Gene plays a role in Williams syndrome

Colleen Morris, M.D., a geneticist with the School of Medicine, helped lead the team of researchers who discovered that the absence of a gene plays a role in Williams syndrome. The syndrome is a complex condition involving blood vessels, connective tissue and the brain, and results in mild mental retardation. However, language development is usually spared, resulting in an engaging personality, with good quantity and quality of vocabulary, auditory memory and social use of language. Because their language skills are so good, mental retardation is often underestimated in children with Williams syndrome.

Dr. Morris and Mark Keating, M.D., an investigator from the University of Utah, led the team of researchers who made the discovery. The team also included investigators from the Eccles Institute of Human Genetics and the Howard Hughes Medical Institute at the University of Utah.

Earlier this year, the same group of scientists found that a mutation in the elastin gene was responsible for a congenital heart malformation (Supravalvular Aortic Stenosis) commonly found in Williams syndrome patients.

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