

Large, innovative autism project sparks hope for better treatments

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Dr. Laura Carpenter enjoys a fun moment with six-year-old Joseph Estell, who has autism. Credit: Sarah Pack

No words can describe how happy Quanita Estell is to hear her six-year-old son come home with hilarious stories to tell. First, she enjoys his sense of humor, but mostly she's thrilled to see Joseph, who was diagnosed with autism spectrum disorder at 2, is becoming better able to communicate.

It's been a long journey involving extensive treatment. But it's paid off.

Now she gets to hear all about his adventures learning to surf in a program that helps children who have autism. Water and music seem to work wonders for him, but she knows they still have a tough road ahead. It's one reason she plans to enroll in a new nationwide genetic study to track down clues to the causes of autism with the goal of potentially developing new treatments. The study opens April 1 at the Medical University of South Carolina.

"We try to get involved with anything that can help them," she says of a disorder that affects an estimated one out of 68 children.

The prevalence of autism has increased significantly over the past two decades, and while some think that it may be leveling off, MUSC autism researcher Laura Carpenter, Ph.D., says the verdict's still out. What she does know is a lot has been spent researching a disorder where few treatments have been developed.

Carpenter says she sees that potentially changing thanks to such studies as SC SPARK or Simons Foundation Powering Autism Research for Knowledge, which has launched a nationwide genetic study with the ambitious goal of enrolling 50,000 families to allow scientists to better understand the genetic changes that contribute to autism. MUSC is the South Carolina clinical partner for SPARK and will be serving the entire state.

Anyone of any age on the autism spectrum, including Asperger's syndrome and PDD-NOS or pervasive developmental disorder/not otherwise specified, is eligible to join. Researchers also want parents and siblings to be a part of the study, which involves providing medical history and saliva samples. Individuals who enroll with parents' participation will receive compensation. There are more than 20 clinical sites nationwide participating. Participants who enroll get access to the SPARK resources online, which include webinars featuring national experts.

"It's a real honor to be chosen," Carpenter says. "Many of the other clinical sites also selected are some of the leaders in autism research. This will open a lot of opportunities for MUSC to partner with other leaders in autism research."

It's those partnerships that can lead to change.

The majority of the money for research in autism has gone towards trying to figure out the genetic factors that lead to autism, and what researchers have found is that there are likely hundreds of genes that play a role, and to complicate matters, there are also likely environmental factors contributing to the development of the condition.

"Now some experts are starting to talk about the 'autisms,' instead of autism—meaning there are multiple pathways to developing autism. We think epigenetics plays a really big role, meaning that the environment probably influences which genes are active and which genes are ultimately implicated in autism. And we think it's probably a complex combination of genes and environment that lead to autism. What we know is that there are multiple genetic disorders that are associated with higher incidence of autism, but there is no one genetic change that results in autism. It's much more complicated than we had originally thought."

What also has become clear is that researchers need a much larger sample size than anyone ever dreamed about. That's what SPARK is all about, she says.

To put it in perspective, she considers a typical large, local study one that enrolls about 300 participants. The state's goal as part of the 50,000 being sought for recruitment is enrolling 1,000 participants, she says. "So, it's just a scale that has never been done before, and I think this is what is needed in order to move the field forward."

The more representative the sample of participants, the better the research. That's why Carpenter wants to get the word out statewide. People can enroll at various community events, by coming to MUSC, and even asking for a home visit. Participants also can enroll online in the comfort and privacy of their own homes.



Drs. Carpenter and Catherine Bradley want to see more treatment options for patients on the autism spectrum. Credit: Sarah Pack

"The state brings something really unique to the network in that we have a really diverse population. I think we have the potential to partner with people who don't necessarily participate in research very often. In order to get answers about a disorder that's so diverse, you really need to have everybody participate so that you have a representative sample," she says.

"We will be holding their hand throughout the participation process. In order for the data to be most useful to the doctors and scientists, we really need people with autism, both their biological parents, and any siblings who are willing to participate."

MUSC is committed to advancing treatments and care to improve the quality of life for those on the spectrum, she says. This initiative adds to many ongoing projects at MUSC, including:

- -Prevalence studies exploring the extent of the disorder, with the goal of raising awareness and encouraging early intervention
- CATS or Carolina Autism Transition Study that is examining the transition from adolescence to adulthood, a critical period for individuals living with autism
- The Learning Enhancement through Neurostimulation in Autism (LENS) study that is examining whether brain stimulation paired with social skills learning can help teenage boys with autism make and keep friends.
- Basic science explorations, such as that of neuroscience researcher Christopher Cowan, who specializes in the genetic underpinnings of autism
- Project REX, a treatment initiative focused on improving quality of life

for families affected by autism

-Piece it Together program, a wellness program geared for young adults with autism sponsored by MUSC's Wellness Center

Carpenter is lead researcher on the CATS study and preliminary findings are showing that too many young adults on the autism spectrum are failing to thrive.

"One of the things we know about autism is that if you intervene early, intensively with behavioral interventions, we can change the trajectory so that some kids will have really outstanding outcomes. So autism is treatable. But we also know that about 50 percent of kids will continue to have significant impairments, even if they get the best early intensive intervention - even the Cadillac of care, the best quality, the highest intensity - at least 50 percent of them will still need a lot of services after that. That tells us that we still need to develop other treatments."

There's another advantage to SC SPARK that shows just how innovative a research project it is. It's a stepping stone to personalized medicine, she says. Should a treatment be developed for a certain genetic sample, for example, those participants who match the profile can be notified in case they want to take advantage of any treatment that has been shown to be potentially effective. The genetic analysis identifies people who carry known genes with autism links.

According to SPARK, an estimated 5 to 10 percent of families are expected to receive genetic results in the first analysis. The genetic information gets re-analyzed every year, and as new autism genes are identified, additional genetic results will be returned. A genetic diagnosis is important because it can help in diagnosis and eliminate the need for additional testing, enable families to better understand the recurrence risk, identify opportunities to network with similar participants and be a

pipeline to learn about clinical trials and/or treatments specific to their diagnoses.

It also takes research to a new level and will enable autism studies of all types to get off the ground more quickly, something that's really needed, says Carpenter. MUSC is rolling this out in April, which is Autism Awareness month. April 2 marks an international celebration of Light it Up Blue, a day dedicated to raising awareness about the disorder. It's a good time to launch enrollment, she says.

"This study is amazing and totally groundbreaking. Not only are we going to get this huge cohort of people and genetic information for this effort, but now these groups of SPARK participants are going to be notified when new ideas come on board. They'll be ready to go. One of the ideas is that [autism](#) may eventually be something that is treated with a more precision approach and, if that is the case and you have this really big sample of people, you might be able to pick out the ones you think will respond better for one treatment versus others."

Provided by Medical University of South Carolina

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