



Uniformed Services University
of the Health Sciences
4301 Jones Bridge Road
Bethesda, MD 20814-4799

Release No.11-07-80

July 27, 2011

Contact: Ken Frager, Office of External Affairs

Voice: 301-295-3981 **Cell:** 240-281-2738

Email: kenneth.frager@usuhs.mil

For Immediate Release:

NHGRI researchers announce finding gene mutation that causes Proteus Syndrome

Bethesda, Md.: After a 15 year search, scientists at the NIH, with support from Dr. Thomas Darling, associate professor, Department of Dermatology at the Uniformed Services University of the Health Sciences, have found the gene mutation that causes Proteus syndrome, a rare disorder that causes tissue and bone to grow out of proportion to the body. The study describing the AKT1 gene mutation was published in the online edition of The New England Journal of Medicine.

According to Dr. Darling, who characterized the skin findings in these patients, "The skin provided important clues that Proteus syndrome was caused by mosaicism – the presence of two populations of cells in the body that differ in genetic composition –and this work confirmed this hypothesis. More importantly, the identification of this mutation will lead to improved diagnosis and hopefully an effective medical treatment."

Proteus syndrome gained wide public attention in 1980, through the movie "The Elephant Man," about a 19th century Londoner whom experts believe may have suffered from the disease.

The full study, along with material distributed by the National Human Genome Research Institute (NHGRI), part of the National Institutes of Health, is available at <http://www.genome.gov/27544866>.

The Uniformed Services University of the Health Sciences is the nation's federal health sciences university. USU students are primarily active-duty uniformed officers in the Army, Navy, Air Force and Public Health Service who are being educated to deal with wartime casualties, emerging infectious diseases, disasters, and other public health emergencies. The vast majority of the university's more than 4,500 physician and 500 advanced practice nursing alumni are supporting operations in Iraq, Afghanistan and elsewhere, offering their leadership and expertise. For more information, visit www.usuhs.mil.