

Patient Advocacy and Patient Perspective of Skin Disease

Malinda Heuring

National Foundation for Ectodermal Dysplasias, Mascoutah, Illinois, USA

Growing up with fair skin and a face full of freckles, I thought I understood the burden of skin disease. I sunburned easily and therefore could not spend my summers at the pool with my friends. And those freckles—they were the cause of countless teasing that usually was followed by tears on my part. It was not until recently through my job as the Director of Education for the National Foundation for Ectodermal Dysplasias (NFED) that I witnessed first hand the true burden of skin disease on families who are affected by Hay-Wells syndrome with severe skin erosion.

Hay-Wells (also known as ankyloblepharon ectodermal defects-clefting, AEC) is one of the 150 ectodermal dysplasia syndromes that is characterized by fused eyelids, sparse hair, cleft lip and/or palate, conductive hearing loss, malformed nails, nonfunctioning sweat glands, and small, malformed teeth. Some individuals affected by Hay-Wells also have skin erosion on their hands, feet, or scalp. Other affected babies have been born with skin erosion on 70% of their body. For the latter, the condition is life-threatening.

For most of these individuals, it is not uncommon to apply lotion up to 20 times a day to their dry, cracked hands or to soak their socks in water before taking them off so that more skin does not come off their eroded feet. It is a part of their daily lives. Some use wheelchairs, not because they cannot walk, but because the skin on their feet is eroded so badly it is too painful to walk. The pain can be so intense that to change the dressings on an affected baby, the baby must be put under anesthesia.

Being denied for medical insurance is also common among these families. The out-of-pocket medical costs can be enormous. If families do have medical insurance, it often does not cover expenses such as dressing changes and lotions, which are significant.

The social stigma attached to a “difference” is often devastating to one’s self-esteem. Many days of school or work are missed for frequent doctor appointments. The effect of skin disease can be seen on the entire family unit. Siblings can feel left out or embarrassed and parents spend many sleepless nights changing dressings and worrying about their children. What is so amazing about many of these affected individuals is that they do not see their disorder as a “burden,” but as an inconvenience in their daily lives.

Hosted by the Department of Dermatology at St. Louis University School of Medicine in St. Louis, Missouri, in September 2003 and funded by the NFED, the Skin Erosion Workshop was the first of its kind in medical history to examine the problem. It was also the largest gathering of individuals affected by Hay-Wells syndrome ever. The NFED researched many funding possibilities including NIH funding, grants, and pharmaceutical support. Although a small

amount of funding was secured from the pharmaceutical industry, the NFED was the main financial supporter of this workshop. It was only through a lay advocacy group that this workshop was accomplished.

The clinical significance of this workshop is still to be seen. Participating physicians and scientists have now more clearly described the nature and impact of skin erosions, which could ultimately lead to accelerated wound healing. Clinicians who attended the workshop came thinking that they understood the severity of this disease and the skin erosion involved. Many left the workshop realizing that there is much more to learn and more to the disease than they first thought. Families, who had tried everything to relieve some of their burden, now have new avenues to explore. These families left the workshop feeling good about themselves and the contributions that they had made to medical research.

The NFED is not the only advocacy group dedicated to providing support to relieve some of the burden of skin diseases. The Coalition of Skin Diseases (CSD) is a group of 23 advocacy groups that work collectively to combat the burden of disease. As a group, these organizations have funded research in excess of more than \$16.5 million over the past 15 years CSD members are in contact with more than half a million individuals affected by skin diseases. They offer affected individuals the latest findings on research, treatment options, and general information on the diseases. Most importantly, they offer emotional support to those affected.

It is through the support and services that lay advocacy groups provide that individuals such as these affected by Hay-Wells accept themselves. Advocacy groups provide a connection among affected individuals. It is almost daily that advocacy groups hear the words “Thank you, I never knew there was someone else like me.” Across the board, all coalition members, representing the 23 skin disease advocacy groups, agree that physical and economic burdens are great and acknowledge that one of the largest burdens their members face in regard to skin disease is feeling as if they are alone in their experience. The patient advocacy groups serve to unite individuals so that no one goes through this alone. Although the CSD cannot offer a cure for all of the skin diseases, they open the lines of communication for individuals to share resources, offer advice, and provide support for one another. The patient advocacy skin disease groups are the conduits by which individuals with skin diseases find each other and in turn find their broader families.

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