

# Advancements in genetic sequencing find long-awaited answers for those with rare diseases

*Advances in genetic sequencing technology give teen correct diagnosis and a path forward*

*Nationwide Children's Hospital*

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## NEWS PACKAGE

<b>SUGGESTED TEASE</b>	ADVANCES IN GENETIC TESTING ARE NOW IDENTIFYING CAUSES OF THOUSANDS OF RARE DISEASES THAT USED TO BE A MYSTERY. COMING UP, WHAT IT'S MEANT TO A TEEN WHO'S WAITED HER WHOLE LIFE FOR ANSWERS.
<b>ANCHOR LEAD</b>	THERE ARE AN ESTIMATED TEN THOUSAND DISEASES CAUSED BY A VARIANT IN A SINGLE GENE <sup>1</sup> , AND UNTIL RECENTLY, THE MILLIONS OF PEOPLE LIVING WITH ONE OF THESE GENETIC CONDITIONS WERE OFTEN LEFT SEARCHING FOR ANSWERS AS TO WHAT WAS ACTUALLY CAUSING THEIR SYMPTOMS. ADVANCEMENTS IN GENETIC TESTING <sup>2</sup> NOW ALLOW SCIENTISTS TO SEQUENCE LARGE AMOUNTS OF D-N-A QUICKLY AND ACCURATELY TO PINPOINT GENETIC VARIANTS AND THE DISEASES THEY CAUSE. BARB CONSIGLIO HAS THE DETAILS ON HOW THIS TECHNOLOGY GAVE A TEENAGER AND HER PARENTS THE ANSWERS THEY'D BEEN SEEKING HER ENTIRE LIFE.
<b>(PACKAGE START) -----</b> <b>CG: Courtesy: Nationwide Children's Hospital</b> <b>:00 - :03</b> Shots of Summer in physical therapy	<i>(Nats of Summer in therapy) :02</i>  SUMMER NAGELE (NEG-lee) IS A KIND AND DRIVEN TEENAGER. BUT SINCE BIRTH SHE HAS HAD SOME UNIQUE HEALTH CHALLENGES. :05
<b>CG: Michael Nagele</b> <b>Summer's dad</b>	<i>"She couldn't lift her head up, she couldn't lift her arms up, so we knew from the first day that there was something different." :08</i>
<b>CG: Autumn Nagele</b> <b>Summer's mom</b>	<i>"My husband and I looked at each other, very scared, // And vowed never to limit our child." :07</i>
Shots of Dr. Connolly greeting family	THE NAGELES SEARCHED FOR ANSWERS, WHICH

