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**PATIENT
PROFILE**



From left, XLHED patients Philip and Nolan Pond, with their brother Dylan and parents, Beth and Michael.

Edimer delivers hope for XLHED patients

BY MEAGHAN CASEY

Beth Pond understands all too well the challenges of living with X-linked Hypohidrotic Ectodermal Dysplasia (XLHED).

Her brother, Steve, and two of her sons, Nolan (age 16) and Philip (age 15), are affected by the rare orphan disease that causes a range of symptoms, including lack of sweat glands, poor temperature control, respiratory problems, sparse hair and missing and malformed teeth. There are a number of secondary features of XLHED that may include severe dry eye, eczema, asthma and dry mucous membranes in the mouth and nose.

In the early years of life, XLHED-affected individuals are at risk for severe life-threatening medical complications, most often related to their inability to sweat, leading to hyperthermia. Pond had a second brother who likely succumbed to those complications when he was only six weeks old.

“He died on a hot, summer day while taking a nap,” said Pond. “Doctors said it was SIDS, but we think that he had XLHED and overheated. It’s important to understand that XLHED can be life-threatening and deadly, not just a host of medical and dental needs.”

Fortunately, through awareness, her sons have been able to manage the ramifications of a lack of sweat glands.

“Growing up, the boys always sought out cool places and things like hardwood floors and the refrigerator,” said Pond, a Hubbardston resident. “They’d be in shorts until December, and they had to be in air-conditioned places as much as possible. They could only go to the park on cold or rainy, cloudy days.”

Later, it became more difficult to control the accommodations in the boys’ school or on the bus. Pond advocated for their elementary school to install air conditioners in their classrooms. She and her husband, Michael, also helped them through the hurdle of outdoor sports. Both boys played

baseball and soccer, while seeking out cool relief.

“Michael would volunteer to coach and we would be on the sidelines spraying them with water,” said Pond. “We always had coolers full of ice, spray bottles, drinks and wet shirts for them to put on.”

Nolan also played ice hockey and both boys have always swum in the summers. Today, they’re avid snowboarders.

Yet, even overcoming the challenges of poor temperature control, their path in life has not been an easy one. As XLHED patients grow older, the shift often focuses to loss of hair and chronic skin and dental issues, with the associated medical, emotional and self-esteem ramifications.

“It can be devastating for these children, especially at this age, in their teens,” said Pond. “I’m glad they’re able to turn to their uncle. He’s able to talk to them about specific things like dentures and personal issues relating to the teenage years and relationships, because he’s already gone through it. It’s really important that he give them hope.”

Because the average adult tooth count in XLHED patients is only six, dentures may be prescribed as early as age 2. With only four teeth at age 3, Nolan was fitted for his first pair. The family’s insurance company initially denied coverage stating that it was cosmetic, but Pond fought it and won. She later initiated a bill for Massachusetts health insurance companies to cover dentures and other medically necessary procedures for XLHED-affected individuals, to ensure other families would not have to go through the same thing.

“I brought my children into this world, and I promised myself that I’d be active advocating for them and other children like them,” said Pond. “It’s very therapeutic for me to be doing whatever I can.”

Pond herself is an affected carrier of XLHED. Though males are affected by X-linked recessive disorders much more

frequently than females, females with one altered copy of the gene may experience some features of the condition. Pond had pegged teeth that had to be filed down, as well as fewer sweat glands. As a carrier, she knew there was a 50/50 chance a son of hers would be affected by XLHED. Her oldest son, Dylan (age 17), beat those odds, but she recognized the signs right away when Nolan was born.

“He had extremely dry skin and no dental ridge at all,” she said. “I knew right away. But unless you know what you’re looking for, it’s difficult to make a clinical decision and start managing the symptoms early on. That’s why it’s so important to educate people and raise awareness about this condition.”

Pond serves as a regional family liaison for the National Foundation for Ectodermal Dysplasias (NFED) and believes strongly in creating a network of communications with other families.

