GeneDx and Mirum Pharmaceuticals Launch Sponsored Testing Programs to Help Patients Find Answers Through Comprehensive Genetic Testing

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Collaboration provides no-cost exome testing for eligible patients with unexplained cholestasis, early-onset cataracts, or suspected or confirmed PBD-ZSD

GAITHERSBURG, Md.--(BUSINESS WIRE)--Nov. 5, 2026-- GeneDx, the leader in rare disease diagnosis and improving health through the power of genomic data, today announced a collaboration with Mirum Pharmaceuticals to offer no-cost exome testing for eligible patients across three sponsored testing programs: cholestasis, early-onset cataracts, and peroxisomal biogenesis disorder–Zellweger spectrum disorders (PBD-ZSD). Across these programs and for patients that meet specific criteria, GeneDx and Mirum are addressing a shared challenge in rare disease care: helping identify the genetic cause of symptoms that may be unexplained, affect multiple organ systems, or overlap with other conditions. Together, the programs aim to uncover underlying genetic conditions to inform diagnosis, clinical management, and treatment decisions.

Identifying the underlying genetic cause can have important implications for patient care. Certain inherited liver diseases[1] can progress to liver failure, early-onset cataracts may signal genetic conditions affecting more than vision, and PBD-ZSD[2] can cause progressive neurological, hearing, vision, and liver complications. Together, these programs aim to help clinicians identify underlying genetic conditions earlier and inform the diagnostic journey, clinical management and treatment decisions.

“Reaching a genetic diagnosis can be an important turning point for patients and families living with rare disease,” said Peter Radovich, President and Chief Operating Officer of Mirum Pharmaceuticals. “GeneDx’s advanced testing and deep rare disease expertise complement Mirum’s commitment to supporting patients throughout their journey by providing information that may support clinical decision-making.”

Eligible patients will receive GeneDx’s ExomeDx™ testing, with proband, duo, or trio testing options available through the programs. Clinicians will receive a comprehensive clinical report with expert interpretation of the results, and GeneDx will also offer post-test genetic counseling to help patients and families understand their results and potential next steps.

The collaboration includes three programs with distinct eligibility criteria:

- **Cholestasis:** Intended for patients with unexplained cholestasis or suspected inherited cholestatic liver disease, including those with unexplained chronic liver disease, certain associated clinical findings, or a family history of genetically confirmed cholestasis. Relevant conditions may include suspected cholestatic liver diseases such as Alagille syndrome and progressive familial intrahepatic cholestasis (PFIC). Patients with suspected extrahepatic causes or total parenteral nutrition–associated cholestasis as the primary diagnosis are excluded.
- **Early-onset cataracts:** Eligible for patients ages 2 to 35 with current or previous idiopathic cataracts not attributable to infection, trauma, or other non-genetic causes.
- **PBD-ZSD:** Intended for patients with diagnosed or clinically suspected peroxisome biogenesis disorder-Zellweger spectrum disorders (PBD-ZSD), a group of rare inherited metabolic disorders caused by variants in *PEX* genes, including patients experiencing neurological, vision, hearing, or liver deterioration.

“Rare genetic conditions can affect multiple organ systems and look remarkably different from one patient to the next, making timely diagnosis especially challenging,” said Lisa Gurry, Chief Business Officer, GeneDx. “GeneDx helps clinicians and patients find earlier answers through advanced genetic testing, expert clinical interpretation, and the world's largest rare disease dataset. Our collaboration with Mirum builds on our commitment to breaking down barriers to genetic testing, accelerating diagnosis and empowering clinicians to make more informed care decisions for patients with rare diseases.”

Testing is fully sponsored by Mirum Pharmaceuticals, and no claim will be submitted to a patient’s insurance to reduce financial barriers to testing. Healthcare providers and families interested in learning more about eligibility and ordering can visit genedx.com/cholestasis, genedx.com/cataracts and genedx.com/pbd-zsd.

About GeneDx
GeneDx’s (Nasdaq: WGS) mission is to empower everyone to live their healthiest life through genomics. GeneDx combines unmatched clinical expertise, advanced technology, and the power of GeneDx Infinity™ – the world’s largest rare disease genomic dataset. This unparalleled foundation powers GeneDx’s ExomeDx™ and GenomeDx® tests – ranked #1 by expert geneticists and granted FDA Breakthrough Device designation – enabling clinicians to deliver precise, fast, and actionable diagnoses. GeneDx Infinity also fuels discovery for biopharma, with the most powerful AI-driven genomic intelligence. A genomics pioneer over the last 25 years, diagnosing more than 4,800 genetic diseases and publishing more than 1,000 research publications, GeneDx is building the network that will drive the future of genomic precision medicine. For more information, visit **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 advance gene-disease discovery, (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, and (iv) future expansion of insurance coverage for exome and genome testing. 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, 2025 and our Quarterly Report on Form 10-Q for the fiscal quarter ended March 31, 2026, filed with the SEC on May 4, 2026, our Quarterly Report on Form 10-Q for the fiscal quarter ended June 30, 2026, filed with the SEC on August 3, 2026 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. Amendola M, Squires JE. Pediatric Genetic Cholestatic Liver Disease Overview. GeneReviews®. Updated May 25, 2023. https://www.ncbi.nlm.nih.gov/books/NBK584020/

2. Steinberg SJ, Raymond GV, Braverman NE, Moser AB. Zellweger Spectrum Disorder. GeneReviews®. Updated October 29, 2020. https://www.ncbi.nlm.nih.gov/books/NBK1448/

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