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## **Rady Children's Institute for Genomic Medicine and Alexion Pharmaceuticals Partner to Speed the Diagnosis of Rare Genetic Disorders in Newborns**

SAN DIEGO & NEW HAVEN, Conn.--(BUSINESS WIRE)-- The Rady Children's Institute for Genomic Medicine (RCIGM) and Alexion Pharmaceuticals, Inc. (NASDAQ:ALXN) today announced a strategic partnership to accelerate the diagnosis of critically-ill newborns with rare genetic disorders. The collaboration combines the Institute's genomic research expertise with Alexion data science and bioinformatics capabilities to advance precision medicine for infants in an intensive care setting.

This Smart News Release features multimedia. View the full release here:

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"Diagnosing acutely ill babies is a race against the clock, so it's essential for physicians to have access to solutions that will provide answers faster and help set the course of treatment," said Stephen Kingsmore, MD, DSc, President and CEO of the Institute. "Winning this race will require collaborative effort, which is why we are delighted to work with the people at Alexion who share our vision for unraveling the mysteries of genetic disease and giving hope to families with critically sick newborns."

There is great need for employing such technology in medicine. As many as 15 percent of babies born in the United States are admitted to neonatal or pediatric intensive care units (NICU/PICU). Among these infants, up to one-third are likely to be affected by genetic diseases or congenital anomalies which are also the leading cause of death among babies in the NICU. 1-11

Rapid diagnosis through genome sequencing can provide definitive answers, allowing physicians to provide timely, targeted treatment that can help prevent a needless diagnostic odyssey and improve medical outcomes. The rapidly falling cost of whole-genome sequencing increases the feasibility for clinical testing for rare genetic diseases. However, the amount and complexity of data continues to grow.

"In rare diseases, rapid diagnosis is made all the more challenging by the significant amount of genomic and phenotypic data a clinician must sift through to reach a diagnosis," said John Reynders, PhD, Vice President of Data Sciences, Genomics, and Bioinformatics at Alexion. "This collaboration will help accelerate an accurate diagnosis for patients with genetic diseases, clarify available paths of intervention and provide hope to families."

Under the partnership, Alexion will share, research and further refine the SmartPanel, a platform developed by Alexion that personalizes and prioritizes suspected rare-disease genes from a patient's next-generation sequenced genome and specific clinical presentation. The Rady Children's Institute for Genomic Medicine is evaluating the SmartPanel in research to establish positive predictive value, enable electronic medical record (EMR) integration for rapid phenotypic extraction and assess overall patient outcomes via earlier diagnosis. Both organizations will collaborate on patient and disease characterization, algorithmic modules and scalability with a shared goal of contributing core capabilities to the open source community to accelerate research in the challenging area of pediatric rare-disease diagnosis.

### **About Rady Children's Institute for Genomic Medicine**

The institute is leading the way in advancing precision healthcare for infants and children through genomic and systems medicine research. Discoveries at the Institute are enabling rapid diagnosis and targeted treatment of critically ill newborns and pediatric patients at Rady Children's Hospital-San Diego. The vision is to develop an integrated process that can be expanded to deliver precision pediatric medicine at children's hospitals in California, the nation and the world. RCIGM is a division of Rady Children's Hospital-San Diego. Learn more at [www.RadyGenomics.org](http://www.RadyGenomics.org).

### **About Rady Children's Hospital-San Diego**

[Rady Children's Hospital-San Diego](http://www.RadyChildrens.org) is a 551-bed pediatric care facility providing the largest source of comprehensive pediatric medical services in San Diego, Southern Riverside and Imperial counties. Rady Children's is the only hospital in the San Diego area dedicated exclusively to pediatric healthcare and is the region's only designated pediatric trauma center. In June 2016, *U.S. News & World Report* ranked Rady Children's among the best children's hospitals in the nation in nine pediatric specialties. Rady Children's is a nonprofit organization that relies on donations to support its mission. For

more information, visit [www.rchsd.org](http://www.rchsd.org) and find us on [Facebook](#), [Twitter](#) and [Vimeo](#).

## About Alexion

Alexion is a global biopharmaceutical company focused on developing and delivering life-transforming therapies for patients with devastating and rare disorders. Alexion is the global leader in complement inhibition and has developed and commercialized the first and only approved complement inhibitor to treat patients with paroxysmal nocturnal hemoglobinuria (PNH) and atypical hemolytic uremic syndrome (aHUS), two life-threatening ultra-rare disorders. In addition, Alexion's metabolic franchise includes two highly innovative enzyme replacement therapies for patients with life-threatening and ultra-rare disorders, hypophosphatasia (HPP) and lysosomal acid lipase deficiency (LAL-D). Alexion is advancing its rare disease pipeline with highly innovative product candidates in multiple therapeutic areas. This press release and further information about Alexion can be found at: [www.alexion.com](http://www.alexion.com).

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## References:

1. Couce ML, Bana A, Boveda MD, Perez-Munuzuri A, Fernandez-Lorenzo JR, Fraga JM. Inborn errors of metabolism in a neonatology unit: impact and long-term results. *Pediatrics international : official journal of the Japan Pediatric Society* 2011;53:13-7.
2. Weiner J, Sharma J, Lantos J, Kilbride H. How infants die in the neonatal intensive care unit: trends from 1999 through 2008. *Archives of pediatrics & adolescent medicine* 2011;165:630-4.
3. Wilkinson DJ, Fitzsimons JJ, Dargaville PA, et al. Death in the neonatal intensive care unit: changing patterns of end of life care over two decades. *Archives of Disease in Childhood - Fetal and Neonatal Edition* 2006;91:F268-F71.
4. Ray JG, Urquia ML, Berger H, Vermeulen MJ. Maternal and neonatal separation and mortality associated with concurrent admissions to intensive care units. *CMAJ : Canadian Medical Association journal* 2012;184:E956-62.
5. Yoon PW, Olney RS, Khoury MJ, Sappenfield WM, Chavez GF, Taylor D. Contribution of birth defects and genetic diseases to pediatric hospitalizations. A population-based study. *Archives of pediatrics & adolescent medicine* 1997;151:1096-103.
6. O'Malley M, Hutcheon RG. Genetic disorders and congenital malformations in pediatric long-term care. *Journal of the American Medical Directors Association* 2007;8:332-4.
7. Soneda A, Teruya H, Furuya N, et al. Proportion of malformations and genetic disorders among cases encountered at a high-care unit in a children's hospital. *European journal of pediatrics* 2012;171:301-5.
8. Hagen CM, Hansen TW. Deaths in a neonatal intensive care unit: a 10-year perspective. *Pediatric critical care medicine : a journal of the Society of Critical Care Medicine and the World Federation of Pediatric Intensive and Critical Care Societies* 2004;5:463-8.
9. Berger TM, Hofer A. Causes and circumstances of neonatal deaths in 108 consecutive cases over a 10-year period at the Children's Hospital of Lucerne, Switzerland. *Neonatology* 2009;95:157-63.
10. Pinar H. Postmortem findings in term neonates. *Seminars in neonatology : SN* 2004;9:289-302.
11. NICU Summary. March of Dimes. (Accessed July 20, 2016, at [https://www.marchofdimes.org/peristats/pdfdocs/nicu\\_summary\\_final.pdf](https://www.marchofdimes.org/peristats/pdfdocs/nicu_summary_final.pdf).)

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