

DebRA Currents

THE QUARTERLY NEWSLETTER OF THE DYSTROPHIC EIPERMOLYSIS BULLOSA RESEARCH ASSOCIATION OF AMERICA

INSIDE:

Pez® Dispenses Sweet Results2

Genetic Testing4

Supplies to Share ...4

Ask the DebRA Nurse Educators5

Research Obstacles Conference6

DebRA Events7

Anna Dittman9

Research Grant10

DebRA Expands Nurse Educator Program; Registry to Reopen

This summer, DebRA expanded its most vital program by funding two EB Nurse Educators based at prestigious institutions, according to Executive Director Suzanne Cohen. "There was an overwhelming demand for the services of our Nurse Educator so we doubled the staffing to meet that need. Along with the expansion, we decided it was to the benefit of our families to have these positions based with EB experts."

Adele Jones Westbrook, RN joined as the DebRA Nurse Educator based at Vanderbilt University in Nashville, TN and Geri Kelly-Mancuso, RN is the DebRA Nurse Educator based at Cincinnati Children's Hospital Medical Center. The positions are funded by grants from DebRA of America to Cincinnati Children's Hospital Medical Center and Vanderbilt

To contact a Nurse Educator, please call 866-DEBRA76 Monday to Friday from 9 a.m. to 5 p.m. Eastern Time or write nurse@debra.org.

University. The entire DebRA Nurse Educator Program is made possible through the generosity of National Rehab, Inc., the Pascucci Family Foundation, the Jennifer DePrizio Memorial Golf Tournament, NYBOT Futures and Options for Kids, Michael and Madeline Weiner, Connie and Paul Flowers, and other DebRA supporters.

continued on page 3

Families Tune into EB in Nashville



Amaya charms during PCC

More than 250 people flocked to Nashville for DebRA's 2006 Patient Care Conference entitled "Tune Into EB." "Music City" played host to the free biennial conference that featured presentations from leading authorities on the various aspects of EB.

DebRA Trustee and Scientific Advisory Board Member Jo-David Fine, MD, was the official host of the conference. The Vanderbilt University professor of dermatology developed the meeting curriculum in conjunction with EB Nurse Educator Madeline Weiner, RN, and both made several presentations throughout the meeting.

The agenda covered topics including the genetics of EB, GI manifestation, wound care, dental issues, psychosocial issues, nutrition, pain management, mobility and physical therapy, hand manifestations, anemia, cancer, insurance, independent living and entering the schools. The materials shared at the conference and

continued on page 2

Pez® Dispenses Sweet Results for EB Community

Who could have guessed that the ever popular and humble PEZ® dispenser could raise awareness and funds for EB. The non-profit Traveling PEZ Dispenser (TPD) organization was founded as a way to raise awareness for breast cancer using a specially made PEZ dispenser that travels weekly to a new donor. Then one day, TPD co-founder Chris Matthai had the opportunity to meet a little boy named Dylan with EB Simplex. Chris also knew the Peshkur family through his mother so he realized that TPD could be helpful to EB awareness and launch a program like it did for breast cancer.

Knowing that a simple way to explain the fragility of the skin in a person with EB was to liken it to the delicate wings of a butterfly, Chris decided to use an existing PEZ dispenser called Florence Flutterfly. It is a lighthearted head of a butterfly with a wide grin. Chris renamed the TPD version "Dylan" in honor of his little friend.

Each week the PEZ dispenser spends time with a donor who makes a minimum

donation of \$5 to host the dispenser. The minimum donation fee is waived for families directly affected by EB. The donor is asked to keep a journal and take photographs of their adventures with the dispenser. The journal and photos then get posted by TPD on their website www.tpd.zoomshare.com.

Dylan kicked off his world tour in October in Long Island at a Peshkur Family Foundation event and in December made a visit to the DebRA offices. Executive Director Suzanne Cohen and Development Manager Abby Meadows took Dylan to see the many holiday sights along Fifth Avenue.

