information, visit genedx.com and connect with us on [LinkedIn](#), [Facebook](#), and [Instagram](#).

About Jaguar Gene Therapy

Jaguar Gene Therapy, LLC is a clinical-stage biotechnology company dedicated to accelerating breakthroughs in gene therapy for patients suffering from severe genetic diseases including those that impact sizeable patient populations. The company is made up of a proven team of experts who have first-hand experience in bringing novel gene therapy treatments to patients and their families. Jaguar is rapidly advancing an initial pipeline of three programs. The company's lead program targets severe neurodevelopmental disorders caused by *SHANK3* haploinsufficiency, due to loss of function pathogenic variant or deletions in *SHANK3* including a genetic form of autism spectrum disorder and Phelan-McDermid syndrome. A clinical trial in pediatric patients is currently underway. The second pipeline program targets Type 1 galactosemia and the third targets Type 1 diabetes. Jaguar's key investors include Deerfield Management Company, ARCH Venture Partners and Eli Lilly and Company. For more information, please visit www.jaguargenetherapy.com and follow Jaguar Gene Therapy on [LinkedIn](#).

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 plans to accelerate and unlock the full potential of precision medicine, (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. 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 our Quarterly Reports on Form 10-Q for the fiscal quarters ended March 31, 2025 and June 30, 2025, 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.

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