“I know a lot of families who, after their first child is born with HED, don’t have any more children and that child is all alone,” said Pond. “Families find support and friendship from other families who are going through the same thing.”

In addition to serving as a liaison for the past five years, Pond also served on the board of the NFED for six years. Through her involvement with the foundation, she was introduced to Cambridge-based Edimer Pharmaceuticals – a company dedicated to delivering a significant and durable improvement in the health and quality of life to future generations affected by XLHED. In February, Edimer announced that it will provide a grant to the NFED to help defray costs associated with the foundation’s annual Family Conference.

“Edimer applauds the efforts of the NFED and supports programs that encourage ongoing communication as we work to develop a treatment for XLHED,” said Neil Kirby, President and CEO of Edimer.

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A DYING WISH

Adriana Jenkins was a publicist and a beloved member of the Mass. life sciences community. She wrote this piece for Forbes magazine shortly before she passed away in February. It is reprinted with permission of Forbes Media.

BY ADRIANA JENKINS

I am dying. We will all die someday, but my expiration date is sooner than most. At age 41, I am facing my second recurrence of cancer. I was first diagnosed with an advanced and rare type of breast cancer in 2001. This led to a large tumor in my brain last year. Now the cancer has spread to my spinal fluid, which will likely seal my fate within weeks.

At my initial diagnosis I participated in a clinical trial evaluating Herceptin, a so-called personalized medicine (PM) drug targeting a mutation believed to be driving my cancer. While most cancer drugs work by targeting mechanisms common to all cancers, such as cell division, PM drugs hit specific genetic mutations driving the cancer's spread. They typically come paired with a diagnostic test to select the subset of patients most likely to benefit. The idea is to get the right drug to the

right patient. (I know about this because I've worked in biotech for 15 years.)

Herceptin likely extended my life by at least nine years. It has had similar benefits for many thousands of patients around the world. It is an amazing example of how the healthcare industry can develop personalized cancer treatments.

But Herceptin, invented at Genentech (now part of Roche), might never have made it to market if it had been tested on all breast cancer patients. The drug aims at the 25% of breast cancer patients whose tumors express high levels of a protein called HER2. Because Genentech tested the drug on this narrower subset of patients, the trials worked. Patients lived longer. Today Herceptin has nearly \$6 billion in annual sales.

Despite Herceptin's success, the pharmaceutical industry seems loath to focus on developing other PM drugs, which often means reducing potential profits. After spending \$1 billion over 15 years to develop a drug, companies are eager to recoup their money fast. The business side of the equation wants as many people to take the drug as possible, whether it's effective or not. I worked at a company whose CEO championed

personalized drugs in public statements. But its first product was not a PM drug. As a public company, it got too much pressure to become profitable for it to further reduce an already small patient population with a diagnostic test.

The result of the focus on testing cancer drugs on all patients is painful trial and error. Chemotherapy is prescribed with no guarantee of effectiveness and can cause wretched, and sometimes fatal, side effects. But cancer patients like me don't have time to waste. How do we convince drugmakers to focus their shrinking R&D budgets on this area of scientific discovery?

One idea is to create an incentive for drugmakers comparable to that in the Orphan Drug Act. Passed in 1983, it encourages companies to develop drugs for diseases that have a small market (fewer than 200,000 patients in the U.S.). Under the law, companies that develop such a drug may sell it without competition for seven years, in addition to often receiving quicker "fast track" regulatory review. This has become such a profitable pursuit that most drugmakers are pursuing at least one "orphan" drug.



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A comparable law could push drugmakers to develop PM drugs for cancer and other deadly ailments. It could combine additional market exclusivity with assurance of accelerated regulatory review. The Orphan Drug Act was passed only because of the support of the National Organization of Rare Diseases. I urge patients, physicians and insurers to create a similar group to support the commercialization of personalized cancer drugs.

