Researchers identify genetic defect that leaves some without fingerprints

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Like DNA, fingerprints are unique to each person or set of identical twins. That makes them a valuable identification tool for everything from crime detection to international travel. But what happens when the tips of our fingers are missing those distinctive patterns of ridges?

It's not the premise for a science fiction movie, but a real-life condition known as adermatoglyphia. It's also known as "Immigration Delay Disease," because affected individuals experience difficulty in passing through security or checkpoints where fingerprint identification is required. Now Prof. Eli Sprecher from Tel Aviv University's Sackler Faculty of Medicine and the Tel Aviv Sourasky Medical Center has identified the genetic mutation responsible for this unusual condition.

Though adermatoglyphia itself is extremely rare, defects that stem from any one genetic mutation give researchers unique insights into the most complex biological phenomena, such as the consequences of lacking a single protein.

The findings have been published in the American Journal of Human Genetics.

Baffling border control

"Immigration Delay Disease" came to the attention of the medical community when it did just that — delay the attempts of one Swiss

woman to cross the border into the United States, which requires that non-citizens be fingerprinted upon entry. Border control personnel were mystified when the woman informed them that she was unable to comply.

Though an exceptionally rare condition — only four documented families are known to suffer from the disease worldwide — Prof. Sprecher was inspired to delve deeper into the causes of the condition, which, in addition to causing an absence of fingerprints, also leads to a reduction in the number of sweat glands. Abnormal fingerprints can also be a warning sign of more severe disorders.

Scientists know that fingerprints are fully formed 24 weeks after fertilization, and do not change throughout our lives. But "the factors underlying the formation and pattern of fingerprints during embryonic development are largely unknown," says Prof. Sprecher. He adds that it isn't only fingertips that have patterned skin — palms, toes, and the soles of the feet also feature these ridges, called dermatoglyphs.

To determine the cause of this rare condition, the researchers did a genetic analysis of the Swiss family, nine of whom have no fingerprints. They compared the genes of those with adermatoglyphia and those without to identify where the genetic alteration lies. They discovered that a skin-specific version of the gene SMARCAD1 has a regulating factor on fingerprint development. The group that presented with adermatoglyphia, Prof. Sprecher explains, were found to have decreased levels of the short skin-specific version of the gene.

An inconvenience, but little more

Now that this gene has been identified, researchers will be able to further investigate how SMARCAD1 regulates fingerprint development. While adermatoglyphia may be intriguing, and can certainly be

problematic for border security, it's also non-threatening. Despite the minor issue of the hand's inability to produce sweat, says Prof. Sprecher, those affected do not otherwise suffer.

This research was carried out in collaboration with Dr. Janna Nousbeck of the Tel Aviv Sourasky Medical Center and Prof. Peter Itin of the University Hospital at Basel, Switzerland.

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