

NEWS

# Firsthand experience with autism can spark new science

BY NICHOLETTE ZELIADT

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**Mark Zylka**, an expert in pain and brain development, never dreamed he would study how mice sleep. Yet that is exactly what he did after meeting families affected by **Angelman syndrome**.

Children with the condition, which is genetically related to some forms of autism, have developmental delays and movement problems. Most of them speak few or no words. But in talking to parents with affected children, Zylka learned they had another major concern: **sleep problems**.

“I think they felt completely helpless. They have these children who are up at all odd hours of the night and doing who knows what,” says Zylka, associate professor of cell biology and physiology at the University of North Carolina at Chapel Hill. “They were worried about them escaping from the house and running away or hurting themselves.”

Zylka and his colleagues decided to explore the molecular roots of sleep problems in a mouse model of Angelman. Last year, his team showed that the mice have intact body clocks but lack the typical need for sleep after a period of wakefulness<sup>1</sup>. “Basically, that whole study was motivated by conversations with parents,” Zylka says.

Zylka’s experience illustrates how firsthand contact with people who have certain conditions can inspire new avenues of research. “It gets you to start thinking outside the box,” he says.

## Tricky translation:

Meeting people with autism, instead of merely reading about them, can alter how scientists approach their work. Mouse researcher **Jacqueline Crawley** says seeing children with autism choose to play with a toy or computer instead of with other children inspired her to develop a now widely used test of social deficits in mice. The test, called the three-chambered social approach assay, probes a mouse’s sociability, defined as spending more time with a novel mouse than with a novel inanimate object.

British researcher **Albert Basson** shifted his focus after meeting children with CHARGE syndrome, a condition characterized by poor growth and congenital defects in the heart, eyes, ears and other organs. About one-third of these children have autism-like behaviors, but Basson noticed some important distinctions.

“They’re really interested in social interactions,” says Basson, associate professor of developmental and stem cell biology at King’s College London. However, he adds, they have trouble with attention and self-control, which can contribute to social difficulties. After noticing this, Basson opted to study brain areas and circuits involved in executive function — which includes planning, attention and self-control, among other aspects of cognition — rather than circuits thought to be specific to social skills.

## Social subtleties:

Similarly, many people with the autism-linked disorder **fragile X syndrome** show **social problems that are subtly different** from those in people with other forms of autism, says **Joseph Piven**,

Thomas E. Castelloe Distinguished Professor of Psychiatry, Pediatrics and Psychology.

He and his colleagues found that these children often have motor-based **repetitive behaviors**, such as hand flapping, as many children with autism do. But most of them do not have cognitive-based ones that are associated with autism, such as an insistence on sameness.

Closer examination revealed that the social problems in children with fragile X syndrome stem largely from severe social anxiety rather than from lack of interest in others. “They take a while to warm up, but they don’t have the sort of reciprocity problems of autism,” Piven says. “If you don’t think about it at that nuanced level, you’ll think there’s just this one thing called autism.”

Interacting with people who have autism can help researchers better understand the condition’s heterogeneity. “I think in order to avoid a very simplistic interpretation, it’s always good to have that human context,” says **Susan Santangelo**, director of the Center for Psychiatric Research in Portland, Maine, who studies children with severe autism symptoms.

Interactions with individuals on the spectrum can also motivate researchers to remain engaged in their work. “For me, the inspiration to work long hours, to toil away, is driven by my firsthand experience with individuals and families with autism,” says **Raphael Bernier**, professor of psychiatry and behavioral sciences at the University of Washington in Seattle.

Bernier says he is eager to take on graduate students and postdoctoral fellows who have known people with autism. “I feel most confident that those scientists will stay in the field and stay dedicated to autism,” he says.

## Lab visits:

So what is the best way for researchers to meet people with autism? Perhaps the easiest approach is to collaborate with clinicians, as Crawley did.

“With the permission of the families, members of my lab and I have had opportunities to observe infants, toddlers, children, adolescents and young adults with autism, either from videos or through one-way observation rooms,” says Crawley, professor of psychiatry and behavioral sciences at the University of California, Davis. “We can really understand what we are working toward when we see the challenges that families and people with autism face daily.”

Some scientific conferences, such as the **International Meeting for Autism Research**, welcome families. And many colleges and universities offer opportunities to meet people with autism as part of their graduate and postgraduate training programs.

“We make sure that individuals with autism and family members are guest lecturers during each course,” says Bernier, who is involved in a graduate certificate program at his university. “The

students feel that is the most relevant and thought-provoking learning experience of each quarter.”

Another approach is to invite people with autism or their families to visit the lab for an hour or a day. These visits have the additional benefit of giving families a glimpse of research in action, Zylka says. “When you meet the family members and the kids with these neurodevelopmental disorders, you really get the sense that your research has meaning.”

**REFERENCES:**

1. Ehlen J.C. *et al. J. Neurosci.* **35**, 13587-13598 (2015) [PubMed](#)