NEWS RELEASE

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FOR THE GOOD HEALTH OF NEVADA

SOM researcher part of team discovery

Lynne Williams, medical school information
July 10, 1996

EMBARGOED UNTIL JULY 11, 6 P.M., EASTERN DAYLIGHT TIME

A team of researchers has discovered that a deleted gene can cause impaired cognitive development in humans. Scientists report that deletion of the gene, Lim-kinase 1, hampers spacial cognition. The findings are the first that point to a molecular basis for a specific human cognitive trait. The team's discovery will be published in the July 12 issue of the journal, Cell.

Colleen Morris, M.D., associate professor of pediatrics and a medical geneticist at the University of Nevada School of Medicine's Las Vegas campus, is part of that team. Mark Keating, M.D., a molecular geneticist at the Howard Hughes Medical Institute (HHMI) at the University of Utah and Carolyn Mervis, Ph.D., a cognitive neuropsychologist at Emory University in Atlanta, are Dr. Morris' collaborators in the group, which has worked together for six years.

The deletion was discovered in individuals who have a disorder known as Williams syndrome. People with this disorder have great difficulty visualizing an object as a set of parts and constructing a replica of the object with those parts. This cognitive ability is known as visuospatial constructive cognition. Williams syndrome individuals cannot copy even simple patterns like a checkerboard consisting of four cubes. As a result, most individuals with Williams syndrome have difficulty with tasks like building a model or assembling a piece of furniture.

People with Williams syndrome have a unique profile of cognitive strengths and weaknesses. Most have mild or moderate mental retardation along with the extremely impaired spatial cognition. In contrast, their auditory short-term memory is often in the

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normal range and the language abilities are relatively good. Most older children and adults speak in grammatical, well-formed sentences, and many have good vocabularies. These people are loquacious, overly friendly to strangers and acquaintances, and overly sensitive to other people's feelings.

One of the keys to the discovery was identifying individuals who had some, but not all, the features of Williams syndrome. Researchers found that these individuals have smaller deletions on chromosome 7 than those with the complete syndrome. They discovered that the loss of one copy of the gene Lim-kinase 1 leads to the impairment described in Williams syndrome.

This is not the team's first discovery. In 1993, the group found that loss of one copy of a gene known as elastin on chromosome 7 was responsible for heart disease and some unusual facial features in Williams syndrome. Elastin provides elasticity to tissues like blood vessels and skin. However, the researchers hypothesized that elastin mutations could not account for the cognitive deficit. Instead, they speculated that other genes adjacent to elastin must also be involved in the deletion that causes this disorder, which led to the latest discovery.

"Individuals with Williams syndrome have many diverse symptoms," says Dr. Morris. "The discovery that a small segment of chromosome 7 was missing was the first step in determining the cause of their problems. Now we are finding that genes in the missing region are responsible for specific characteristics. This is particularly exciting because this gene is important in the function and development of the brain."

The team's research is supported by the Howard Hughes Medical Institute, the National Institutes of Health, and the Williams Syndrome Association.

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