Since the program began, Dylan has visited Germany, Japan, Canada and many U.S. states. PEZ continues to support the program and recently repackaged just 25 of the now retired dispensers with a Golden Ticket. Like in "Charlie and the Chocolate Factory", each Golden Ticket entitles the bearer to a private PEZ tour in Connecticut. TPD arranged for some



Dylan hails a cab in NYC

of these Golden Tickets to be used for a PEZ factory tour for a small group of EB Butterflies of all ages. PEZ does not give private tours so the event was truly special and led by "Pezident" Joseph Vittoria. Today, the remaining Golden Ticket Dylans are slowly being auctioned on Ebay to raise additional funds for the TPD project.

Proceeds of the Dylan EB project will be shared between DebRA and EBMRF. To sign up to participate, please email a phone number, email address, and mailing address to travelingpez@hotmail.com.

Nashville continued from page 1

presentations from most sessions are available in the 2006 PCC section at www.debra.org.

The PCC is free for attendees and was supported through the generosity of Gold Sponsors Mölnlycke and National Rehab, Inc., Silver Sponsor Tyco Healthcare/Kendall, Kids Room Sponsor Smith & Nephew and through educational grants from Organogenesis, the Pascucci Family Foundation and RegeneRx. Kids Room Supporters were the Forever Young Foundation, Hudson's Hope Foundation and Wayne-Westland Friends of EB. The conference received general support from William and Sarah Baum and William and Cathleen Donnelly and gifts-in-kind from Winfield Laboratories and Pedors Footwear.

More than 250 people flocked to Nashville for DebRA's 2006 Patient Care Conference entitled "Tune Into EB"

"The conference presents the unique opportunity to gather experts in the key aspects of EB in one place and offer families direct access to ask questions," DebRA Executive Director Suzanne Cohen said. "But just as important, it gives

everyone a chance to meet in person, share their stories and tips, and just spend time together."

An opportunity to further spread EB awareness arose at the conference when a film crew joined the activities. The Cripps family was being filmed at the PCC for a planned Discovery Channel documentary on siblings with rare diseases that will also include the Thurmond family. Dr. Fine and his fellow DebRA Scientific Advisory Board Members Anne Lucky, MD and Richard Azizkhan, MD were interviewed for the film as well.

The next Patient Care Conference will be held in 2008 with the date and location to be determined. Details will be shared as they are available.

Nurse Educator Program continued from page 1

SECOND NURSE HELPS REOPEN EB REGISTRY

In the newly created second Nurse Educator position, Ms. Westbrook will spend 30 hours per week on patient and family calls and emails and the remaining 10 hours helping her supervisor, Jo-David Fine, MD, with the National Epidermolysis Bullosa Registry. Dr. Fine, a DebRA Trustee and Scientific Advisory Board Member, is a world-class clinical and scientific expert on EB and Principal Investigator and Project Head of the National Epidermolysis Bullosa Registry.

The EB Registry was developed by Dr. Fine with support from DebRA of America and the N.I.H. and is the largest medical database of its kind in the world. The Registry collects information from patients with EB, characterizes the many forms of this disease, establishes the frequency of various manifestations of EB and determines the risks of specific clinical outcomes within EB types. It has yielded a great deal of knowledge about the disease and many of its findings have been published in peer-reviewed scholarly journals. The Registry was closed temporarily when Dr. Fine moved from University of North Carolina at Chapel Hill to Vanderbilt University.

Ms. Westbrook was already a Vanderbilt University employee whose prior position was as a Dermatology MOHS Micrographic Surgical Unit Nurse. She has extensive experience in dermatology, pediatrics, plastic surgery, oncology and orthopedic nursing. She attended Vanderbilt University School of Nursing

The entire DebRA Nurse Educator Program is made possible through the generosity of National Rehab, Inc., the Pascucci Family Foundation, the Jennifer DePrizio Memorial Golf Tournament, NYBOT Futures and Options for Kids, Michael and Madeline Weiner, Connie and Paul Flowers, and other DebRA supporters.

and is a Magna Cum Laude graduate of Aquinas College, Nashville, TN.

