The Educator

Newsletter of the National Foundation for Ectodermal Dysplasias Fall 2012

Magical Moments

National Foundation for Ectodermal Dysplasias

Fall 2012

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We invite your correspondence and welcome your comments, observations, and suggestions. Please send your letters to the following:

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Board President's Message

By Anil Vora
Board President

Exciting times at the NFED -- In 2012, we continue to fund research, educate health professionals and provide treatment for our families. This October, the NFED hosted the Classification Conference in Charleston, South Carolina. Judy Woodruff, Mary Fete, and Mary Kaye Richter participated in this important conference.

The goal of this conference was to develop a functional, robust, and powerful grouping of ectodermal dysplasias, which will have long-reaching implications for future research and treatment. In addition, we also began a research project for Goltz syndrome. These projects will require your participation, as we progress into the future.

First and foremost, the NFED is a family to family foundation. Over the years, the NFED has connected families, providing personal assistance, education, and Family Conferences. New families continue to join, learn and get connected. In order to keep these services current and complete, we must rely on gifts from many different sources. Since the Foundation does not receive government funding, we depend on donations from our families, the community, corporations and grantors.

Due to the current economic climate, our finances are stretched extremely thin. To keep our mission strong, I ask that each of you help in raising the funds necessary to meet our budget. For without this income, we are not able to provide the services so many individuals have come to rely on.

Please consider the following ways you can help the NFED:

Contributions

- If you are able, please increase the amount of your annual gift to the Foundation.
- If you have already made a gift to the NFED this year, consider sending a second gift.
- If you have never given or haven't given in a few years, consider a gift.

Employers

- Check if your company has a Matching Gift Program - it's the perfect way to double the impact of your gift.
- If you are a U.S. government employee, you can contribute through the Combined Federal Campaign. The NFED is #10604.

Events

- Instead of special-occasion gifts (e.g. birthday), ask friends to donate to the NFED in your honor.
- Hold a fundraiser that benefits the NFED – a Walk or an event with which you are comfortable.

Please contact the NFED if you can help with any of the above, or if you have other ideas that can benefit the NFED.

Volunteer

Our goal this coming year is to engage, connect, and empower more of our families in many different aspects of the NFED's daily life. You might be interested in volunteering as an Ambassador who provides community awareness; being a family liaison, or as a program participant, like our young adults who conducted the Virtual Bash this year; or as an advisor who researches and assists with alternative treatment funding. Contact the NFED if any of these volunteer opportunities interests you. We'd love to work with you.

Stay Connected

Help us stay in touch with you and reduce our mailing costs, by providing your email address to the NFED (info@nfed.org).

Your gifts and those of your family and friends will make a tremendous difference in these programs that impact the lives of people affected by ectodermal dysplasias. Please know how much we appreciate your time, talent, and gifts in any of these areas.

Find the Magic in You

2012 NATIONAL FAMILY CONFERENCE
JULY 18-21, ORLANDO
NATIONAL FOUNDATION FOR ECTODERMAL DYSPLASIAS

On the Cover

Madison Hoffmann, who is visually impaired, meets Minnie Mouse.

Conference Scholarships

Thanks to the NFED and Edimer Pharmaceuticals, 28 families received scholarships to attend the conference.

Connecting people!

Syndrome ribbons and regional stickers helped families connect with other families who are affected by the same syndrome or who lived within their region.

By The Numbers

- 302 Attendees
- 121 Kids
- 101 Affected Individuals
  AEC syndrome, Clouston syndrome, EEC syndrome, Goltz syndrome, HED, TDO syndrome, Unknown
- 6 Countries Represented
  USA, Brazil, Canada, Pakistan, Saudi Arabia, Switzerland
- 30 States Represented
- 2 Care Providers Who Want to Learn More
- 38 Volunteers

You can check out all the conference news by reading the archives on the NFED Conference Blog

at

www.nfededucator.wordpress.com.

Opening Night Kick Off

Caitlin Surubbi (below right) opened the conference with her inspiring story.

Buddy Program

This year, we launched the Buddy Program to connect first-time attendees with veterans to make their first conference more meaningful. The families report that it was a huge success!

Repeat Offenders

We recognized our "Repeat Offenders." These individuals attended five or more National Family Conferences.

5-9 years of attendance - 28 people

10-14 years of attendance - 3 people

15+ years of attendance - 5 people

Pictured below: Rhonda Page, John and Virginia Dickie, Anil and Sean Vora.

Syndrome Specific Workshops

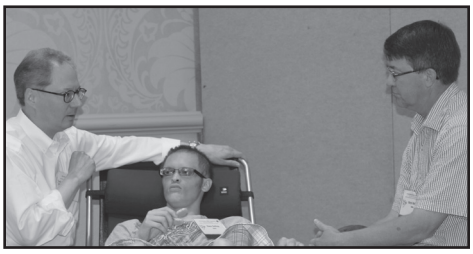
Syndrome specific workshops allowed families who are affected by same syndromes to learn from the experts and share their experiences with each other. Doctors and dentists from our Councils who are experts in their fields and who have extensive knowledge of the ectodermal dysplasias presented lectures, workshops, and panels on topics ranging from pediatric care, dental care for children and adults, women's health issues and many more.

The Family Conference is truly the best venue to getting the most accurate information on ectodermal dysplasias as the world's leading experts on ectodermal dysplasias are available to the families.

Pictured above are the EEC families.

Clinical Research

Four NFED dental experts, including Dr. Clark Stanford (pictured right) evaluated patients and consulted with them about their personal oral health issues.



Edimer Pharmaceuticals, Inc., the NFED and Washington University School of Medicine collaborated on a study to examine whether special software can recognize the facial features of male infants affected by hypohidrotic ectodermal dysplasia (HED) soon after birth as compared to later in life. The study also collected data to determine if there are nutritional deficiencies in males with HED that cause health and growth issues.

