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.

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