Scientists Unravel Mystery of People with No Fingerprints

September 13 2006

Researchers at the Technion-Israel Institute of Technology have succeeded in unraveling the genetic basis of two rare congenital diseases in which afflicted persons have no fingerprints. The results will be published in the October 2006 issue of the *American Journal of Human Genetics*.

A team led by Professor Eli Sprecher of the Technion Faculty of Medicine found that both Naegeli syndrome and dermatopathia pigmentosa reticularis (DPR) are caused by a specific defect in the protein known as keratin 14.

The defect induces the body to mark cells in the upper layers of the skin for death. As a result, people with these afflictions lack fingerprints, as well as the ability to perspire normally. They also suffer from thickening of the palms and soles and may have developmental anomalies of the teeth, hair and skin.

Biological samples from Naegeli and DPR patients has been available for more than 20 years, but only recently did Technion graduate student Jennie Lugassy discover that the disease results from the abnormal function of keratin 14. Defects in this protein were known for years to be associated with a number of skin diseases with very different manifestations, so it had never been considered as a cause of Naegeli syndrome and DPR.

According to Sprecher, this new discovery was made possible by efforts

initiated more than 20 years ago by former Professors Gabriele Richard (now with GeneDx, Inc. in Gaithersburg, Md.) and Peter Itin (University Hospital in Basel, Switzerland). The pair assembled an extraordinarily large group of individuals affected with these rare diseases; 25 individuals from five extended families from Switzerland, the United States and the United Kingdom participated in the study.

The researchers are now trying to understand the exact connection between the unusual features of the disease – such as the absence of fingerprints – and enhanced programmed cell death. According to Sprecher, the data from such research could have implications for many skin disorders.

Source: American Technion Society