# SOUTHWESTERN NEWS

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## GENE LOCATED FOR RARE FAT DISORDER

DALLAS – September 14, 1999 – An international team led by researchers at UT Southwestern Medical Center at Dallas has located the gene for a rare fat-developmental disorder called congenital generalized lipodystrophy.

Lipodystrophy disorders cause selective loss of body fat and are associated with the same metabolic complications as obesity – lipid disorders and diabetes.

The researchers reported finding the location of a gene that causes congenital generalized lipodystrophy in the September issue of the *Journal of Clinical Endocrinology and Metabolism*. The work was done by Dr. Abhimanyu Garg, professor of internal medicine at UT Southwestern, former UT Southwestern faculty member Dr. Anne Bowcock, now at the Washington University School of Medicine in St. Louis, colleagues from UT Southwestern and researchers from the National Institutes of Health (NIH), Pakistan, Turkey and the United Kingdom.

Congenital generalized lipodystrophy is the most severe form of the lipodystrophies. It is characterized by a lack of body fat from birth. Patients appear extremely muscular and have little fat. They develop severe insulin resistance, and diabetes occurs during teen-age years. They also have high triglyceride levels and low levels of HDL cholesterol– the "good" cholesterol.

The researchers collected genetic data on 17 families from around the world with congenital generalized lipodystrophy. The gene for the disorder was found on the long arm of chromosome 9, and studies are under way to isolate the gene. Research indicates a second gene may be implicated in congenital generalized lipodystrophy. The second gene has not yet been located or identified.

This breakthrough comes 45 years after the initial description of the disorder.

"Locating a gene for congenital generalized lipodystrophy certainly opens doors for investigation into common obesity," Garg said. "If we can find out why these genetic defects cause loss of fat and complications associated with insulin resistance, we hope to be able to learn more about common types of obesity, such as truncal obesity and how it leads to insulin

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resistance, diabetes and lipid problems."

The UT Southwestern researchers last year located the gene for another form of lipodystrophy - familial partial lipodystrophy - on chromosome 1.

Other researchers in the study include UT Southwestern Center for Human Genetics colleagues Ross Wilson, a research associate, and Robert Barnes, a research analyst; Dr. Elif Arioglu and Dr. Simeon Taylor of the NIH; Dr. Zohra Zaidi of Zinnah Postgraduate Medical Center in Karachi, Pakistan; Dr. Figen Gurakah and Dr. Nurten Kocak of Hacettepe University Faculty of Medicine in Turkey; and Dr. Stephan O'Rahilly of the University of Cambridge in England.

Garg will continue researching both forms of lipodystrophy with the aid of a four-year NIH grant he was awarded in the spring.

Grants from the NIH, the National Center for Research Resources, the General Clinical Research Center at UT Southwestern and Southwestern Medical Foundation supported the research.

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