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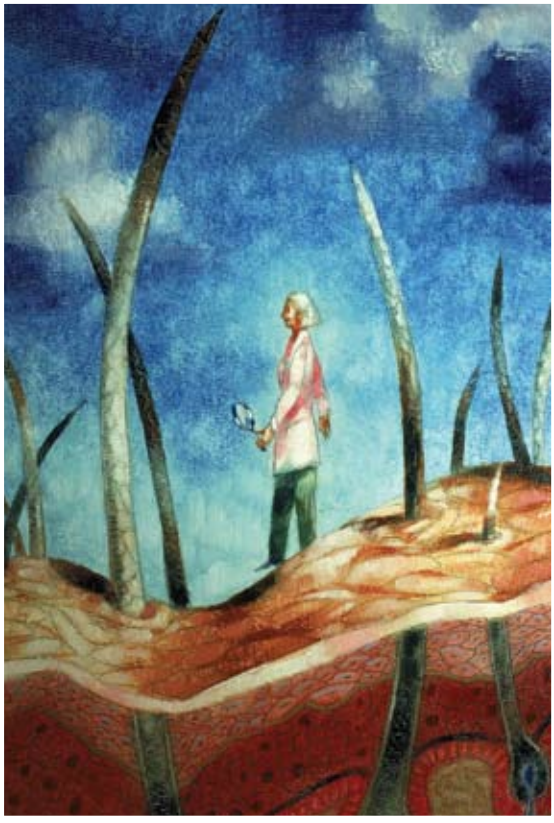

SUPPLEMENT for the Hannah's Hope Fund

Official Journal of the Society of Dermatology Physician Assistants

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From The Patient's Perspective
Our Mission to Save Our Daughter's Life

By Lori Sames

"Your daughter has Giant Axonal Neuropathy. This is a very rare, recessive, neurodegenerative disorder. I'm sorry to tell you, life expectancy is typically the second or third decade." This is what we were told in March of 2008 about our precious four-year-old daughter, Hannah.

In August of 2006, when Hannah was two and a half years old, my mother noticed her left arch seemed to role inward when she turned a corner. For the next nine months, a podiatrist, orthotist, and her pediatrician assured us that she was fine even though both arches were collapsing and her gait had become awkward. By June of 2007, we were convinced Hannah had something seriously wrong with her. We sought the help of a pediatric neurologist and a geneticist who collaborated and ordered several different rounds of testing over the next six months. When everything came back normal, we had a follow-up appointment with Hannah's neurologist to discuss the next steps. He strongly suspected she had some type of peripheral neuropathy because she didn't have a reflex response from the knee down. He entered the exam room with a large textbook in hand and flipped to the 'Neuropathy' section. There was a photograph of a boy who looked to be around eight years of age and had ankle foot orthotics that rose to just below the knee. The boy had very kinky, curly looking hair. The doctor froze. He began reading out loud. This boy had ataxia, low muscle tone, elastic joints, low nerve conduction velocity scores, abnormally kinky, frizzy hair that lacked luster, and pale, dry skin. He had stumbled upon Giant Axonal Neuropathy (GAN), a genetic disorder that will eventually take away Hannah's ability to speak and swallow, and render her a quadriplegic, dependent on a feeding tube and ventilator to breathe before dying of pneumonia.

"Hannah's skin has a tendency to be very dry and her hair is kinky and frizzy. Like many other children with GAN, these cutaneous findings can be an early clue for providers that there may exist an underlying genetic abnormality."

Hannah is our youngest of three beautiful daughters; Madison is ten years old and Reagan is eight. Hannah was a completely healthy, happy, and strong baby who potty trained and began speaking early. Just a few weeks before Hannah's invasive testing at Columbia Presbyterian, I read my journal where I kept track of milestones and funny, cute things our girls were saying and doing. I read the following entry: "Hannah will be fourteen months old next week. She's running and bounding along all of the furniture. Bet she'll be walking independently within a week." I broke down. I knew there was something terribly wrong with Hannah and feared for her life. We had just celebrated her fourth birthday and her run had become a staggering walk. She was dragging her toes as she pulled them forward upon ambulation. She used exaggerated arm movements to help her balance when walking.

