

NEWS RELEASE

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Genomic Medicine, Physician Dedication Give Family Answers About Teen's Rare Disease

(COLUMBUS, Ohio) – Summer Nagele loves photography. She hopes to one day become a professional sports photographer, capturing moments of football, basketball, baseball and hockey. At age 18, Summer is focused on her college plans, but when she was born, she faced many immediate challenges – struggles with breathing, feeding and mobility, and a cleft palate that was repaired before her first birthday – and many uncertainties.

Summer's condition remained a mystery for many years. An initial diagnosis of congenital myasthenic syndrome (CMS) gave the family some direction, and medication proved to be helpful. However, some of Summer's symptoms did not overlap with CMS, such as her cleft palate and short stature. Her family's search for answers brought them to Nationwide Children's.

Through whole exome sequencing performed in 2020 at the <u>Steve and Cindy Rasmussen Institute</u> for <u>Genomic Medicine</u> at Nationwide Children's, the Nagele family was finally able to put a real name to Summer's rare disease: Birk-Barel Syndrome, a genetic condition that has been reported fewer than fifty times worldwide, in scientific literature.

"There would have been no possible way to diagnose Birk-Barel Syndrome 30 years ago," said <u>Anne</u> <u>Connolly, MD</u>, chief of the<u>Neurology</u> division at Nationwide Children's Hospital, and Summer's physician. "In Summer's case, we guessed a group of maybe 10 diagnoses that might fit, and none of them were correct. With whole exome sequencing, some of that guesswork is taken out. The diagnostic odyssey is absolutely shortened."

In addition to identifying Summer's condition, participating in whole exome sequencing as a family provided another insight: Autumn Nagele, Summer's mom, was found to also have a genetic disorder, a type of limb girdle muscular dystrophy. Now, both Summer and Autumn see Dr. Connolly twice a year in the muscular dystrophy clinic at Nationwide Children's.

"Whole exome sequencing, I believe, is becoming a more standard approach," said <u>Alayne Meyer</u>, <u>MS, CGC</u>, genetic counselor at Nationwide Children's. "It is becoming more common practice to jump straight to the exome, because we know that there are so many new genetic conditions being discovered that smaller tests don't always catch."

"Dr. Connolly made a promise to my daughter at 8 years old, that she would find out what her diagnosis was, even if she had to name it after both of them," said Autumn. "It took a while, but she kept her promise, and that meant the world. She never stopped looking, she never stopped trying."

To learn more about Summer's story, click here.

About The Abigail Wexner Research Institute at Nationwide Children's Hospital

Named to the Top 10 Honor Roll on U.S. News & World Report's 2022-23 list of "Best Children's Hospitals," Nationwide Children's Hospital is one of America's largest not-for-profit free-standing pediatric health care systems providing unique expertise in pediatric population health, behavioral health, genomics and health equity as the next frontiers in pediatric medicine, leading to best outcomes for the health of the whole child. Integrated clinical and research programs are part of what allows Nationwide Children's to advance its unique model of care. As home to the Department of Pediatrics of The Ohio State University College of Medicine, Nationwide Children's faculty train the next generation of pediatricians, scientists and pediatric specialists. The Abigail Wexner Research Institute at Nationwide Children's Hospital is one of the Top 10 National Institutes of Health-funded free-standing pediatric research facilities in the U.S., supporting basic, clinical, translational, behavioral and population health research. The AWRI is comprised of multidisciplinary Centers of Emphasis paired with advanced infrastructure supporting capabilities such as technology commercialization for discoveries; gene- and cell-based therapies; and genome sequencing and analysis. More information is available at NationwideChildrens.org/Research.