FAMILY CONFERENCE

Kays' Kids Camp

Thanks to Louis and June Kay, who sponsored this year's Kays' Kids Camp, 120 children were busy with scavenger hunts, laughing, art projects, games, and making life-long friends.

Talent Show

We had an all-time record of 49 acts for our Talent Show. Our families amazed us with their talents. Skyler Long sang "in the Navy" while dad was giving cues, dancing and singing in the audience. Simply adorable!

Anthony Barbar was a hit as he sang the Miami Dolphin fight song and Alex Quintanar brought tears to our eyes as he overcame his stage fright to sing. Executive director Judy Woodruff and her husband, Mike, closed the Talent Show by singing, "What a Wonderful World." Crystal Mata showed true talent as our Talent Show MC.

Mickey and Minnie Mouse

On Thursday evening, families grabbed their swim gear and cameras for a pool party sponsored by Edimer Pharmaceuticals. Families enjoyed swimming and making new friends. Mickey and Minnie Mouse even made a guest appearance to the delight of the kids both young and old. Thank you Edimer for sponsoring their visit!

My Conference Experience

By Crystal Mata

The very first Family Conference that I attended drastically changed my life. It was held in Houston in 1998. I had been spending my day at the Kids Camp, and, without warning, a fellow camper had a seizure right in front of me.

After a long 13 years, I was able to save up enough money to attend a National Family Conference. Coming back to this tight-knit community only reminded me of how important it is to be a part of this unwavering support group.

I learned that being there for new families was more important now more than ever. The Conference is more than a time for us to talk to doctors and professionals about medical problems and solutions for day-to-day issues. It's a time for us to come together as a family to learn from each other, support each other, and create long-lasting relationships with one another.

Meeting everyone often is the best part of the Conference because you finally get to see that you're not alone. There are dozens of other families going through and overcoming the exact same thing. I hope, however helpful this information might be, that I will have the opportunity to see familiar faces and meet new families in Houston next year. Come join our family!

This Conference Outshone All of the Others!

By Dee Dee Olsen

After every Family Conference there are many attendees who say, "This conference was the best one yet!" They say this, despite the fact that most of them are conference veterans, having attended these conferences for years.

This year's Family Conference in Florida was our family's eighth. And, though I have said this many times before, this time I say it with the utmost conviction: This conference outshined all of the others!

There were so many highlights! Often I find myself flipping through the photos of our wonderful experience at this conference. I can't stop viewing the smiling faces, the children holding hands, the countless poses with Mickey and Minnie. My video clips of various kids performing at the Talent Show will forever bring a smile to my face. There is no doubt that everyone had a great time! However, if you paid close attention, there was something far more important going on than just a good time. If you were attending conference as a family who is affected with Goltz syndrome, then something truly miraculous was transpiring!

This year at the conference, the largest number of Goltz families ever to congregate in one place spent an entire day together. These eight families not only shared common concerns, but we also made history! Goltz syndrome is an ectodermal dysplasia that affects mostly girls. These eight girls had the pleasure of an unforgettable bonding experience and, more importantly, they moved mountains. These girls were given the opportunity to participate in a groundbreaking research study. This study has the potential to alter, if not end, Goltz syndrome altogether.

I am delighted to tell you a small number of doctors have taken an interest in studying Goltz syndrome. The doctors were able to gather information and even collect biopsies from our brave girls. It was explained to our girls that the samples gathered on that day would be able to live on in a laboratory indefinitely! The samples could be used again and again to answer endless questions without them having to undergo that testing themselves. We are all excited at what this could possibly mean. We look forward to hearing what these doctors can discover almost as much as we look forward to next year's conference.

Volunteers Needed

We are launching a new volunteer program in 2013 and we need your help! We are looking for people who can help us in one of these areas:

- raise awareness in your local community;
- coach families on the insurance process;
- mentor families to be advocates at their children's school; and
- more.

Stay tuned for more information!

I Found the Magic in Me!

By Lindsay Harris

We all have our own stories about how we came to become part of the NFED family. This is my story.

I was born in January of 1982 with an extra digit or a second "thumb" on my right hand. Both thumbs and my index finger were abnormal and both the extra thumb and index finger were missing nails. Otherwise, I was a seemingly healthy child. As a few months passed, chronic conjunctivitis led to the discovery of malformed lachrymal ducts. As an infant, my parents chose to have my second digit removed and attempted to fix the lachrymal duct situation. As I grew, I continued to surprise my parents with little medical quirks.

When I was a two or three-year-old child, my eyes were red, swollen and goopy; the conjunctivitis never quite seemed to cleared up. I was extremely photosensitive. I was catheterized when I needed to urinate, I was susceptible to chest congestion, bloody noses and frequently had nausea. My parents were amazing and were vigilant about taking me to all the best doctors and trying to take care of me.

It was after a toddler visit to the dentist that the term "ectodermal dysplasia" was introduced to my family. Essentially, a Xeroxed definition was handed to my parents and the doctors said something to the degree of "we think she may have this." I was seen by a geneticist who agreed and believed I had symptoms of ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome and lacrimal lacrimo-auriculo-dento-digital syndrome. But nothing was ever confirmed. No one seemed to know enough about any of it to determine what exactly was "wrong" with me. My parents treated each issue and event separately as best they could and with nothing less than acceptance and unconditional love.

> **"The "Find the Magic in You" NFED Family Conference changed my life 180 degrees.**

After multiple hand surgeries, extensive dental work resulting in the preservation of most of my baby teeth until I was 18 followed by complete dental implants and jaw realignment, I had made it through high school and college and into adulthood. I suffered through the typical difficulties of adolescence compounded by extra teasing through junior high because of my differences. Yes, it was hard. Yes, it hurt. Yes, I wondered why I wasn't a "normal" girl. No, I wouldn't change any of it. I AM normal. I didn't and don't know anything different. I developed a unique ability to navigate the world and the hardships in a way that I would always end up on top. Yes, I would have loved to have known anyone who was like me.

