## Meet the Scientist

# Identifying Genetic Markers to Assess Health Risks in Newborns


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There can be few experiences more heartbreaking than learning a newborn infant has a severe, life-threatening disorder. Dr. Kate Ackerman is working to help these infants by more accurately identifying genetic markers that can alert caregivers to disorders that may not be otherwise apparent.

While performing her fellowship at Boston Children's Hospital, Dr. Ackerman saw that despite what might appear to be a straightforward problem in an infant, might actually be more complicated.

"We had babies that we considered low risk, but their lungs or heart never functioned properly, and they would die in six months," says Dr. Ackerman. When she came to the University of Rochester, she asked, "What are the related factors we're not seeing? At the time, no one anywhere really knew the answer."

Dr. Ackerman was the first to identify a gene that was causing particular defects in both the development of the diaphragm and the lungs. She is now part of a multicenter study that allows her to take a gene suspected to play a role in normal heart, diaphragm, or lung development and compare it to real-world effects in a human very quickly. When children are identified with birth defects caused by problems with specific genes, Dr. Ackerman studies mice with the same genetic problems and tests extensively for other congenital problems, such as kidney disease, that might arise as the mouse ages—giving a crucial heads-up to doctors that a similar unknown problem might arise in the child months or years down the road.

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"It's so important because this program gives us opportunities to identify potential therapeutic targets for these kids," says Dr. Ackerman. "My hope is to use cutting-edge genetics to make the difference in our care of the complete child. That's the goal."