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<p>Shots of Summer in physical therapy</p>	<p>LED THEM TO DOCTOR ANNE CONNOLLY, WHO HELPED FIND EFFECTIVE THERAPIES AND MEDICATIONS FOR SUMMER. :06</p> <p><i>8149 (Summer running) "Keep going. You got it." :02</i></p>
<p>Shots of Dr. Connolly doing exercises with Summer</p>	<p>AND DOCTOR CONNOLLY PROMISED THAT ONE DAY THEY WOULD HAVE A CLEAR DIAGNOSIS FOR HER CONDITION. :04</p>
<p><b>CG: Anne Connolly, MD</b> <b>Nationwide Children's Hospital</b></p>	<p><i>"I will walk with you. Even if I don't know the diagnosis today, I will keep trying." :07</i></p>
<p>Shots of IGM lab</p>	<p>AS SUMMER GREW, SO DID ADVANCEMENTS IN GENETIC TESTING. AND AT AGE SIXTEEN, SHE UNDERWENT WHOLE EXOME SEQUENCING AT NATIONWIDE CHILDREN'S HOSPITAL, WHICH EXAMINES D-N-A SEQUENCES THAT IMPACT HOW GENES FUNCTION. :11</p>
<p><b>CG: Alayne Meyer, MS, CCG</b> <b>Licensed Genetic Counselor</b></p>	<p><i>"We just give the symptoms that the patient has and they look through all of their genes to try to assess whether any alterations there could be explaining those symptoms." :09</i></p>
<p>Shots of Summer in appointment</p>	<p>THE RESULTS REVEALED THAT SUMMER IS ONE OF ABOUT FIFTY KNOWN CASES OF A RARE GENETIC CONDITION CALLED BIRK-BAREL SYNDROME. :05</p>
<p><b>Autumn Naegele (CG'd earlier)</b></p>	<p><i>"She kept her promise to my daughter and that meant the world. She never stopped looking, she never stopped trying." :07</i></p>
<p>Shots of Dr. Connolly working with Summer</p> <p><b>Dr. Connolly (CG'd earlier)</b></p>	<p>BUT THEIR JOURNEY IS FAR FROM OVER. :02</p> <p><i>"Once you know the gene, that's just the starting point. You have to know what the gene is doing, and most importantly, can you help improve whatever mechanism this is impairing." :13</i></p>
<p>Shots of Summer and Dr. Connolly</p>	<p>THE PERSISTENCE OF SUMMER'S CARE TEAM CHANGED HER LIFE... :02</p> <p><i>Dr. Connolly holding Summer's baseball card "Flip it." (has note on back for Dr. C.) :02</i></p> <p>AND GAVE HER A CLEARER PATH FORWARD. :02</p>
<p><b>CG: Summer Nagele</b> <b>Diagnosed with rare genetic disease</b></p>	<p><i>"I just like to try to be as normal as I can be, but in my own way." :05</i></p>

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<p>Shots of family and Dr. Connolly</p> <p><b>(PACKAGE END) -----</b></p> <p><b>ANCHOR TAG</b></p>	<p>AT NATIONWIDE CHILDREN'S HOSPITAL, THIS IS BARB CONSIGLIO REPORTING. :03</p> <hr/> <p>RESEARCHERS AT NATIONWIDE CHILDREN'S HOSPITAL HAVE FOUND THAT AMONG THEIR PATIENTS WHO COULD NOT BE DIAGNOSED THROUGH PREVIOUS TESTING METHODS, A THIRD RECEIVED A CLEAR ANSWER THROUGH EXOME SEQUENCING.</p> <p>SUMMER'S DIAGNOSIS ALLOWED HER FAMILY TO CONNECT WITH OTHERS WITH THE SAME CONDITION AS WELL AS LEADING EXPERTS WHO ARE DEVELOPING NEW TREATMENTS.</p>
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**SOCIAL MEDIA**

<p> <b>Share it! Suggested tweet:</b></p> <p> <b>Suggested post:</b></p>	<p>After searching her whole life for answers, advancements in genetic testing at <a href="https://www.nationwidekids.org">@NationwideKids</a> finally gave Summer Nagele a definite diagnosis for her rare inherited disease and a clearer path for treatment. <a href="https://bit.ly/3X4e6BE">https://bit.ly/3X4e6BE</a></p> <hr/> <p>When Summer Nagele was 8 years old, Dr. Anne Connolly made a promise to one day give her a clear diagnosis for her rare genetic condition. And with the help of advancements in genetic testing at <a href="https://www.nationwidechildrens.org">Nationwide Children's Hospital</a>, she was able to fulfill that promise when Summer was 16 years old, giving her a clearer path forward for treatment. <a href="https://bit.ly/3X4e6BE">https://bit.ly/3X4e6BE</a></p>
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**EXTRA BITES**

<p><b>CG: Anne Connolly, MD Nationwide Children's Hospital</b></p>	<p>Connolly describes new advancements in genetic testing and treatment:  <i>"There are early diagnoses that can be diagnosed by newborn screening and things like that, that if you know early enough an intervention may be possible. So, where I see it going is, I would say earlier and more often."</i> :18</p> <p>Connolly says Summer's symptoms greatly affect her life:  <i>"If you've met Summer, she's active. She wants to be doing things. Having to stop and not do things in the afternoon or evening, not be with friends, not do extracurricular activities very much impacts her life."</i> :20</p> <p>Connolly says they were not able to identify Summer's condition before genetic testing:  <i>"We knew that she had weakness and we knew she had fatigue. So we tested her for a common set of genetic disorders that would link those together and she didn't have any of them."</i> :14</p>
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**CG: Anne Connolly, MD**  
Nationwide Children's Hospital

