Rain Man and Einstein

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50 years ago, autism was little understood, and its casualties were relegated to institutions. Today, UCSB is leading the way both in understanding the mechanisms of the disease and in developing effective treatments for it.

Decades ago, children with severe social problems?many who didn?t talk and didn?t behave?were dispatched to psychiatric hospitals. Called ?autistic,? they languished in a life of hopelessness, separated from family, friends, and normal life. Now UC Santa Barbara is leading the way, on two major fronts, in understanding and treating what has come to be recognized as a complex of disorders manifested in varying degrees of severity.

It was at the infamous Camarillo State Hospital that Lynn Koegel first encountered people with what?s now known as autism spectrum disorder. Now, years later, she is one of the most renowned clinicians in the field; she runs UCSB?s Koegel Autism Center together with her equally renowned husband, Robert Koegel, whom she first met at Camarillo.

?Things have really changed a lot,? Lynn Koegel says. Severely autistic people are no longer consigned to an invisible, institutionalized life, and the number of people diagnosed with autism spectrum disorder has increased considerably in the last few years, now affecting 1 in 150, although it?s not clear why.
This is an epidemic that won’t go away, Koegel says. It’s not just people being more aware—although they are—it really is an increase in numbers. Awareness has increased in recent years due both to the efforts of autism-focused nonprofits, and to the 1988 Oscar-winning movie Rain Man, which featured Dustin Hoffman as Raymond Babbitt, the autistic older brother of Charlie Babbitt, played by Tom Cruise.

Autism is a developmental disability that causes problems with social interaction and communication. Autism is a spectrum disorder which affects individuals with a considerable range and severity of symptoms. People with Asperger’s syndrome, the mildest form of autism spectrum disorder, may have good verbal, quantitative, or artistic skills, but they typically have trouble understanding body language and other non-verbal cues, and often have an obsessive interest in a single object or topic. Famous scientists and artists, including Einstein, Newton, Darwin, Orwell, van Gogh, and Mozart, are often said to have had Asperger’s. People whose autism is severe typically have far more limited engagement with the world around them—they may not speak and may engage in repetitive behaviors, such as rocking back and forth or waving their hands repeatedly.

It’s not yet known what causes autism. One of the more prevalent theories of recent years—the idea of a link between childhood vaccines and autism—has failed to hold up in scientific studies. Every case of autism is a little different, says Ken Kosik, co-director of the Neuroscience Research Institute at UCSB.

There’s no cure for the disorder, but skilled intervention can minimize problems and improve the lives of patients and their families. Our end goal, Koegel says, is for people with autism to be fully productive members of society with good jobs, loving families and friends, and a satisfying life... The Koegels believe this someday will be possible, for even the most severe cases.
While clinicians like the Koegels have made great progress in figuring out how best to help people with autism, little is known about what happens in the brain to give rise to its symptoms. "Something's not working right? in the brains of people with autism, Lynn Koegel says. "but we don't understand what?"

Because autism encompasses such a range of symptoms that vary widely in their severity, it's exceedingly tough to figure out the mechanisms that give rise to the disorder. "It's very complex," Kosik says. "There are certainly many different ways to acquire it. Different genes can go wrong in different people."

Some cases of autism are known to be caused by abnormalities in certain genes, but those are the minority of instances. In most cases, Kosik says, there are almost certain to be small contributions from many genes—as is the case for other complex disorders, such as diabetes and heart disease, that are very challenging to understand and address. Kosik, who has spent most of his career researching the mechanisms in the brain that produce the devastating symptoms of Alzheimer's disease, recently turned his attention to autism.

While he and the Koegels haven't yet worked together, Kosik says he's "looking forward to having some discussions with them" and "building some bridges here" between their clinical work and his focus on the genetic and biochemical mechanisms that underlie brain disorders.

To try to figure out what sets apart the autistic brain, Kosik chose to focus on snippets of genetic material called microRNAs, to see how they might be involved in the disorder.

MicroRNAs were discovered about 15 years ago; since then, these tiny fragments have had a big impact. The study of microRNAs is "one of the most explosive fields in all of biology," Kosik says. They play a key role in regulating genes, and scientists have linked them to some cancers and other diseases.

MicroRNAs are found in all animals—humans probably have close to a thousand of them, according to Kosik—and they serve a kind of fine tuning role. "They take a biological system and tweak it so it's working properly," he says.

MicroRNAs play a particularly important role in the brain, Kosik says, especially in the early years of life, when it develops most rapidly. Because that's also the time when symptoms of autism start to show up, Kosik decided to look at whether microRNAs are somehow involved in the disorder. "It was a very logical connection," he says.

