GENE PLAYS ROLE IN BEHAVIOR DEVELOPMENT: Researchers identify cause of Williams syndrome

Lynette Boggs, University of Nevada School of Medicine, 702-383-2638
Dorothy Dart, University of Utah, Eccles Institute of Human Genetics, 801-585-3470

EMBARGO: August 31, 1993, 3 p.m. (PST)

New studies show that the absence of a gene plays a role in Williams syndrome, a developmental disease that causes mental retardation but simultaneously enhances language skills and results in a gregarious, "cocktail party" personality. This new finding provides some of the best evidence to date that a single gene can play a significant role in specific behavior patterns in humans. The study, conducted by scientists at the University of Nevada School of Medicine in Las Vegas and Reno, and the University of Utah, will be published in the September 1, 1993, issue of *Nature Genetics.*

Williams syndrome is a complex condition involving blood vessels, connective tissue and the brain. Mild mental retardation commonly occurs and is accompanied by poor visual-motor integration. As a result, affected individual have problems visualizing a complete picture but instead see only parts. Affected individuals may also have Attention Deficit Disorder. Language development, by contrast, is relatively spared and some elements of speech may be enhanced, particularly the quantity and quality of vocabulary, auditory memory, and social use of language. Many individual rarely forget a name. Because of their engaging personalities, language skills and loquaciousness, mental retardation is often underestimated in children with Williams syndrome.

Colleen Morris, M.D., a clinical geneticist at the University of Nevada School of Medicine, and Mark Keating, M.D., a Utah investigator, led the team of scientists that made these discoveries. Researchers also included scientists from the Eccles Institute of Human Genetics and Howard Hughes Medical Institute at the University of Utah, and researchers at the University of Nevada School of Medicine in Las Vegas and Reno. Research support was provided by the National Heart, Lung and

-more-

Blood Institute; the Williams Syndrome Foundation; the American Heart Association, the March of Dimes and the Utah Genome Center.

"We are proud to be a part of this medical breakthrough and delighted at the prospects for the future," says Terry Monkaba, vice president for the national Williams Syndrome Association. Several members of the organization, comprising more than 2,000 families, participated in this research study. "This discovery and the results of subsequent studies will go a long way toward helping professionals understand the many facets of Williams syndrome which will, in turn, help make our mission a reality and the future brighter for our children," Monkaba says.

Earlier this year, scientists found that a mutation in the elastin gene was responsible for Supravalvular Aortic Stenosis (SVAS), a congenital heart malformation commonly found in Williams syndrome patients. It was this find that led the group to study the elastin gene in individuals with Williams syndrome. Scientists discovered that individual having Williams syndrome inherited only one intact elastin gene. The other elastin gene was either absent or deleted. *(A chromosome deletion, a condition that occurs during reproduction, is the loss of part of a chromosome. During the development of human eggs and sperm, genetic material or DNA that form the blueprint for new life is duplicated. Sometimes this duplication process is incomplete, resulting in the loss or "deletion" of genetic material. If the missing material is important, as in the case of Williams syndrome, development is altered).*

This discovery will improve diagnosis of both SVAS and Williams syndrome and may lead to new treatments for these conditions. SVAS is an abnormality of the blood vessels which may lead to heart failure and even death. SVAS occurs in approximately 1 in 25,000 births and affects both children and adults. The disease often worsens with age. Currently, the only treatment for SVAS is surgery and there is no cure for many of the abnormalities of Williams syndrome. Further studies on the elastin and neighboring genes will give new understanding to the role genes play in behaviors and development. Studies may also lead to better understanding of the causes of deletions in this and other disorders.

###