Fast forward to Christmas of 2011, two weeks before my 30th birthday. I had been having intense eye pain, swelling, temporary blindness, and extreme photosensitivity to the point that I decided to go to an eye doctor. It was then that I was told I had bilateral corneal abrasions.

OUCH! The doctor wasn't sure what it was or why but was concerned with the preservation of my vision and urged me to see a specialist. My mother and I had talked over the years and we always felt that somehow all of my medical issues were interconnected, including the new diagnosis of the corneal abrasions, but we never knew for sure.

When I got home from the eye doctor, I was distraught. I am a professional makeup artist and an esthetician. My eyes are vital to my career. My hand never stood in my way of anything I ever wanted to do. I wasn't about to let my eyes. Once I calmed myself to the point of rational thought, I decided to learn what I could about my new diagnosis. I Googled ectodermal dysplasia. And there it was: www.nfed.org. I called the phone number listed, eager for information, and got the holiday voicemail. I left a message that I can only imagine sounded incoherent and slurred through nervous tears.

On January 3, 2012, my phone rang. In that moment my life changed. I spoke to one of the NFED angels and I told her that I wanted some information. When the packet came in the mail, 30 years of loneliness started to evaporate with the realization that I was not alone. The images of those precious children brought me to my knees. I looked more like them than I did anyone in my immediate family.

Lindsay Harris, JoAnna Nix and Heather McKelvie

I wanted to get involved immediately. The NFED told me the best way would be to attend the Family Conference in Orlando. I read about what the Conference was about and signed up. I realized I needed a roommate! The NFED gave me the name of Heather McKelvie. I sent her a text and we started talking about ectodermal dysplasias, and for the first time in my life I was talking to someone else who had it. I was elated! We became Facebook friends and she introduced me to JoAnna Nix. The three of us decided we would be great roommates. The countdown to Orlando was on!

The "Find the Magic in You" NFED Family Conference changed my life 180 degrees. I learned more about myself in three days than I knew what to do with. It was an emotional roller coaster. Those of you who met me know that I cried most of the conference. My tears were not out of fear or sadness, but relief and acceptance. Perfect strangers were more welcoming, understanding, accepting, loving, patient, caring and kind than I was prepared for.

I saw children who looked like me as a little girl, and I met a group of adults who all related to me on such a level that words cannot describe. My entire life, the people who loved me TRIED to understand, tried to relate, tried to comfort me. And now, as an adult, these strangers actually UNDERSTOOD! They lived through the same medical obstacles I did. They understood what it was like getting dentures, having painful skin and nails, not being able to breathe, having people look at you and whisper. I had finally found a place where I felt like everyone else!

The NFED has become more than a website with information for me. It has become a family. People I met over three days have become people I talk to regularly. I said the Conference changed my life 180 degrees. It changed a part of my being. It changed my thought process. It confirmed my belief that there is something greater than myself. My heart has opened. My mind has expanded. My hope has grown. My fear has lessened. My loneliness is gone. I have found peace within myself. I am stronger knowing I'm not alone. I know now that no matter what life throws at me next, my immediate family and my new extended family will be there to help me through it.

I truly found the magic within me.

To read the rest of Lindsay's story, go to the NFED website, under Meet Our Families.

Our First Employee Retires After 31 Years!

One day in 1981, Mary K. Richter asked her friend, Beverly Meier, if she would help her write a few letters to dental schools across the country. Lucky for us, she said yes! She became the National Foundation for Ectodermal Dysplasias' first and longest-standing employee.

It's now 31 years later and Beverly recently retired from her NFED family. We honored Beverly at the National Family Conference this past July. Crystal Mata serenaded her. The children each brought her chocolate kisses. Following is the tribute that Virginia Dickie, Rhonda Page, and Lynn Van Cavage gave her.

Here's to Beverly!

Beverly did type a lot of letters, but that's not all she did. There isn't a job at the NFED that Bev hasn't done.

For the first 25 years or so, it was Beverly's cheerful voice on the end of the line that you heard when you called the NFED office. She was the one who welcomed new families, listened to their stories, and offered care and advice.

She made copies, stuffed envelopes, planned Family Conferences, and took minutes at Board meetings. She managed the finances and books, cleaned office toilets, opened the mail, packed endless boxes and cleaned out storage sheds.

She took care of kids in Kids' Camp, walked on her knees to entertain them and was "Baby Spice" in the Spice Girls performance at the 1999 Family Conference. She's been the office mother to the staff. She's kept them in line with her little reminders to close their windows, turn in their time sheets, and clean out their old food from the fridge.

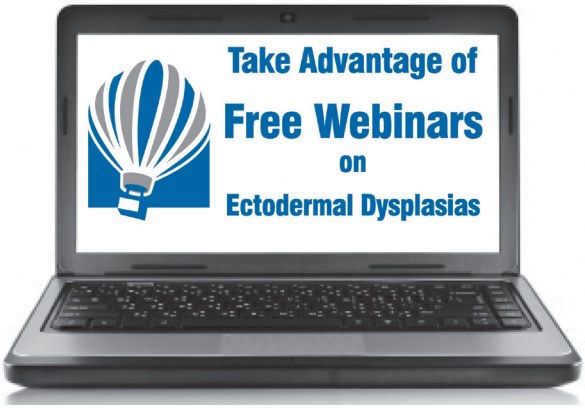
She was the one who's always crossed every "t" and dotted every "i". She's the hugger who always listened to families' stories, sharing in their joys and sorrows. She's the one who's always laughing, whose cackle can be heard from great distances.