I am so grateful for the extra time a PM drug gave me. My hope is that future patients have the same chance to benefit from personalized medicine.

RARE DISEASE DAY

More than a hundred patient advocates, biotechnology industry stakeholders and legislators gathered to mark the third annual Rare Disease Day at the State House on Feb. 28.

The day calls attention to the public health issues associated with rare diseases, which affect nearly 30 million Americans and countless others around the world.

People with rare diseases often face challenges that occur less frequently with more common diseases, including delay in getting an accurate diagnosis or a missed diagnosis, few treatment options and difficulty finding medical experts.

Speakers at the event included Blair Van Brunt, President of the Shwachman Diamond Syndrome Foundation, Neil Kirby, CEO of Edimer Pharmaceuticals, Alison McVie-Wylie, a researcher at Genzyme, Mark Baldry of Shire Pharmaceuticals, Rep. David Linsky of Natick and John Heffernan from MassBio.



Innovation 2011 announced

The Innovation 2011 Vendor Expo, which will link MassBio members with the latest and greatest technologies, equipment, supplies and services, is quickly approaching. This is a not-to-miss event for research and development scientists, as well as purchasing, safety, facility and operations professionals.

Co-hosted by MassBio and Fisher Scientific, the event will be held on May 2 at the Boston Marriott Cambridge.

Innovation 2011 will also offer four high-level, application-based seminars focused on the latest technologies and cutting-edge topics most important to

MassBio members. The seminars, which will be held from 10 a.m. to 3 p.m., will include presentations by Millipore, Promega, Thermo Fisher Genomics and Thermo Fisher Chromatography.

The event is free to all employees of MassBio member companies and registration is not necessary. There is also still exhibit space available for members and non-members.

Companies interested in exhibiting can register at www.massbio.org or contact Lauren Perna at 617-674-5100.

INNOVATION 2011 SCHEDULE

11 a.m.-3:30 p.m.	Expo hours
10-10:45 a.m.	Presentation by Millipore
11-11:45 a.m.	Presentation by Promega
12:30-1:15 p.m.	Presentation by Thermo Fisher Genomics
1:30-2:15 p.m.	Presentation by Thermo Fisher Chromatography

A boxed lunch will be served in the Expo room from 11:30 a.m. to 1:30 p.m.

Edimer Pharmaceuticals delivers hope for XLHED patients

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Edimer was established in 2009 with investment from Third Rock Ventures and VI Partners. Last year, Third Rock Ventures invited Pond to speak at its Rare Disease Day event, where she met members of the Edimer team.

"They wanted to hear about the family perspective that brings it all to life," said Pond. "It was an awesome group. They wanted to be more informed, and it's wonderful that they wanted to learn from my life experiences. Being connected, I can be that go-between,

bringing other families to Edimer."

Pond encourages families to join the Edimer Patient Network so they can get information and updates on their work.

"Beth has been such a tremendous resource," said Tessa Lorenze, who is responsible for patient outreach at Edimer. "She and her brother are willing to come in and test out equipment and share their stories. It's less clinical and more anecdotal. It helps us stay in tune with what's going on and what's important to patients."

While there are currently no specific therapies for the treatment of XLHED, Edimer is developing an innovative therapy, EDI200, which has been shown in animal studies to substitute for Ectodysplasin-A1 – a protein that is involved in the formation and development of skin and teeth. EDI200 is in natural history studies right now and is expected to go into clinical trial in late 2011/early 2012.

"It's so exciting and life-changing," said Pond. "My first thought was, 'I can't believe this is happening in my lifetime.'"

Though to be effective, EDI200 will likely have to be given within the first few weeks of life, before the ectodermal appendages are already established.

"It's heartbreaking to me that it won't help my brother or sons, but it does give hope to future generations," said Pond. "All of Philip and Nolan's daughters will be carriers, so hope is all we've got."

To learn more about Edimer Pharmaceuticals and EDI200, please visit <http://edimerpharma.com>.