"Adele brings nursing experience from several fields that are directly relevant to the varied and complex issues that arise in EB," Dr. Fine said. "This Nurse Educator position presents unique challenges as it demands someone with a range of nursing skills and a strong research orientation married with an overriding sense of caring for the EB community. This is not an easy combination to find but Adele possesses all these qualifications."

CINCINNATI CHILDREN'S HOSTS NURSING POSITION

The other EB Nurse Educator position will benefit from DebRA's partnership

with Cincinnati Children's Hospital. Ms. Mancuso is working under the supervision of DebRA Scientific Advisory Board Member Anne Lucky, MD, the co-director of the hospital's Epidermolysis Bullosa Center.

"Basing the position at Cincinnati Children's will enable us to access the depth of EB knowledge in their outstanding program," Cohen said. "Geri will be working in tandem with Adele to assist our families and together they will give us a strong nursing team."

Ms. Mancuso has extensive pediatrics experience as a school nurse and nurse at South Bronx Children's Health Center. Prior to joining Cincinnati Children's Hospital, she was the Clinical Nurse Manager of the Hope Center at St. John's Riverside Hospital in Yonkers NY, where she managed the facility's outpatient AIDS clinic. In addition, Ms. Mancuso speaks Spanish and will help DebRA meet the growing demand for outreach to Spanish-speaking families, according to Cohen. Ms. Mancuso earned a bachelor's degree in nursing from Hunter College in New York City.

"Geri's pediatrics background and skills in managing the cases of people of all ages with a serious, chronic disease gives her a strong grounding in the issues EB families face," Dr. Lucky said.

To contact a Nurse Educator, please call 866-DEBRA76 Monday to Friday from 9 a.m. to 5 p.m. Eastern Time or write nurse@debra.org.

Need more up-to-date and accurate information?

Visit our website at
www.debra.org

Genetic Testing Q&A

Editor's Note: the following article is based on an interview with Ellen Pfindner, PhD, Director, Epidermolysis Bullosa Diagnostics Program, GeneDx

Q. WHAT TESTS ARE USED TO DIAGNOSE EB?

A. For proper diagnosis, a skin sample must be taken by a dermatologist and sent to a specialized laboratory for analysis. This allows the pathologist to determine the cleavage plane in the skin which determines the EB subtype. After that has been determined, DNA analysis may be done to identify the specific genetic mutation present and to determine the mode of inheritance (recessive vs. dominant). This is helpful in regard to future family planning.

Q. HOW ARE THE GENETIC TESTS USEFUL?

A. When there is no family history of EB, finding the particular mutation helps determine the risk that EB will occur in a future pregnancy and is needed for prenatal testing. The results also tell us something about the severity of the disease because it tells us the subtype of EB present. In some cases, knowing the mutation can also help determine how severe the disease will be in the future as all types of EB show a range of severity depending upon the mutations in the affected individual.

Q. WITH A PRENATAL TEST, WHAT PREPARATION AND TIMEFRAMES NEED TO BE FOLLOWED?

A. If the affected individual has not previously had their mutation identified, he or she should be biopsied to determine their type of EB. A blood sample from the affected individual should be sent for mutation detection to identify mutations six months before a pregnancy is started.

Once the mother becomes pregnant and the obstetrician determines how many weeks old the fetus is by ultrasound, then chorionic villus sampling (CVS) is scheduled at 10-12 weeks or an amniocentesis is scheduled at 16-19 weeks. The sample is taken by obstetrician under ultrasound guidance and sent to hospital lab where a portion of the sample may be retained for chromosome studies.

CVS samples are dissected, while amniocentesis samples can be used directly and both are sent to a DNA diagnostic lab where DNA is extracted. At this point, tests are done for specific mutations in that family. Turnaround time is usually 2 weeks for the lab to read and report the results to the physician.

Only labs that offer the prenatal genetic testing for EB can perform this testing. Currently GeneDx is the only lab in the USA that offers prenatal testing for EB.

