WAYNE STATE UNIVERSITY
news release

Release Date: September 26, 2007
Contact: Julie O'Connor
Voice: (313) 577-8845
E-mail: ag2712@wayne.edu
Fax: (313) 577-3626

Professor at Wayne State University cracks code for genetic eye disease

DETROIT— For over 18 years, Jayne S. Weiss, M.D., professor of Ophthalmology and Pathology at Wayne State University's School of Medicine, has been studying a rare inherited corneal disease, Schnyder's crystalline corneal dystrophy (SCCD). With her collaborators, she has discovered the abnormal gene which causes this visually disabling disease. The lipid processing gene that causes SCCD, UBIAD1, should give a window into understanding other diseases involving abnormal lipid accumulation such as hypercholesterolemia or atherosclerosis.

SCCD causes visual loss by resulting in progressive opacification of the front layer of the eye, the cornea. This progressive clouding of the cornea which causes gradual visual loss, results from an abnormal deposition of cholesterol and fatty acids (phospholipids) in the cornea. In the 1990's, Dr. Weiss' work with Dr. Howard Kruth's research group at the National Institutes of Health, revealed that the pathology of the corneal deposits in SCCD was similar to atherosclerosis, a condition caused by cholesterol in the bloodstream.

Subsequently studying over 35 families with SCCD around the world, she found that the disease resulted in significant visual disability with 50% of affected individuals above the age of 50 having undergone corneal transplant surgery because of visual loss. In addition, two-thirds of patients with SCCD also have hypercholesterolemia. When Dr. Weiss first began studying this disease eighteen years ago, it was her hope that finding the gene for SCCD might lead to further understanding about systemic cholesterol metabolism and possibly atherosclerosis.

--more--

Dr. Weiss' research is the largest and longest-running patient study of SCCD, making her the world's leading expert on the disease. What began as a coincidence in 1986 with three patients diagnosed with SCCD – all three with the same surname or maiden name – has led to a collection of more patients with this disease than had ever been studied or reported in medical literature. With the large volume of patients in the study, and the length of the study, Dr. Weiss has been able to clearly paint a picture of what happens to patients with this disease over a long span of time.

The surname discovery led to the recognition that a large portion of patients diagnosed with the disease are of Swede/Finn descent. This allowed Dr. Weiss to zero in on recruiting patients for her study.

"My goal 18 years ago when I began this work was to not only be able to tell a patient that they were developing a disease that would result in visual loss, but more importantly to discover a way to prevent disease progression," commented Dr. Weiss. "Discovery of this gene is the first step of that goal. This truly has been an international collaboration with many groups within the United States and abroad, starting with our first discovery of the abnormal chromosome in 1995 with researchers at MIT, to the assistance of doctors around the world who enrolled their patients, to the work narrowing the genetic interval with Drs. Kuivaniemi and Tromp at Wayne State's Center for Molecular Medicine and Genetics and the final localization of the gene and the mutation analysis with researchers at Transgenomic in Maryland and NIH."

"From the start, I thought understanding the genetic defect in this disease would give us a window into further understanding of lipid metabolism," added Dr. Weiss. "This appears to be true as the gene that causes SCCD is involved in lipid metabolism. I hope further work will shed some light on lipid metabolism in the rest of the body and ultimately translate into preventative therapy. My hope

is that my first hunches 18 years ago were correct and that understanding the gene abnormality is a link to understanding the genetic and metabolic causes of atherosclerosis and problems with lipid metabolism."

"Dr. Weiss has made a tremendous mark on the study of Schnyder's Crystalline Corneal Dystrophy," commented Dr. Gloria Heppner, associate vice president for research at Wayne State. "She has made a poorly understood disease into one that is more easily diagnosed and predictable due to the length of her research study. By cloning the gene that is linked to the development of this disease, Dr. Weiss will now be better able to focus on drug discoveries which ultimately may lead to a non-invasive cure for Schnyder's."

Dr. Weiss, a resident of Bloomfield, Michigan, is also a sought-after expert in the area of refractive surgery. She was the former chair of the FDA Ophthalmic Devices Panel and is presently chair of the entire American Academy of Ophthalmology (AAO) Basic and Clinic Sciences Course (BCSC) on Refractive surgery, editing and serving as one of the authors on the first BCSC book on the subject used to teach every refractive residency programs in the United States. She is the past recipient of the AAO Secretariat Award and Honor Awards, and the FDA Service Advisory Award.

# # #

Wayne State University is one of the nation's pre-eminent public research universities in an urban setting. Through its multidisciplinary approach to research and education, and its ongoing collaboration with government, industry and other institutions, the university seeks to enhance economic growth and improve the quality of life in the city of Detroit, state of Michigan and throughout the world.