Beverly has done it all for us for the past 31 years. The NFED would not exist had it not been for her tireless efforts. We thank Beverly Meier with all of our hearts. Mary Kaye was leading the charge and making the NFED soar. And Beverly was quietly in the background providing the wind beneath her wings.

We thank Beverly and wish her a wonderful retirement!

Go to our website to see more photos of Beverly through the years and to watch Mary Kaye's tribute video.

If you would like to send a note to Beverly, you can mail it to the NFED office and we'll forward to her.



2013 Webinar Schedule

All webinars take place at 5 p.m. Pacific / 6 p.m. Mountain / 7 p.m. Central / 8. p.m. Eastern. Webinars are free and last one hour. Visit our Web site to register for each Webinar.

Miss a webinar? *- No problem! Go to the NFED website where you can access recordings of all webinars.*


How to Handle Teasing and Bullying

Tuesday, February 12

Presenters: Mary Fete, R.N., M.S.N., C.C.M., NFED
Margot Stein, Ph.D., University of North Carolina

Join the discussion on how to handle teasing and bullying in today's world. Ms. Fete and Dr. Stein will provide coping tips and strategies.


Genetics and Reproductive Health

Tuesday, May 14

Presenters: Dorothy Katherine Grange, M.D., St. Louis Children's Hospital
Jill Powell, M.D., St. Mary's Health Center

Join us to learn from our experts. Drs. Jill Powell and Kathy Grange will discuss current approaches to pregnancy planning and genetic diagnostic testing. Topics to include preimplantation diagnosis, chorionic villus sampling (CVS), in vitro fertilization (IVF) and amniocentesis. Learn the benefits of preconception genetic counseling with a geneticist, a genetic counselor, high risk OB and other specialty providers.

EDI200, Treatment of X-Linked Hypohidrotic Ectodermal Dysplasia Update

Tuesday, August 13

Presenter: Kenneth Huttner, M.D., Ph.D., Edimer Pharmaceuticals

Edimer Pharmaceuticals' vision is to improve the health and quality of life of future generations affected by X-linked Hypohidrotic Ectodermal Dysplasia (XLHED). Their goal is to make EDI200 (XLHED treatment) available to all families affected by XLHED. When will clinical trials begin? Has EDI200 passed FDA regulations? Join us to learn all of these answers and hear how Edimer is progressing.

Pediatric Dental Needs

Tuesday, November 12

Presenter: J. Timothy Wright, D.D.S., M.S., University of North Carolina

When should dental treatment begin? What is bonding? Does my child need both lower and upper dentures? Dr. Wright will answer these questions and share information about other pediatric dental concerns. This is your chance to talk to a dentist experienced in treating children affected by ectodermal dysplasias and get answers.

Family Liaison Program

Marc and Virginia Make 19…19 Family Liaisons That Is!

Virginia Higgins lives in St. Louis, Mo., with her husband, Brian, son, Zane, and daughter, Selah, where she homeschools, is active in ministry, and writes at thatbaldchick.com. Virginia and Zane are affected by ankyloblepharon-ectodermal defects-cleft lip and palate (AEC) syndrome. Virginia grew up with the NFED and was taught by her mom at an early age that people ridicule that which they do not understand; make them understand, and they will not ridicule. That life lesson empowered her to educate others about ectodermal dysplasias and she relishes opportunities to raise awareness. Virginia believes that you are only as limited as you allow yourself to be, and being affected by ectodermal dysplasias doesn't have to be a limit. Whether by phone, email, Facebook, or in person, Virginia would love to connect with you.

Marc Steingesser lives in Ellicott City, Md., with his wife, Mary, and daughters, Rachel and Sarah. Marc is affected by ectrodactyly ectodermal dysplasia-clefting (EEC) syndrome. Although it wasn't until he was in his 40s, Marc was profoundly and positively affected to learn that there were others "just like" him. Marc says that he has found the NFED to be an incredible organization and community that's made up of kind, knowledgeable, and caring people. He looks forward to helping others become enriched through their connection to the NFED.

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Toward A Cure for Skin Erosions in AEC Patients

Maranke Koster, Ph.D.
Peter J. Koch, Ph.D.
University of Colorado, Denver

Ankyloblepharon ectodermal dysplasia and clefting (AEC) syndrome is an ectodermal dysplasia characterized by skin fragility, often causing the skin to erode. Currently, there is no cure for skin erosions in AEC patients. Our goal is to develop a novel approach for treating skin erosions in AEC patients.

This treatment will be based on the use of induced pluripotent stem (iPS) cells. iPS cells are a special type of stem cells that is generated in the laboratory. These cells can be made out of any cell of the human body, but usually skin cells are used. The reason for this is that skin cells are easy to obtain (from a skin biopsy) and that skin cells are very easy to grow in the lab. The skin cells are then treated in a special way that will allow them to start to function like a specific type of stem cell, called pluripotent stem cells.

This type of stem cell can be turned into any cell type of the human body. Further, we can repair DNA mutations in this type of stem cell, something that cannot be accomplished in other types of cells. Therefore, the ultimate goal is to repair the disease-causing mutation in iPS cells derived from AEC patients and to turn the repaired cells into intact and healthy skin. This repaired skin can then be used to graft onto lesions of AEC patients.

We have already obtained skin biopsies from seven AEC patients at the 2011 NFED Family Conference. We have grown skin cells from these biopsies in the laboratory and have generated iPS cells from the skin cells. Currently, we are using novel techniques that will allow us to correct the disease-causing mutation in these iPS cells. We will then determine if we can turn the corrected cells into healthy skin, as we expect. Ultimately, our goal is to generate healthy keratinocytes for the treatment of skin erosions in AEC patients.