Q. WHAT ARE THE LIMITATIONS OF THE GENETIC TESTS? WHY?

A. First a biopsy needs to be done to determine the EB type. The genetic tests are designed to sequence the gene that is implicated in the disorder. For EB there are ten different genes, so the subtype; either JEB, DEB, EBS, EB-PA, EB-MD must be determined in order to narrow down the list of gene candidates for sequencing. Second, the sequencing should be done on a sample from a related, affected individual. Sometimes samples from the parents can be used but if the parents are not carriers as in the case of a new mutation, the test will not detect any mutations. Finally, there are cases where no mutations are identified because the mutations lie outside what we call the coding portion of the gene, in regions involved in gene control. Since the consequences of changes in those regions cannot be predicted by the geneticist those regions are not studied by typical genetic testing labs. And it is important for the laboratory to have accurate information about the family—who the biological parents are and who is affected in order to perform the testing accurately.

Q. WHAT ARE THE COMMON MISCONCEPTIONS ABOUT THESE TESTS?

A. The main misconception in EB testing is that we can always determine how severe the disease will be. Often we can, but there are plenty of times when we are surprised by how mild the disease is when the mutations we see should predict severe disease. For this reason we

continued on page 6

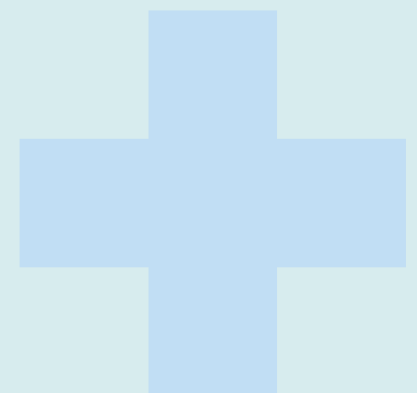
Supplies to SHARE

DebRA's Wound Care Clearing-house accepts donations of wound care products which are passed on, free of charge, to people in need. Please contact DebRA at 866-Debra76 or send an email to nurse@debra.org.

Availability of supplies varies; contact DebRA to discuss your specific needs.

AVAILABLE SUPPLIES

- **Vigilon**
- **Kendall Kerlix Super Sponges**
- **Pedialite**
- **Kendall Conform 4 inch rolled gauze**
- **Enteral nutrition bags**
- **Adaptic**
- **8x8 Allevyn Non-adhesive dressing**



• Ask the DebRA Nurse Educators •

Dear DebRA Nurse:

My son gets blisters and skin breaks down when transported in a car seat. I have protective foam and gel packs that I place under the belts so they won't irritate his skin but I'm looking for any other tips that you may have regarding things to use to help prevent blistering from the car seat. I am seeking any tips or experience you have with this.

**Signed,
Traveling Mom**

Dear Traveling Mom:

First let me begin by saying that car seats are a must for transporting any child or infant in an automobile. The risks of serious injury or death due to the improper or lack of use of car seats are far greater than the blistering that might occur from having your child properly restrained.

Let me start with the car seat itself. Infant seats should be easy to remove from the car and to install properly back into the car when you are ready to go on the road. Some causes for blistering in a car seat or child restraint device are the seatbelt(s)

rubbing and the heat. I have learned from EB moms that the sheepskin seatbelt covers available in many discount stores (such as WalMart) are perfect for covering the straps of the car seat. The adult seatbelt covers can be cut to size to fit over the child seat straps. They cushion the straps and do not hold heat like some other types of strap covers. Sheep skin or fleece car seat covers which are under the child also provide good padding and protection for the baby. A thin piece of foam can be placed under the fleece to add additional padding (use caution not to obstruct any of the working parts of the child restraint device). Fleece can also be used to cover toddler seats and older child booster seats and seatbelts.

It is important that the car seat and the car itself are not hot when you put the child in. If possible remove the car seat, particularly infant seat, from the car and take it inside. (BE SURE TO FASTEN THE SEAT BACK INTO THE CAR EACH TIME CAREFULLY FOLLOWING MANUFACTURERS INSTRUCTIONS). In hotter weather be sure to cool the car before placing the child inside as heat can cause increased blistering.

So remember, PAD, COOL, FASTEN CORRECTLY and SAFE TRAVELS!



