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Tourette's DNA center established at Rutgers

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Jay Tischfield, a world-renowned geneticist at Rutgers University, remembers the expression on his son's face during a visit to a neurologist more than a decade ago.

The boy was being teased in school for repeatedly flickering his eyes. The kids called him "Blinky" and made him an outcast. The reason for the eye tic, the physician told the boy, was that he had Tourette's syndrome, an inherited neurological condition that can produce unusual behaviors. The neurologist said he immediately knew what it was because he also suffered from the same disorder.

"At that very moment, my kid realized he wasn't doomed," said Tischfield, the Duncan and Nancy MacMillan Professor of Genetics, whose son, now 25, is a student at Harvard University.

Though Tischfield's own epiphany about the syndrome came much later, the scientist, working with parents advocates, yesterday announced the opening of the country's first public cell and DNA repository for research on Tourette's.

"All my career, I stayed away from scientific topics that were of personal interest to me," Tischfield said. "I was afraid I couldn't be objective. Then I thought to myself, 'Idiot! Dummy! You can make a difference here.' And what's wrong with that?"

By making such vital information available to researchers worldwide, Tischfield and others who have family members with Tourette's believe scientists may find answers to the still-mysterious disorder, much as researchers with access to the genes of families with autism and schizophrenia are making strides.

And there's always the hope that such knowledge could bring cures.

"This is an important path to developing treatments," said Sol Barer, the CEO of Celgene Inc. in Warren, a company known for developing trail-blazing remedies for rare diseases. "It's a scientific, rational way to do it."

Barer's 19-year-old son, Joshua, also suffers from Tourette's syndrome. The elder Barer and Tischfield have been friends since Tischfield joined the Rutgers faculty in 1998.

Genetic and environmental factors play a role in the disorder, which usually arises at age six or seven, but the exact causes are unknown. Once considered rare and bizarre, the syndrome, named for a 19th century French physician, is now believed to occur in 1 out of 100 children, according to federal figures. Of the many physical and vocal tics that can occur with the syndrome, eye-blinking and throat-clearing are the most common behaviors.

Coprolalia, the exclamation of obscene words or socially inappropriate remarks, popularly considered a hallmark of Tourette's, actually occurs only in about 15 percent of cases.

What solidified Tischfield's ambition was a phone call three years ago from Faith Rice, executive director of the Tourette Syndrome Association of New Jersey in Somerville. Rice, a former IBM employee, had gone through her own odyssey in the 1990s, joining the association and building it up after running into problems finding treatment for her own son, now grown.

"I recognized early I wasn't going to be able to cure him," Rice said. "But I could start to change the world around him."

She had read that Rutgers' Human Genetics Institute, a vast storehouse of cells, had won a multimillion-dollar federal grant to fund the storage of DNA samples of families with a host of mental disorders. She wanted to know why Tourette's was not on the list and what could be done to make that change.

"It was like a hand of fate reaching out to me," Tischfield said. "It was a time of my life when I was examining my work and my goals. And I was feeling almost guilty I had not done anything about the disease."

Since that conversation, they have worked together. Part of the \$250,000 in state funds for the New Jersey Center for Tourette Syndrome and Associated Disorders at Rutgers, established by Rice and others, will support the cell repository.

Recently, Rice mailed a questionnaire to 250 families in her organization about whether they would consider donating blood samples for research. The response, she said, was overwhelmingly positive.

To understand these families' willingness to seek answers, Rice said, it helps to comprehend the difficulties of living with the disease.

No one dies from Tourette's, but those who live with it say it can lead to social isolation and wasted lives. "It affects every aspect of a kid's life," Rice said.

Just before a tic occurs, most people with Tourette's are aware of an urge that is similar to the need to sneeze or scratch an itch. They describe it as a buildup of tension that they consciously needed to release. Tics are suppressible but irresistible.

"These kids' lives are miserable," Tischfield said. "For most families, this is a disaster."

Medical treatment can come in the form of medications to treat other disorders often associated with Tourette's syndrome, such as attention-deficit hyperactivity and obsessive-compulsive disorder. Some work well; others do not.

Genetically speaking, Tourette's is viewed as a heterogenous disease, meaning that it involves different groups of genes in different families. Classically homogenous diseases include hemophilia, a rare blood disorder caused by a defect in a single gene. Tischfield suspects that the disorder may occur when one of the main neural connections in the brain that controls impulsivity is somehow perturbed.

Health workers will start to collect blood samples from Tourette's families at the end of this month. They will be frozen and stored in cryogenic tanks at the Human Genetics Institute on Rutgers' Busch campus. As with everything else in the collection, the identities of those contributing samples will be kept confidential.

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