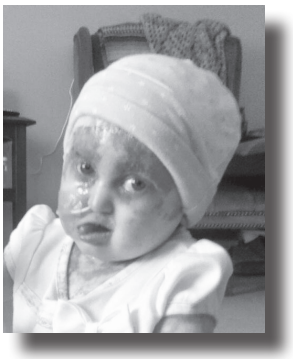
Characterization of A Novel Conditional Mouse Model For AEC Syndrome

Caterina Missero, Ph.D.
CEINGE Biotecnologie Avanzate
(Center for Genetic Engineering)

AEC syndrome is caused by mutations in the p63 gene, encoding for a protein that plays an essential role in early development. The p63 protein is a transcription factor, which means that it attaches (binds) to DNA and controls the activity of several genes involved in development of several ectodermal structures, such as the skin, hair, teeth, nails, and also the palate epithelium. The p63 gene mutations responsible for AEC syndrome interfere with the ability of p63 to turn target genes on and off at the right times.

To study AEC syndrome at the molecular level, we generated a mouse model that closely resembles the human syndrome. This model has been instrumental to demonstrate that skin atrophy and cleft palate are caused by reduced growth of the epidermis and of the palate epithelium during fetal life, due to defective expression of the receptor for the Fibroblast Growth Factor. In addition, the AEC mouse model is affected by epidermal fragility resembling the skin erosions observed in patients. However, further studies aimed at understanding the molecular cause of the erosions and the therapeutic implications have been impaired by neonatal lethality caused by cleft palate.

To overcome this problem, we recently generated a novel mouse model in which the AEC mutation can be conditionally induced during development, thereby overcoming neonatal lethality due to cleft palate. With this potent tool in our hands, we propose to study the molecular alterations that occur in AEC mice after birth focusing on epidermal fragility. Specifically, we will study alterations in the intercellular adhesion (adhesions among epidermal cells), which are the likely cause of the epidermal fragility. The goal of this project is to identify the cell adhesion components that are affected in AEC syndrome and that contribute to the skin erosions. Understanding the molecular mechanisms that lead to skin erosions will be essential to identify therapeutic treatment that prevent or alleviate the symptoms.



The proposed research also addresses a relevant question often asked by patients concerning the propensity to develop skin cancer. Skin erosions and the consequent inflammation are risk factors in skin areas subjected to mechanical stress. We will test whether AEC mice are more or less susceptible to skin tumors compared to normal mice using a well-establish chemical carcinogenesis protocol. Taken together, these studies will shed light on the molecular defects underlying skin erosions, which will allow us to design a strategy to prevent or alleviate a number of postnatal symptoms typical of the syndrome.

Studying Focal Dermal Hypoplasia Using Stem Cells

By Karl Willert, Ph.D.
University of California, San Diego

We have collected skin biopsies from five individuals with Goltz syndrome and are awaiting an additional four biopsies. After several weeks of growing these skin biopsies in a dish in the laboratory we obtained several million cells, some of which we have frozen for long-term storage.

As an initial characterization of these cells, we examined the DNA sequence of the PORCN, the gene that is changed in Goltz syndrome. In addition, we started the first experiments to convert these cells to stem cells. This conversion process is called "reprogramming" and the resulting stem cells are called "induced pluripotent stem cells" or iPS cells. We are growing these iPS cells so that we have enough cells for future experiments.

The next step in this project will be to show that the iPS cells do in fact behave like stem cells and are capable of growing indefinitely and giving rise to mature cell types, such as skin cells.

The NFED granted Dr. Willert $25,000 in funding and access to patients in 2012.

Goltz Families Needed to Participate in Major Research Conference

By Mary Fete, R.N., M.S.N., C.C.M

In 2013, we will sponsor a research conference dedicated to Goltz syndrome in conjunction with Baylor College of Medicine & Texas Children's Hospital. The dates will be July 22nd and 23rd in Houston, Texas. We need individuals who are affected by Goltz syndrome to help us and to participate in evaluations and screenings that will be accomplished at Texas Children's Hospital.

Please contact me at mary@nfed.org if you are interested in participating in this conference.

2013 Events Calendar

28 Days of Change
February 2013

Kayla's 5K to Benefit the NFED
Sunday, February 10, 2013
Port Richey, Fla.

Don't Sweat It Walk
Saturday, April 27, 2013
Fairview Heights, Ill.

Don't Sweat It Golf Classic
Friday, June 7, 2013
Fairview Heights, Ill.

Zach Hamm's Don't Sweat It Golf Classic
Sunday, July 21, 2013
Spring, Texas

Goltz Research Conference
July 22-23, 2013
Houston, Texas

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Growing Up With Goltz Syndrome

By Abigail Ernst

Many of us can reminisce on the many times a Band-Aid saved the day making all scrapes, scratches and boo-boos better. The sticky friend helping most children doesn't hold the same meaning for 7-year-old Emma Miller.

Band-Aids are usually a mother's go to, along with a kiss, in making an injury better. For Rebecca, Emma's mother, Band-Aids are the first thing she stays away from. This sticky friend is too harsh for Emma's fragile skin. Instead, gauze is the go-to bandage when needed.

Diagnosed within days of being born, Emma is affected by Goltz syndrome. Her skin conditions, including papilloma, rash, thinness and sensitivity are only part of her symptoms. She also has nine fingers and nine toes, colobomas and blind spots in both of her eyes, kidney reflex, not being able to sweat and cleft palms and feet.

Rebecca reminisced of when Emma was born, "We were lucky there was a geneticist at the hospital who had recently dealt with another individual affected by Goltz. She knew right away when she saw Emma."

Only given the diagnosis of Goltz syndrome, Rebecca went to the internet and started researching. She came across the National Foundation for Ectodermal Dysplasias, which led her and her daughter, seven years later, to attend their first National Family Conference, "Find the Magic in You!" in Orlando.

"Conference was awesome," Rebecca said , remembering the feelings created while at Conference. "It was almost peaceful to know other people are going through the same thing."