Conference Seeks to Clear Research Obstacles

The conference "Obstacles to Translation" was held earlier this year at University of California, San Francisco with the aim of exploring the difficulties associated in translating what is known genetically and scientifically about skin diseases into actual treatments for a variety of diseases. The conference, of which DebRA of America was a sponsor, gathered scientists, physicians, pharmaceutical industry executives, government representatives and non-profit executives to hear talks on current research in a number of rare skin diseases, including EB, and the roadblocks faced. Then the participants were called upon to form groups to brainstorm possible ways around these roadblocks.

The four types of impediments to progress discussed were barriers to identifying and producing useful compounds to treat diseases, obstacles in treating skin diseases, regulatory issues and financial issues.

The organizers of the Conference were DebRA Scientific Advisory Board member Ervin Epstein, Jr., MD, Professor of Dermatology, UCSF; Barbara Gilchrest,

MD, Professor and Chair of Department of Dermatology, Boston University School of Medicine Boston University; and Leonard Milstone, MD, Professor of Dermatology, Yale University School of Medicine. The organizers moderated the productive and lively discussions after each talk.

There was extensive discussion that large pharmaceutical companies no longer fund the early stages of drug discovery and development in rare diseases and do not want to become involved until a compound shows demonstrated promise in humans. This leaves biotechnology companies to pick up the basic, and extremely costly, investment in such vital research. Participants felt that if a compound could be used for an additional purpose for a larger disease (e.g. cancer, diabetes) there was greater hope of it gaining the financial support needed from drug discovery to marketing.

"While pharmaceutical companies can no longer be relied upon to find the effective treatments and a possible cure for EB, there is tremendous hope," DebRA

Executive Director Suzanne Cohen said. "We have seen interest from biotech and medical device firms in exploring the benefits of their products for EB patients. And, today there is a robust international research program that attracts a scientific community dedicating their lives and careers to EB."

"It was productive to gather the key leaders from the fields involved in every step from bringing discoveries in the lab to the pharmacy," Dr. Epstein said. "In inherited skin diseases like EB, there are many challenges on the road to treatments and a cure, so this meeting helped us take a step toward clearing the obstacles and advancing research."

The hope was to have a set of action steps emerge from the conference but it was found there was need for additional work before that could come to fruition, according to Dr. Epstein. As a result, a follow-up workshop will be held in conjunction with the 2007 American Academy of Dermatology meeting.

Q&A continued from page 4

are cautious about predicting severity especially in newborns.

There is also a misconception that we can predict whether a fetus is affected without testing the affected family member. This is incorrect. It is important to have the mutations identified in the affected family member before a pregnancy is started because if you wait until you are already pregnant it is possible we cannot find the mutation in time for a prenatal test especially if you plan to have a CVS.

Q. WHAT DOES IT COST? COVERED BY INSURANCE?

A. The cost of testing depends upon the gene. For big complex genes like COL7A1 involved in DEB, there are 118 separate regions that must be sequenced. So the cost is higher (\$5500 currently) than say for LAMB3 one of the genes involved in JEB where there are 22

separate regions to sequence (\$2500 currently). In addition in JEB if the mutations are not in LAMB3 there are 3 other gene candidates each with a separate charge. So it can be very expensive if you have to do all four genes (more than \$10,000). It is frequently covered by insurance but you have to check with the insurance company. They will ask for the cpt4 codes which are listed for each gene test on the GeneDx website and once they know these they can tell you how much of the testing will be covered..

Q. WHERE DOES SOMEONE GO TO BE TESTED?

A. GeneDx is the only company in the USA offering comprehensive EB testing. There are also a few laboratories in Europe that do certain genes. The GeneDx website (www.genedx.com) has information about testing and all the

forms you will need. Payment is expected when a sample is submitted or you may include a letter giving written approval from the insurance company. You can arrange for testing yourself by having your family doctor draw blood and sending it to GeneDx with the appropriate forms and payment. The process is often easier and more understandable if you meet with a geneticist or genetic counselor to arrange the blood draw and get the forms filled out and to handle shipping. In addition, they can explain what the testing will tell you and explain what the results mean. Some dermatologists can do this as well, but that is not the rule. Once the sample is submitted it will take 6-12 weeks to get a result and the result is always sent to the physician or genetic counselor. Prenatal samples are accepted only through an obstetrician or genetics professional.