Kosik studied microRNAs in the brains of people with autism, using samples donated to tissue banks after their deaths. He found that a number of the several hundred microRNAs he analyzed were expressed differently in those samples, compared to tissue from people without autism.

"We are by no means trying to say that the microRNA regulation that we observed is cause and effect," Kosik says. "There's almost certainly something more fundamental going on," he says. "We're in very early days in terms of understanding of this disorder."

A better understanding of the mechanisms in the brain that underlie autism could help
scientists develop drug treatments for the disorder, or perhaps ways of preventing it. In the meantime, however, interventions like those pioneered by Lynn and Robert Koegel are the gold standard for autism treatment.

“We’ve come a long way since the days when children with autism weren’t getting an education,” says Lynn. Rather than keeping children with autism out of normal classes, the Koegels advocate mainstreaming. That way, children with autism benefit from being exposed to the typical behaviors of their peers and to the expectations educators and parents put on children without autism. The Koegels’ approach to managing autism has changed the way clinicians, teachers and families around the country, and the world, deal with individuals with autism.

In the past, Koegel says, “we just punished kids (with autism) when they had bad behaviors. Now we figure out why they are behaving badly. A lot of the time it’s because the kids are bored or the work is too hard.”

The Koegels’ treatment strategy focuses on positive reinforcement that takes advantage of a child’s interests. “We let them choose the activity?” Koegel says. “Toys, books, activities they enjoy. Let’s say they like balls. If they say they want to bounce the ball, we’ll let them bounce the ball.”

“It seems so logical,” Koegel adds, “but back in the early days not many people knew what to do. We’re also learning that if you vary the task, children with autism learn a lot more.”

Rather than trying to deal with problem behaviors one at a time, the Koegels’ method—termed “Pivotal Response Treatment”—focuses on key aspects of a child’s development, such as motivation or social initiations. They have researched and developed “Pivotal Behaviors” that, once taught, have a widespread positive effect on lots of other untreated behaviors. They are, in a sense, “pivotal” to learning.

“It’s really important,” Koegel says, “to figure these kids out and learn some of the tools. Those tools can also help children without autism. For example, she says, kids will take more easily to handwriting if they pen postcards to friends, rather than repeatedly looping words on a page.

While much of the research and training done at the Koegel Autism Center focuses on children, Lynn Koegel and others at UCSB have also begun working with UCSB students who have been diagnosed with Asperger’s syndrome.

The disorder has been paid little attention until now, Koegel says, because the problems
associated with it are relatively mild. People with Asperger’s typically do fine academically, but “their social communication with peers is a real problem,” she says. “A lot of them end up spending their lunch in the library.”

People with Asperger’s syndrome generally want the same things as most of their peers, she says—a good job, friends, a family of their own—but find it hard to establish and maintain relationships. Those difficulties can lead to a multitude of problems, from legal tangles triggered by misunderstandings to depression.

The UCSB program aims to help the students succeed beyond the realm of books and exams. Participants get help upgrading their social skills—learning to look people in the eye when they’re talking, for example—and even run through practice dates.

The program, made possible by a large donation from Eli and Edythe L. Broad, only began last year, but Koegel says the students are definitely socializing more and are happier.

As encouraging as those results are, one of the keys to success in treating autism—whether the relatively mild difficulties associated with Asperger’s syndrome or the devastating problems that accompany more severe forms of autism—is early intervention.

These days, fortunately, educators and doctors place tremendous emphasis on detecting problems in young children and beginning treatment as soon as possible. That constitutes a major break with previous attitudes, Koegel says, when “pediatricians used to say, ‘Just wait??’

Signs of the disorder usually appear in the first years of life. Children with autism tend to be slow to develop language skills, don’t wave or gesture like other children, and don’t respond to their name being called. If a child over the age of 18 months isn’t talking, Koegel says, a specialist should evaluate them?

“We’re happy to get them at two (years of age), but our goal would be to get them in infancy some day,” Koegel says.

With early intervention, she says autistic children can show marked improvement. She cites the example of a child she worked with as part of an episode of the television series “Supernanny,” in which a childcare specialist—sometimes helped by professionals like Koegel—comes to the rescue of a family in crisis. The boy with autism, aged three, wasn’t speaking before he met Koegel, but began talking during the week she worked on the show.

Early intervention makes sense to Kosik. “In terms of the biology,” he says, “it’s very reasonable.” At that age, “the brain is very plastic,” Kosik adds, and with early intervention, “we may be able to direct the formation of pathways in directions that might make a person healthier.”

Links:

Autism information from the National Institutes of Health: www.nichd.nih.gov/health/topics/asd.cfm [2]
UCSB Koegel Autism Center: education.ucsb.edu/autism [3]

Ken Kosik: lifesci.ucsb.edu/mcdb/faculty/kosik [4]

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