Matched with another Goltz family, Rebecca found comfort in talking about her experiences. The families shared birthing stories, surgeries they have gone through with their children and day-to-day challenges. For Emma, it was the first time she had ever met anyone else like her. Before, she had always known being affected by Goltz syndrome was, "just the way God made me."

Conference gave Emma the chance to meet other girls that know what it is like growing up with Goltz syndrome.

Emma, back at home and school, goes to bed knowing that there are other girls, also affected by Goltz syndrome, going through the same thing. Knowing she is affected but not letting her symptoms stand in her way of growing up, Emma continues to have a wild imagination and loves spending time with her two pet cats whom she refers to as her brother and sister.

Answering Your Questions about the Adults with XLHED Study

Participants asked the following questions on a webinar that the NFED hosted in August regarding Edimer Pharmaceuticals' Adults with XLHED Study. Edimer provided the answers. To read more of the Q and A and listen to the webinar, go to our website under research news.

EDI200

When will the studies take place? Are specific dates available or is it dependent upon the participant's schedule?

The adult study will take place in late 2012 and early 2013.

What class of drug is EDI200 and how does it work?

EDI200 is a fully humanized replacement protein.

Can you explain the difference between protein (EDI200) and gene therapy?

EDI200 will treat the symptoms of XLHED by replacing the missing protein that is needed during the development of things like hair follicles, sweat glands, tooth buds, etc. It will disappear from the body within a few weeks. Gene therapy is intended to persist in the body. EDI200 does not treat XLHED at the genetic level and will not correct the genetic mutation.

Is there a possibility for an allergic reaction to the protein treatment?

There is the possibility of unanticipated effects, like an allergic reaction. However, the course of EDI200 is a short one and individuals participating in the adult study will be under constant medical supervision for the 24-hour period following each dose of EDI200.

Is treatment expected to address symptoms only and not the genetic makeup of the individual and their likelihood to pass on XLHED?

EDI200 is not gene therapy. EDI200 will potentially treat symptoms only and will not impact an individual's potential of passing the mutation to his/her children.

Do you expect that the drug would be needed as a lifetime treatment?

Results from the animal studies suggest that a single course of therapy given early in life is sufficient to correct many of the symptoms associated with XLHED, and that this benefit lasts through adulthood.

What are the potential side effects for humans?

When EDI200 was given to animals that most closely resemble humans (monkeys) and in a manner that is consistent to how it will be administered in our studies (slow IV infusion), no adverse effects were observed. Given these results we do not anticipate any side effects in humans. However, unanticipated effects (like allergic reactions) are always a possibility.

Are the teeth in dogs of normal in appearance and function?

In the best case of early XLHED dog therapy with EDI200 there are no missing teeth and the teeth are close to normal in structure.

Will participating in this study have any impact on ectodermal dysplasias for me? I assume that nothing is supposed to happen since I am an adult.

We do not anticipate that there will be any clinical benefit of giving EDI200 to an XLHED affected adult.

From studying the treated animals, would life expectancy be altered for the humans being treated?

Results from treating XLHED dogs with EDI200 demonstrate a reduction in life-threatening illness and a normal life span.

Ectodermal Dysplasias Classification:

A Model For Integrations Of Clinical, Systems Biology and Bioinformatics

By Laurie Jo Mattson
NFED Intern

The following is an interview with Carlos F. Salinas, D.M.D., F.A.C.D. and J. Timothy Wright, D.D.S., M.S.

The NFED-sponsored Classification Conference was held on October 18-20 in Charleston, SC. Carlos F. Salinas, D.M.D., F.A.C.D., Conference Chair, reports the goal of this conference was to have scientific experts from all over the world come to an international consensus toward the development of a more inclusive classification model for ectodermal dysplasias.

While Salinas admits that this was challenging because many symptoms of this condition are not simply "black or white," he remains optimistic.

"I have really high hope that we are going to have an agreement in the near future," Salinas says.

If they are successful, this new classification model will ideally be made into an interactive online database that J. Timothy Wright, D.D.S., M.S. says will optimize communication between researchers, clinicians, and families dealing with ectodermal dysplasias.

Wright says that if the conference comes out with a "very functional, robust, powerful" grouping of ectodermal dysplasias, then it will have long-reaching implications for future research.

"People will say, 'Wow, the NFED and the folks dealing with the ectodermal dysplasias really set the groundwork and led the way in this area," Wright says.

Right now, both Salinas and Wright stress that the current classification system is lacking some key components to fully understanding ectodermal dysplasais at its multiple, complex levels. The current classification is based on the clinical (or physical) appearance of this condition, whereas the new classification method would take a multi-axis approach that would include a gene-based methodology as well.

Having a combination of the genetic and clinical domain will help bring researchers and clinicians more clarity.

While clinicians are more adept at recognizing the physical symptoms, they often might lack a more in-depth knowledge of the disorder at a molecular level. Conversely, researchers might be experts of disorders at a cellular level, but they might have trouble understanding symptoms at a physical level.

Dr. Wright says all three of these domains are important to having a full understanding of ectodermal dysplasias and how to treat it best.

The first axis, clinical, is the physical manifestations of the disorder. For example, the clinical characteristics of a person with hypohidrotic ectodermal dysplasia could be sparse hair, missing teeth, or diminished sweat glands.

The second, gene-based axis revolves around identifying if the gene is known or unknown and how the disorder might have been inherited.

The third axis, functional pathways, is looking into how certain genes interact with other genes. Often, when certain genes intermingle with each other, they cause a trigger-reaction, or cascade.

"[It explains] why you have that gene that causes this protein to be altered in this way and that causes that protein to then not function appropriately in this cascade. That's why cells do what they do. Or, do what they shouldn't do. Or, they don't do what they should do," says Dr. Wright. "That's what causes the clinical symptoms."

Wright thinks that this could draw a fair amount of attention to people that might not have originally been interested in ectodermal dysplasias.