Connolly says Summer's mom was also diagnosed with a genetic disease after exome sequencing:  
*"It turned out that Summer's mother, whose name is Autumn, was able to... she gave her DNA and it turned out that she had a genetic disorder as well, and hers was related to an adult onset muscle disease."* :19

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Meyer says exome sequencing also revealed a condition in Summer's mom:

*"Through that testing, we also identified that Summer's mother has a type of limb-girdle muscular dystrophy. So that was a little bit unexpected, but we had talked about the possibility of having these unexpected findings up front. And so that actually allowed her mother to get the care that she needs as well."* :19

**CG: Alayne Meyer, MS, CCG**  
Licensed Genetic Counselor

Meyer says it means a lot to families to get a diagnosis:

*"This is a family who's had questions about where these symptoms are coming from since their daughter was a baby. And so it does have a lot of meaning for those families to have a name to put to those symptoms and also be able to connect with, even though it's a small community, a community of other families who have that same diagnosis."* :18

Meyer says broad genetic testing was needed to give Summer a correct diagnosis:

*"We looked at what had been done and thought that there was some gaps as far as what still could be evaluated. And given that Summer had a variety of symptoms, we thought that doing a more broad genetic test made the most sense to try to figure things out for her."* :15

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**CG: Summer Nagele**  
Diagnosed with rare genetic disease

Summer states her plans for her future education and career:

*"I would like to be a professional sports photographer. But right now I'm really focused on going to a community college for a few years, and then maybe going on to a university."* :17

Summer explains some of her physical limitations:

*"I'm not as fast as everybody else and I have to rest more than everybody else."* :10

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**CG: Autumn Nagele**  
Summer's mom

Autumn says they vowed never to limit Summer's development:

*"The neonatologist told us 'your baby may never walk, talk, or eat on her own.' So it was at that point that my husband and I looked at each other, very scared, very new to parenthood in that NICU. And we vowed never to limit our child."* :19

<p><b>CG: Autumn Nagele</b> Summer's mom</p>	<p>Autumn says they received four diagnoses that gave them a lot of information: <i>"One test, four diagnoses, we call it the Big D day, the big diagnosis day. They gave us a lot of information that we likely would not have learned for many, many years."</i> :16</p> <p>Autumn says they did everything possible to find a diagnosis: <i>"We knew that as a family, we were doing everything possible and we were taking every step possible to find that diagnosis. And we did."</i> :13</p>
<p><b>CG: Michael Nagele</b> Summer's dad</p>	<p>Michael says they were excited about the option to have exome sequencing: <i>"We tried everything else. We tried the biopsy, we tried limited genetic testing, and we still didn't have an answer. So I think the opportunity presented itself and we were on board right away."</i> :15</p> <p>Michael says they're hopeful for new treatments, but just having a diagnosis is a relief: <i>"Maybe they'll come out with a different treatment or something down the road, but 05:09:03 having an answer, that is a big change because you don't have that cloud of uncertainty kind of hovering over your life."</i> :17</p> <p>Michael says Dr. Connolly was helping Summer, but continued on to find a diagnosis: <i>"Before the diagnosis, she helped improve her life on a day-to-day basis, but then to continue on and to give us the opportunity to get the genetic testing, to get a diagnosis, it just really changes things."</i> :15</p>

### References

<sup>1</sup>*Impact of Genetic Diseases. Genehome, 2021. Online:*

<https://www.thegenehome.com/basics-of-genetics/disease-examples>

<sup>2</sup>*What are whole exome sequencing and whole genome sequencing? MedlinePlus, July*

**2021. Online:** <https://medlineplus.gov/genetics/understanding/testing/sequencing/>

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