MedicalXpress

New genetic pathways linked to severe lung disease in preemies

October 24 2018

Scientists from Stanley Manne Children's Research Institute at Ann & Robert H. Lurie Children's Hospital of Chicago and colleagues identified promising new genetic pathways associated with severe lung disease in extremely premature infants, as well as pathways linked to faster recovery from lung disease in this population. The study is the largest to date to perform whole exome sequencing—or examine all the genes that code for proteins—in relation to respiratory outcomes of prematurity. This method is considered to be an efficient way to establish direct links between genetic changes and disease. Their findings were published in *BMC Genetics*.

"Our results lend further support to the theory that some chronic respiratory problems in premature babies have a genetic basis," says lead author Aaron Hamvas, MD, Division Head of Neonatology at Lurie Children's and Professor of Pediatrics at Northwestern University Feinberg School of Medicine. "Some of the genetic pathways we found make sense biologically and warrant further research. Ultimately, we hope that early genetic testing could help us identify infants at high risk for severe lung disease, and reveal the precise genetic cause of their disease, so that we can treat it most effectively. Better understanding of genetic causes of lung disease in these babies will bring us closer to developing more precise treatments."

The study completed whole exome sequencing on 146 extremely premature infants born at less than 29 weeks of gestation, examining genetic variations and pathways in connection to the extremes in

respiratory outcomes. The group with the most severe extreme of lung disease required continuous respiratory support up to 36 weeks post menstrual age (PMA). The group with the least affected extreme only required respiratory support for less than two weeks after birth and did not require any respiratory support at 36 weeks PMA.

One of the promising genetic pathways identified by this study relates to the gonadotropin releasing hormone, which is involved in sex differences and reproductive functions. Dr. Hamvas and colleagues found that this pathway is overrepresented in babies with the most severe chronic lung disease.

"Our observation that a hormonal pathway is related to more severe lung disease is intriguing because we know that there are sex differences in the risk for chronic lung disease in premature infants, with boys more susceptible to worse outcomes," says Dr. Hamvas, who also is the Raymond & Hazel Speck Berry Professor in Neonatology. "Could the sex differences we see clinically be the result of genetic changes in this pathway? More studies are needed to answer this question."

Another promising genetic pathway that is overrepresented in preemies with severe lung disease involves genes that encode heart development.

"Our discovery that genetic changes in a cardiac pathway are associated with chronic lung disease might explain why so many of these babies go on to develop pulmonary hypertension," says Dr. Hamvas. "One of our research projects is actively pursuing this connection, trying to understand the direct mechanisms involved."

More information: et al, Exome sequencing identifies gene variants and networks associated with extreme respiratory outcomes following preterm birth, *BMC Genetics* (2018). DOI: 10.1186/s12863-018-0679-7

Medical Xpress

Provided by Ann & Robert H. Lurie Children's Hospital of Chicago

Citation: New genetic pathways linked to severe lung disease in preemies (2018, October 24) retrieved 2 May 2024 from https://medicalxpress.com/news/2018-10-genetic-pathways-linked-severe-lung.html

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