

Research finally reveals reason for perplexing red skin disease ichthyosis

October 7 2016, by Marla Paul

The children and adults travel from as far as Australia, Africa and Mongolia to see her, because she is one of the few dermatologists in the world who specializes in the rare and perplexing genetic disease that causes their skin to be disfigured by redness and dark scales across their entire bodies.

There aren't any great treatments for the disease with the hard-to-pronounce-name, ichthyosis.

But now Dr. Amy Paller, who has spent 30 years researching the disorder, for the first time has found the reason the disease causes the red skin and has a promising biologic drug to begin testing soon in clinical trials.

In a new study published in the *Journal of Allergy and Clinical Immunology*, Paller, the Northwestern Medicine chair of dermatology, together with Dr. Emma Guttman-Yassky of Mount Sinai Medical School, discovered that an arm of the immune system – the Th17 pathway – in these patients is way too active, and the higher its activity, the worse the <u>disease severity</u>.

Paller is about to launch a clinical trial to test a new biologic (a cuttingedge drug) for it, with the goal of targeting and calming down this pathway.

In ichthyosis, the skin barrier is abnormal, so the skin is inflamed, dry



and scaly.

"These patients are tremendously disfigured by this skin disease," said Paller, also an attending physician at the Ann & Robert H. Lurie Children's Hospital of Chicago. "It can be painful, itchy and easily gets infected. They may have trouble using their hands and walking."

Patients also have trouble sweating, so it's hard for kids and adults to participate in sports.

The drug Paller will test, secukinumab, has already been highly effective in psoriasis, a more common skin disorder with an increase in this Th17 pathway, leading to inflammation and scaling. And with Paller's new discovery, she thinks the drug could be even more helpful in ichthyosis because the overactive immune pathway was actually more strongly correlated with ichthyosis than with psoriasis.

More information: Amy S. Paller et al. An IL-17–dominant immune profile is shared across the major orphan forms of ichthyosis, *Journal of Allergy and Clinical Immunology* (2016). DOI: 10.1016/j.jaci.2016.07.019

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