Having these conferences and then publishing the proceedings in different scientific and genetics journals increases the exposure to ectodermal dysplasias by letting people know what the current science is, and what actions are being taken.

"Once those papers come out in journals, there is a lot of exposure and a broad readership, and not only that, but they're available to everyone in the world," Wright said. "That's a huge benefit to having the conference."

Salinas adds that this kind of attention could also help raise funds for more innovative research on the subject as well.

"When you bring this to the attention of the researchers, researchers are going to be pushing for funding support … in order to advance in their research," Salinas explained.

Because the database is only in the planning stages, Wright says that funds will also be crucial to get the database up and running. To do this, they not only need researchers who understand the condition and all of its variations, but also people who are" tech-savvy" and know their way around the complicated software needed to complete this project.

Mary Fete, R.N., M.S.N., the NFED's Sr. Director of Programs reports that she was thrilled with the Conference.

"It was amazing to see experts from around the world come together and arrive at a consensus on how to classify the ectodermal dysplasias. They made major strides that will change how we all look at the conditions."

Women's Survey Results Help Us Understand Female-Specific Issues

By Jill Powell, M.D. St. Mary's Health Center and Mary Fete, R.N., M.S.N., C.C.M., NFED

The results are in! The NFED sponsored a survey to further characterize female-specific issues and their possible connection to ectodermal dysplasias.

Background

Seventy-nine female respondents affected by ectodermal dysplasias completed a pilot survey about topics ranging from breast development and function, to menstruation and puberty. This survey laid the groundwork to verify that breast and vaginal concerns are associated with ectodermal dysplasias. A larger, follow-up survey provided results which fostered the growing correlation between these characteristics and ectodermal dysplasias.

Of the 2,333 surveys sent, 427 women between the ages of 12 and 83 responded. These women had many different diagnoses of ectodermal dysplasias, but the majority (118 women) were hypohidrotic ectodermal dysplasia (HED) or HED carriers (159 women). Here is what we learned with their help.

Sweating

- Fifty-five percent of all respondents reported decreased sweating, while 35% had normal or excessive sweating. Ten percent claimed to have no sweating at all.
- Sixty-four percent of women with HED also reported having decreased sweating. Twenty-one percent had no sweating at all, and 15% had normal to excessive sweating.

Puberty

Fifty-two percent of the women reported having their first menstrual period at age 12 or 13. Both women with HED and HED carriers reported that they had their first period between the ages of 11 and 14. The majority (73%) of the total group reported that their regular cycles last between 4 to 7 days.

Breast Issues

- Twenty-five percent of the total group have one or both of their nipples that were either absent or hypoplastic, or, in other words, abnormally small.
- At close to 30%, the majority of those surveyed with HED also reported having underdevelopment or incomplete development of nipples. Other issues included no breast development on either the left or right side, or no breast development on both sides. A small percentage also reported to either having no right or no left nipple.
- HED carriers had a smaller percentage of reported breast issues. The biggest concern at 5% was that there had been no breast development on either their right or their left side. Less than 1% reported the absence of a nipple.

Breastfeeding

One of the predominant concerns was breastfeeding. While 76.2% of the total group wanted to breastfeed, only 31.4% were successful. Almost half of the women needed to supplement breastfeeding with formula.

Pubic Hair

- Twenty-three percent of the total group has absent or sparse pubic hair, while 40% reported absent or sparse axillary, or armpit, hair.
- A more significant number of women with HED reported having this problem. Eighty-five present reported sparse or thin head hair, while 64% said they had absent or thinning axillary hair.
- For HED carriers, the majority at 66% claimed to have sparse, absent, or thin head hair. Forty-four said they experienced sparse, or thin pubic hair and 51% reported having these symptoms with their axillary hair.

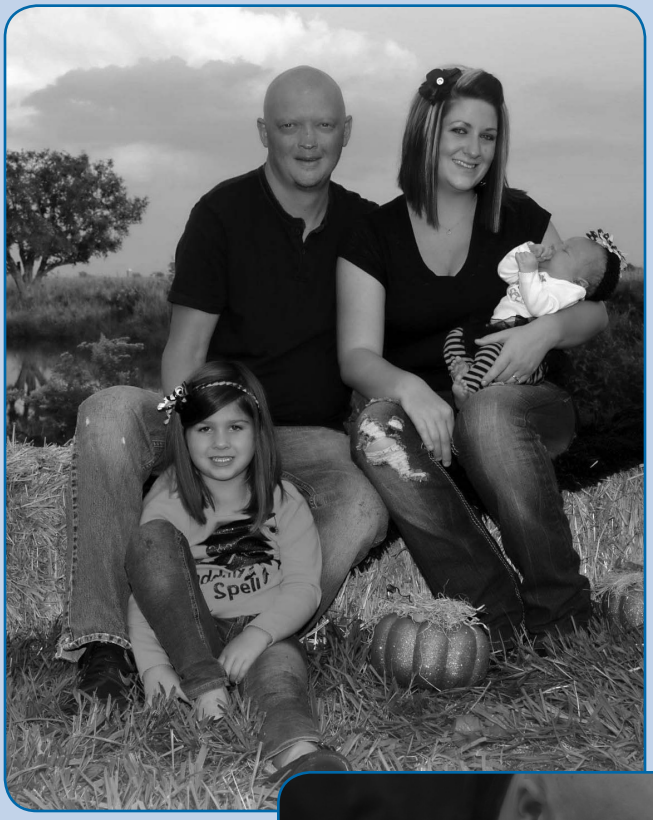
Vaginal Issues

- Unlike the commonly reported breast abnormalities, there was not a large reported increase of abnormal uterus or vagina development as compared to the general population.
- A troubling characteristic that is shared among these women affected by ectodermal dysplasias, however, is the reported percentages of those who stated intercourse was often painful. Eighty-five percent of the total group of 18 to 40-year-olds reported this as well as 100% of the women who were 41 years and older. Forty percent of HED women reported chronically painful intercourse.
- Yeast infections also seemed to be prevalent in these surveys. Sixty-two percent reported them as a frequent occurrence. The majority of the women who reported this as a recurrent problem were 41 years and older.
- Fourteen and a half percent reported having reoccurring bladder infections. Frequent, nighttime, or painful urination was also reported as well as urinary incontinence. These results, however, are not increased above general population rates.

