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Gene Discovered in Childhood Language Disorder Provides Insight Into Reading Disorders

The recent discovery of a gene associated with specific language impairment (SLI), a disorder that delays first words in children and slows their mastery of language skills throughout their school years, offers new insight into how our genes affect language development. The finding, published in the [Journal of Neurodevelopmental Disorders](#), is the result of a collaborative team effort headed by Mabel Rice, Ph.D., a University of Kansas professor and NIDCD-funded scientist.



Dr. Mabel Rice reading with a young friend.
Credit: Steve Puppe

The gene, KIAA0319, appears to play a key role in SLI, but it also plays a supporting role in other learning disabilities such as dyslexia. The finding is important for children with SLI and their families, and it is also likely to improve the classification, diagnosis, and treatment of other language, reading, and speech disorders.

SLI affects an estimated 7 percent of 5-6 year olds. Yet it is often overlooked as a diagnosis because children with SLI typically don't have severe communication problems or an obvious cause for the impairment, such as hearing loss. "These children are less likely to start talking within a normal timeframe," says Dr. Rice. "They may not begin to talk until they're three or four. And when they finally do talk, they use simpler sentence structure and their grammar may seem immature." Language impairments such as SLI also appear to increase the risk for reading deficits.

Often childhood language difficulties are seen as only a mild problem, or something kids eventually grow out of, but Dr. Rice says that's not true. "It persists. We know they don't catch up and their limitations in language continue as they move forward in school and then out into the workplace."

Because SLI tends to run in families, scientists suspected that genes played a role. But tying the presence of a specific genetic mutation to SLI, or to any inherited language impairment for that matter, had eluded researchers until recently.

A total of 322 individuals took part in the study, selected from a large pool of children, parents, and other family members participating in an ongoing investigation of the long-term outcomes of children with SLI. Each individual in the study was put through a battery of tests to assess speech, language, and reading skills. Standard diagnostic tests—the same tests that speech pathologists use to diagnose language and learning disabilities—were used to establish measurable behavioral traits that can act as symptoms of SLI, much as how fever is a symptom of the flu.

Using saliva samples to collect the DNA, the team identified a group of candidate

genes—genes that previous studies indicated might have an association with speech or reading disabilities—and looked for mutations that corresponded with SLI's behavioral traits. Dr. Rice and her team scanned millions of letters of genetic code looking for mutations that family members have in common.

They discovered that mutations in one of the candidate genes for reading disability, KIAA0319, had a strong effect on the language traits that are characteristic of SLI, traits that can also be present in dyslexia, some cases of autism, and speech sound disorders (conditions in which speech sounds are either not produced, or produced or used incorrectly).

The next question, according to the researchers, is what does this gene do to affect how we learn language? "It could be a gene that's necessary in the development of the cortex, the area of the brain where we do most of our language processing," says Dr. Rice. "Or maybe it's a gene that's important for setting up neural pathways that are responsible for allowing language to emerge on time. It could be a gene, or one of a family of genes, that sets the stage to make language happen."

Dr. Rice contends that these findings lend support to the idea that difficulties with reading and understanding printed text may be coming from the same genes that influence difficulties in learning language. If this is so, she says, early detection and diagnosis will be the key to helping children with SLI close the reading gap between themselves and their peers. Interventions targeted to the preschool years, she adds, when neural pathways in the brain's language regions are still plastic and open to change, can give preschoolers the chance to develop their vocabulary and language skills in play settings and improve their ability to communicate once they enter school.

Even better, this discovery takes the shame and blame out of SLI. In the past, parents were often blamed for their child's disability and told that they hadn't read to them enough. Children with SLI were called lazy or accused of not working hard enough. Now, with the evidence that SLI is caused by a genetic mutation, parents and children know that talking on time or speaking correctly isn't something that youngsters with SLI can will themselves into doing.

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