



## **Behind The Seizure™ Program Expansion Helps Speed the Diagnosis of Genetic Epilepsy in Younger Children Experiencing Unprovoked Seizures**

*—Stoke Therapeutics and Xenon Pharmaceuticals Join Founders BioMarin and Invitae to Offer a No-Cost Epilepsy Gene Panel Testing Program for Children Up to 60 Months Who Have Had an Unprovoked Seizure—*

**SAN FRANCISCO, February 26, 2019** -- As Rare Disease Day focuses attention on the challenges of diagnosing rare, often genetic conditions, [BioMarin Pharmaceutical Inc.](#) (NASDAQ: BMRN), [Invitae Corporation](#) (NYSE: NVTA), [Stoke Therapeutics](#) and [Xenon Pharmaceuticals](#) (NASDAQ: XENE) today announced the expansion of [Behind the Seizure](#), an innovative, cross-company collaboration that aims to provide faster diagnosis for young children with epilepsy. Previously available to patients ages 2-4 years, the program will expand eligibility to make no-cost genetic testing available for healthcare providers to order for children who have had an unprovoked seizure from birth up to age five (0 to 60 months).

“Genetic testing can be invaluable in helping families and healthcare providers better understand how to care for children living with epilepsy,” said Amy Brin Miller, executive director and chief executive officer of the Child Neurology Foundation. “We applaud this unique effort to bring multiple partners together to help increase access to genetic testing for more of their patients, including their youngest ones.”

Since the program began, hundreds of children have received genetic testing through Behind the Seizure and research has shown participants in the program were diagnosed 1-2 years sooner than historic averages.<sup>1</sup>

“Through this program we can secure comprehensive genetic testing for a child who has had an unprovoked seizure at no cost to the patient. That testing helps us get to a diagnosis faster so we can focus on providing the most accurate and timely treatment for our patients,” said Raman Sankar, M.D., professor and chief of pediatric neurology at UCLA Mattel Children's Hospital in Los Angeles. “When dealing with such young patients, this type of program can be very important, shortening the time to diagnosis, particularly for neurodegenerative conditions such as CLN2 disease.”

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<sup>1</sup>Miller, Nicole, et al, "Behind the Seizure: A No-Cost 125-gene Epilepsy Panel for Pediatric Seizure Onset Between 2-4 Years." Presented at the American Society of Human Genetics Meeting: October 16-20, 2018, San Diego, CA.

Partners in the program sponsor the cost of testing using the Invitae Epilepsy Panel, which includes up to 180+ genes associated with both syndromic and non-syndromic causes of epilepsy, including neurodegenerative conditions. With the expansion of the program, healthcare providers can now order the test for patients under the age of five with unprovoked seizures. Test results are available quickly (14 days on average).

“For many of the genetic disorders which cause serious debilitating morbidities in young children, early diagnosis is of paramount importance, and targeted early genetic testing is one way to overcome the challenges of delayed diagnosis. Over the years we’ve heard from physicians, advocacy groups, and parents about the challenges they face in implementing genetic testing early, to provide accurate and timely diagnosis that enables them to tailor their patients’ care,” said Adrian Quartel, group vice president and head of medical affairs for BioMarin. “We’re proud to be working with our partners to further expand the Behind the Seizure program and foster earlier genetic testing – especially for the youngest patients.”

“More than half of epilepsies are based in genetics. When a child experiences a seizure, performing genetic testing quickly using a comprehensive gene panel is a highly cost-effective and informative diagnostic tool. Quickly finding an underlying cause for infantile epilepsy can be crucial for making a difference for that patient with precision management and treatment options,” said Robert Nussbaum, M.D., chief medical officer of Invitae. “We’re proud to partner with BioMarin, Stoke and Xenon to help more children get access to genetic testing sooner.”

“At Stoke, we’re focused on developing therapies that address the root cause of genetic epilepsies, starting with Dravet syndrome. As precision medicines become available, it is increasingly important for patients to get early and accurate genetic testing so they can be matched with the appropriate treatment. That’s why we’re delighted to support this critical expansion of the Behind the Seizure program,” said Ed Kaye, M.D., chief executive officer of Stoke Therapeutics.

“Xenon is proud to be part of the Behind the Seizure collaboration, which aims to rapidly identify the genetic cause of seizures in children, including KCNQ2 and SCN8A epileptic encephalopathies. By offering this genetic testing to very young patients, it is hoped that the results will lead to earlier diagnoses and improved treatment outcomes through the introduction of personalized medicine approaches,” said Simon Pimstone, M.D., chief executive officer of Xenon Pharmaceuticals.

To learn more about the Behind the Seizure program please visit <https://www.invitae.com/en/behindtheseizure/>.

### **About BioMarin**

BioMarin is a global biotechnology company that develops and commercializes innovative therapies for patients with serious and life-threatening rare and ultra-rare genetic diseases. The company's portfolio consists of seven commercialized products and multiple clinical and pre-clinical product candidates. For additional information, please visit [www.biomarin.com](http://www.biomarin.com).

### **About Invitae**

Invitae Corporation (NYSE: NVTX) is a leading genetics company whose mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people. Invitae's goal is to aggregate the world's genetic tests into a single service with higher quality, faster turnaround time, and lower prices. For more information, visit the company's website at [invitae.com](http://invitae.com).

### **About Stoke Therapeutics**

Launched in 2018, Stoke Therapeutics is a biotechnology company working to increase gene expression to treat a wide array of severe genetic diseases, including genetic conditions affecting the central nervous system, eye, liver and kidney. Stoke's lead program addresses Dravet syndrome, one of the more common causes of genetic epilepsy. Stoke has raised \$130 million in funding from two rounds of financing; investors include Apple Tree Partners, RTW Investments, RA Capital Management, Cormorant Asset Management, Perceptive Advisors, funds managed by Janus Henderson Investors, Redmile Group, Sphera Funds Management, and Alexandria Venture Investments. For more information, visit [www.StokeTherapeutics.com](http://www.StokeTherapeutics.com) and follow Stoke on Twitter [@StokeTx](https://twitter.com/StokeTx).

### **About Xenon Pharmaceuticals**

Xenon Pharmaceuticals is a clinical stage biopharmaceutical company committed to developing innovative therapeutics to improve the lives of patients with neurological disorders, including rare central nervous system (CNS) conditions. We are advancing a novel product pipeline of neurology therapies to address areas of high unmet medical need, with a focus on epilepsy. For more information, please visit [www.xenon-pharma.com](http://www.xenon-pharma.com).

### **Safe Harbor Statements**

This press release contains forward-looking statements within the meaning of the Private Securities Litigation Reform Act of 1995, including statements relating to the benefits of genetic testing and the Behind the Seizure program, including shortening the time to diagnosis and improved treatment outcomes for patients. Forward-looking statements are subject to risks and uncertainties that could cause actual results to differ materially, and reported results should not be considered as an indication of future performance. These risks and uncertainties include, but are not limited to: the company's ability to use rapidly changing genetic data to interpret test results accurately and consistently; the ability of genetic testing to result in faster or more accurate diagnosis; laws and regulations applicable to the company's business; and the other risks set forth in Invitae's filings with the Securities and Exchange Commission, including the risks set forth in its Quarterly Report on Form 10-Q for the quarter ended September 30, 2018. These forward-looking statements speak only as of the date hereof, and Invitae Corporation disclaims any obligation to update these forward-looking statements.

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