



GeneDx Expands Commercial Footprint for Exome and Genome Testing with Inborn Errors of Immunity as a New Indication

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Focus on Inborn Errors of Immunity expands access to exome and genome testing for the known 200,000+ patients in the US¹ impacted by these conditions and opens new opportunities for GeneDx and biopharma partners to accelerate the path to treatment for patients

GAITHERSBURG, Md.--(BUSINESS WIRE)--Apr. 3, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced the commercial expansion into Inborn Errors of Immunity (IEI), a group of nearly 500 genetic disorders that impair immune function², increasing susceptibility to infections, autoimmunity, and inflammatory conditions. The strategic expansion reinforces GeneDx's mission to improve patient outcomes by providing exome and genome testing solutions for an ever-growing number of patients, now including those with inherited immunological conditions.

With an expanded commercial focus on IEIs, GeneDx accelerates patient care with the adoption of exome and genome testing, empowering ordering clinicians with more accurate and comprehensive genetic insights to better treat this patient population. Today, clinicians treating IEIs recognize the importance of genetic testing, and with the rapid pace of new gene-disease discovery in IEI¹, exome and genome sequencing are better equipped than panel-based testing to stay up to date with new discoveries in this area. Genomic sequencing provides a diagnostic yield of approximately 40% for IEI patients^{3,4,5} — higher than the 29% yield from multi-gene panels.³

With superior diagnostic accuracy and growing reimbursement support, the shift toward an exome and genome-first approach accelerates times to an accurate diagnosis and informs personalized treatment decisions such as bone marrow transplantation, gene therapy, biologic supportive therapy, and Ig replacement therapy. Therapies are available for more than 50% of individuals with an IEI.⁶

"GeneDx's commercial focus on Inborn Errors of Immunity furthers our commitment to providing patients, families and clinicians with the most comprehensive genetic testing solutions at the times they need it most," said Britt Johnson, PhD, FACMG, and Senior Vice President of Medical Affairs at GeneDx. "By transitioning from panel-based testing to exome and genome sequencing, GeneDx will improve diagnostic precision, enabling earlier interventions, and ultimately enhancing patient outcomes by accelerating the path to treatment."

In the past 18 months, GeneDx has sequenced more than 5,000 patients suspected of having IEIs. This commercial and patient focus not only empowers providers with comprehensive genetic insights to deliver optimal patient care but also opens new opportunities for biopharma partners to advance research, drug discovery, and therapeutic development by leveraging GeneDx's industry-leading dataset of more than 750,000 clinical exomes and genomes already enriched with IEI data.

"Inborn Errors of Immunity can be difficult to diagnose based on clinical features alone due to complex and overlapping phenotypes. Comprehensive exome and genome sequencing enable precise diagnoses and allow patients to access clinical trials and targeted therapies that may improve their quality of life and clinical outcomes," said Heather McLaughlin, PhD, FACMG and Senior Director, Molecular Diagnostics at Pharming.

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 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 (vi) 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.

References:

1. Rider, NL, Truxton A, Ohrt T, et al. Validating inborn error of immunity prevalence and risk with nationally representative electronic health record data. *J Allergy Clin Immunol.* 2024 Jun;153(6):1704-1710. doi: 10.1016/j.jaci.2024.01.011. Epub 2024 Jan 24.
2. Tangye SG, Al-Herz W, Bousfiha A, et al. Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. *J Clin Immunol.* 2022 Oct;42(7):1473-1507. doi: 10.1007/s10875-022-01289-3
3. Vorsteveld EE, Hoischen A, van der Made CI. Next-Generation Sequencing in the Field of Primary Immunodeficiencies: Current Yield, Challenges, and Future Perspectives. *Clin Rev Allergy Immunol.* 2021 Oct;61(2):212-225. doi: 10.1007/s12016-021-08838-5.
4. Chen Y, Li D, Yin J, et al. Diagnostic yield of next-generation sequencing in suspect primary immunodeficiencies diseases: a systematic review and meta-analysis. *Clin Exp Med.* Jun 18 2024;24(1):131. doi:10.1007/s10238-024-01392-2
5. Platt CD, Zaman F, Bainter W, et al. Efficacy and economics of targeted panel versus whole-exome sequencing in 878 patients with suspected primary immunodeficiency. *J Allergy Clin Immunol.* Feb 2021;147(2):723-726.
6. Quinn J, Modell V, Johnson B, Poll S, Aradhya S, Orange JS, Modell F. Global Expansion of Jeffrey's Insights: Jeffrey Modell Foundation's Genetic Sequencing Program for Primary Immunodeficiency. *Front Immunol.* 2022 Jun 10;13:906540. doi: 10.3389/fimmu.2022.906540.

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Investor Relations Contact:

Investors@GeneDx.com

Media Contact:

Press@GeneDx.com

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