Infertility (Only Reported for Women with HED)

The majority of the women with HED reported having no problems with conception. Only 5% of the 18 to 40-year-olds reported having difficulty, while 21% of women older than 41 reported having previous difficulty.

Charlie's Story Continues with the arrival of Baby Charley!



If you have been with the NFED for any amount of time, you know that Charlie Richter is the reason we exist. His mom and NFED founder, Mary K. Richter, chronicled his birth and childhood in "Charlie's Story."

We are happy to share with you the latest chapter. Charlie, now 34, married his wife, Bethany, in early 2012. Together with their daughter, Kylie, they welcomed second daughter, Charley Reece Richter.

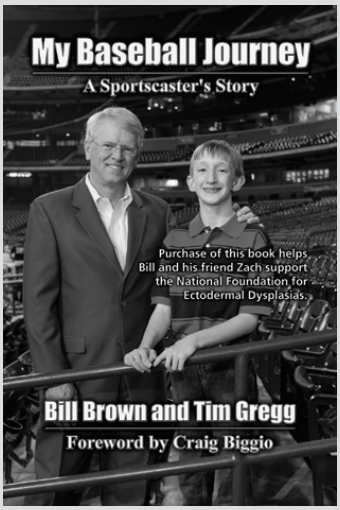
We congratulate the Richters, the very proud daddy and the happy grandma!

We Welcome Amy to the Program

Amy Rush is the administrative assistant to the program team. She is helping Mary Fete and Kelley Atchison with execution of all programs that we offer.

Amy says, "I am privileged to work for NFED where everyone is a family. As the newest member of the staff, I can't wait to meet families and hear their stories, which will ultimately help me work with the program team for support, research and treatment of ectodermal dysplasias."

Baseball Broadcaster's New Book Benefits the NFED



Bill Brown, the television broadcaster for the Houston Astros for the last 26 years, met 12-year-old Zach Hamm and was touched by his resiliency and heart. Wanting to help Zach and the NFED, Bill wrote a book titled *My Baseball Journey: A Sportscasters Story*, and is donating all proceeds to the NFED. The book is a must read for every baseball fan. Bill brings to life the stories of those who have inspired him, and he talks openly about his struggles to gain a foothold in the broadcasting business. He features Zach in chapter 26. Baseball great Craig Biggio wrote the foreword.

Bill and Zach have raised incredible awareness for ectodermal dysplasias and the NFED in promoting this book around the country at book signings, in the media and at numerous events. We are grateful to Bill for all that he is doing to help our NFED family!

Order Your Book Today!

Visit our website to see photos from their book signings or to purchase the book. It would make a great holiday gift! You can also learn more about the book at mybaseballjourney.com.

Thanks to These Families for Hosting NFED Fundraisers!

Runners and walkers picked up the pace at the Huxman Run 2 Sweat in October in Kan. Thank you, DeAnn and Chris Huxman for all of your hard work on this successful event!

We thank Ashli Matus George, Tom George, Kristin Matus-Kelso, Mark Kelso, and Terri and Jerry Matus who hosted the 8th Annual Rally for Ally in June in Vienna, Va.

- Joy Booth held a "Tea With Callie Booth" in Ga. and guests made a gift to the Foundation.
- Julie and Craig Claeys sponsored their first tournament called "Sweat Nothing Golf" in June in Mich.
- Dena and Kevin Davis of Ill. and their family stuff magazines in their local newspaper and donate their earnings.
- Sharon Garza of Texas recruited her friends to help her raise money for the 30 Days of Change campaign.
- Zach Hamm's Don't Sweat It Golf Classic was bigger and better than ever this past August in Spring, Texas. Thank you, Paul, Susan and Zach Hamm!
- Angela and Dante Purro had their first Animals for Ava event at Turtle Back Zoo in N.J. in honor of their daughter, Ava.
- Kudos to the Swierczewski crew on their 17th Bruno's Golf Outing for NFED in Blue Island, Ill. Golfers and friends helped raise much needed funds and awareness.
- The Wang Family once again held a concert at their children's school in Calif. to raise awareness and funds.
- For nearly three decades, Stanley Zwirn of Conn. has sold entertainment books with proceeds benefiting the NFED.

Don't Sweat It Walk 2012
NATIONAL FOUNDATION FOR ECTODERMAL DYSPLASIAS

The NFED began a national initiative this year, to raise funds and awareness through the Don't Sweat It Walks. We wanted to create an event that would appeal to many people around the country. Other organizations have had much success through walks, runs, and cycling events, so we created an event that our families could easily manage from their own communities. A tool-kit provides information on how a successful walk can be created. The response this first year has been overwhelming, and we want to salute our families who "stepped" up and hosted walks in their communities.

Start planning now to have your Don't Sweat It Walk in 2013! Call Katie at the NFED office to schedule yours.

Thanks to…

Delta Theta Tau Sorority in Bethel, Mo.

Kerri Fasulo in Poughkeepsie, N.Y.

Jennifer Hagerty in Tracy, Calif.

Tara Huber in Grove City, Ohio

Pam Kennedy in Canton, Ga.

NFED's Walk in Mascoutah, Ill.

Briana Pinon in Newark, Calif.

SIU School of Dental Medicine in Edwardsville, Ill.

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Save the Dates!

We Want You to...

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July 24-27, 2013
Houston, Texas

Get Your NFED Spirit Wear Online Today at www.NFED.org!