Everyone knows Thomas Edison was a brilliant scientist. Most people don’t know he was dyslexic. Kicked out of school because he was “dumb,” the inventor of the phonograph, motion picture camera and light bulb also struggled with words and speech.

UNMC’s Shelley Smith, Ph.D., has studied the relationship between genetics and learning disabilities, including dyslexia, and she may have the answer as to why Edison struggled. A few years ago, Dr. Smith and her collaborators discovered a link between a particular gene and language impairment. The “KIAA0319” gene on chromosome 6 previously had been associated with reading disability. The finding, which has since been replicated by others, shows it can affect both reading and language.

“It had been thought that reading disability, language impairment and speech disorder...
were entirely separate conditions, and they are usually diagnosed separately and treated separately,” Dr. Smith said. “The finding confirms that there are common factors. It also helps explain why some kids have problems in more than one area.”

The director of developmental neuroscience at UNMC’s Munroe-Meyer Institute, Dr. Smith localizes and identifies genetic mutations that influence developmental disorders, primarily language disorders such as dyslexia. This spring, she was named the 2010 Science Laureate, the highest honor for a UNMC investigator.

“Dr. Smith’s diligence and hard work are inspiring and have led to her achieving a global reputation as a leader in 21st-century genetics,” said Tom Rosenquist, Ph.D., vice chancellor for research. “She truly embodies the characteristics we seek in a Scientist Laureate.”

While language involves vocabulary and grammar, and speech involves accuracy of sound production, both contribute to a child’s ability to read. The finding that a certain gene could influence all three abilities suggests a common pathway contributes to overlapping strengths or weaknesses across speech, language and reading.

“Some thought it was impossible to find anything because so many genes influenced these disorders,” she said. “We, and others, continue to show we can find these genes.”

Redefining the conditions to recognize the overlaps could lead to more effective treatment, she said, whether it is through clinical therapy or medication.

Previous studies had not found such overlap. Reading disability genes appeared on certain chromosomes, and language impairment genes had been localized to other chromosomes.

“It made us think that maybe there were fundamental differences between the two disorders, but that seemed unlikely since they were often seen in the same kids,” Dr. Smith said. “Our finding helps to clarify things. We’ve shown that in some cases there really are genetic overlaps between the two disorders.”

“We also found some association with speech problems. To test that completely, we are studying a population of kids who are primarily diagnosed with speech disorder.”

As the genes that influence learning disabilities are defined and the cognitive overlaps between disorders worked out, genetic studies will be able to confirm which diagnoses have common genetic effects and which are distinct.

“It’s likely that the current diagnostic criteria may be modified,” Dr. Smith said. “A better definition of the three disorders should lead to optimal treatment procedures based on deficits that are closer to the underlying causes than clinical symptoms alone.”

Dr. Smith believes other genes will be found that account for all three disorders. She hopes to do further studies to determine how the genes interact in order to better understand how the brain handles reading and language information.

“Next, basic science researchers can determine exactly what these genes do and how they really interact, which will tell us about normal brain development and what goes wrong in these disorders.”

Dr. Smith’s interest in genetics grew out of a thesis project involving a Swedish family with significant dyslexia. “It looked like a dominant trait and in those days those were the types of traits you could find genes for,” she said.

“We naively went into this thinking we could find these dominant families and the gene localization would be easy. We actually did find some families and they serendipitously led us to the right place.”

They published a linkage in 1983, but also realized that the inheritance of dyslexia was much more complex than a simple dominant trait. It wasn’t until technology and computing power advanced that they were able to discover the more-difficult-to-find quantitative trait genes. “Now, everyone is interested in complex genetic traits.”

As technology advanced, so did her research. In the mid-90s, better DNA markers further propelled her work and her group identified a region on chromosome 6, which was quickly replicated by researchers at Yale and Oxford Universities. “Everything just broke loose,” she said. “The work remains intriguing. I’m lucky to have a fantastic group of people in the laboratory (Judy Kenyon, Denise Hoover and James Askew) and great collaborators across the country.”

Dr. Smith also oversees a $10.4 million Center of Biomedical Research Excellence (CoBRE) grant that provides research opportunities as well as career-development and mentoring for junior faculty to jumpstart their careers. She also co-directs the M.D./Ph.D. program, which has grown to 26 students.

While she enjoys traveling, rock climbing and gardening, Dr. Smith’s passion lies with the research and the families being served including one that sends the lab cookies each year.

“I think of science as being really elegant,” she said. “There’s a constant challenge of wanting to see how it works because the answers are lovely.”

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Dyslexia is the most common learning disability with a prevalence of about 9 percent in American school children. Language impairment is the second most common at about 7 percent. Speech disorders rank high as well.