A sural nerve biopsy revealed GAN. Every individual has, on average, four to six mutated recessive genes. My husband and I had each passed our damaged, recessive copy to Hannah's GAN gene, a gene responsible for expressing an intracellular protein called gigaxonin. The role of gigaxonin is to degrade its binding partners, which are neurofilament proteins. In absence of functional gigaxonin, these proteins are toxically accumulating on the axon portion of the nerve causing densely packed, disorganized, giant axons. This prohibits the transmission of nerve signals from the central nervous system to the peripheral nervous system and causes progressive nerve death resulting in this devastating disorder. The gigaxonin protein has a role in the function of skin and hair filament. Hannah's skin has a tendency to be very dry and her hair is kinky and frizzy. Like many other children with GAN, these cutaneous findings can be an early

From The Patient's Perspective

clue for providers that there may exist an underlying genetic abnormality.

Paralyzed by unexplainable grief, my husband Matt and I struggled to function. I don't think we told our parents for a few days. I kept saying to Matt, "We have to feel thankful we have time to try to save her. She just turned four!" Refusing to accept this horrific fate for Hannah, we formed Hannah's Hope Fund, a 501(c)(3) public charity devoted to raising funds for a treatment and cure of GAN. We are the only organization in the world for this devastating disorder. At the time Hannah was diagnosed, GAN was not being researched in the US. In August of 2008, we held the first ever GAN symposium. Twenty research scientists from around the world attended the symposium and discussed what was known about GAN to date and prioritized possible therapeutic approaches. Thankfully, scientists know exactly what causes GAN. The causal gene was identified in 2000, allowing researchers to focus on therapy development.

Between May and December 31 of 2008, family, friends and total strangers helped us raise almost five hundred thousand dollars. There are two therapeutic approaches being considered and a clinical trial is scheduled to begin by fall 2011. More money will be neededbe needed to sustain this research and to pay for the treatment of these children and young adults during a clinical trial. The Doris Buffett 'Sunshine Lady Foundation' has agreed to match all funds raised through February 15th, 2010, up to five hundred thousand dollars. Please spread the word of our life saving mission by giving our website address to everyone you know and asking that they do the same.

www.hannahshopefund.org

Precious and innocent lives depend on us all.

Lori Sames *is Hannah's mother and the Executive Director of Hannah's Hope Fund.*

TAKE HOME POINTS for DERM PAs:

By Steven K. Shama, MD, MPH

- We were taught over and over again during our pediatric training, when examining a child who is too young to be verbal, that we should always listen to the parents. This was the case with Hannah's parents when they knew something was not right with their child, almost a year before the diagnosis was made. Listen...
- The skin is often a reflection of the internal environment of the body. When examining a patient who has findings that cannot be explained at that moment, like kinky, fizzy hair and pale, dry skin, never forget those findings. Never give up on making *all* of your clinical findings make sense. Never mind asking a colleague or putting off your final thoughts on an examination until you have checked further. You might just help parents like Hannah's understand what is happening to their child and perhaps save a life and/or make the remaining years of a patient's life that much more precious.

Dermatology Physician Assistants

Physician Assistants (PAs) in dermatology play a number of varied and vital roles.

PAs are medical providers licensed to practice medicine with physician supervision. From patient care and education, to skin surgery, treatment of chronic skin conditions, and cosmetic procedures, PAs are dynamic members of the healthcare team. PAs practice in every medical and surgical specialty and have been collaborating with dermatologists for 30 years, providing a wide variety of services. These include diagnosing, prescribing medications, ordering and interpreting lab tests, wound suturing, and medical or surgical treatment of a wide variety of clinical diseases. As with all PAs, dermatology PAs are legally and ethically bound to practice only under physician supervision.

PAs are trained in intensive, accredited education programs.

Because of the close working relationship that PAs have with physicians, PAs are educated in the medical model designed to mirror and complement physician training. PAs take a national certification examination and to maintain their certification, they must complete 100 hours of continuing medical education every two years and take a recertification exam every six years. Graduation from an accredited PA program and passage of the national certifying exam are required for state licensure.

How a PA practices dermatology varies with training, experience, and state law. In addition, the scope of the PA's responsibilities corresponds to the supervising physician's scope of practice. In general, a PA will see many of the same types of patients as the physician. Referral to the physician, or close consultation between the PA and physician, is based on the dynamic relationship between the physician and PA.

The Society of Dermatology Physician Assistants (SDPA) is a non-profit professional organization, composed of members who provide dermatologic care or have an interest in the medical specialty of dermatology. Fellow members provide medical services under the supervision of a board certified dermatologist.

More information can be found at **www.dermpa.org** and **www.aapa.org**.