DebRA Events

EVENTS AROUND THE COUNTRY SUPPORT DEBRA'S MISSION

The programs and services of DebRA are made possible in large part to the generosity of the individuals who support its mission. Throughout the year, many people hold events from which the proceeds benefit DebRA. The following is a round-up of recent events. If you plan to hold an event to benefit DebRA, the staff is happy to assist you. If you would like your event included in a future newsletter, please send information to Current Newsletter, DebRA of America, 5 West 36th Street, Suite 404, New York, NY 10018, or via email to staff@debra.org.



Jennifer Kirk Deprizio

JENNIFER DEPRIZIO MEMORIAL GOLF TOURNAMENT

"Grammy" Shirley Jones held her first Golf Tournament in Melbourne, Florida in January in memory of her granddaughter Jennifer DePrizio, a beloved member of the EB community who had Recessive Dystrophic EB. A field of golfers braved an initial downpour to play 18 holes of golf on the Gary Player designed Baytree National Golf Links followed by a sumptuous steak dinner donated by Outback Steakhouse. Proceeds of the event, whose key sponsor was National Rehab, benefited the EB Nurse Educator Program, which includes the EB Registry at Vanderbilt University. Other sponsors included Molnycke, MP Total Care Medical, RJ Cameron Construction, Mike Erdman Toyota Scion, UBS, Hometown News and Direct Medical. A second annual event is being planned for Saturday, January 20, 2007.

GOLFING FOR EB

In late 2005, the Furjes family of Missouri held their third annual golf tournament in honor of their daughter, Ella, who has EB Simplex-Dowling Meara. The field of golfers hit the links at Sunset Lakes in Fenton, MO. Proceeds of the tournament benefited the DebRA International Research Grants Program and DebRA of America program and services.

FISH RODEO

Mandy and Tod Taylor of Summerville, Georgia teamed again with the Waterville Baptist Church and held their fourth annual Fish Rodeo. More than 300 anglers showed up to raise funds and compete for prizes and enjoy a variety of games and activities. The event is held in honor of Taylor's son, Grant, who has Recessive Dystrophic EB.

"A DELICATE AFTERNOON" FASHION SHOW LUNCHEON

Lindsey and Jason Gregg of OH held its first fashion show at Miami University of Ohio in honor of their three-year-old son, Max, who has Dominant Dystrophic EB. Max's uncle Travis Gregg, an Indy Car driver, was on hand for the festivities which included a silent auction. Proceeds of the event will benefit the New Family Advocate Program.

5K EMERALD NUTS WALK

Samantha Hotch of CA raised funds for DebRA by seeking sponsorships for her participation in this walk. Samantha has no personal connection to EB but was captivated by "The Boy Whose Skin Fell Off". She completed the walk while six months pregnant.



Samantha Hotch

SLUSHY SALE



Erik Wilander

Erik Wilander, an eight-year-old with EB Simplex, used his entrepreneurial skills to mobilize classmates to raise money through a slushy and cookie sale at his school in Idaho. The sale was part of the school's Crazy Hair Day.

DRESS DOWN DAY

Theresa Evans of New Jersey sold DebRA EB Awareness bracelets at her daughter Kristin's high school as part of an area Dress Down Day for EB. More than 2,000 students participated in the festivities.

WALK-A-THONS

Middle school students at Friends Academy in Long Island made DebRA one of the beneficiaries of its annual Walk-a-thon. Students at Pentucket Lake Elementary School in Haverhill, Massachusetts made DebRA the beneficiary of its Walk-a-thon in honor of their fellow student Jonathon Connolly who has Recessive Dystrophic EB.

TREY WEEK

The Jackson Family of Wisconsin launched an EB awareness effort in memory of their son Trey, who had Junctional EB. Their outreach included media interviews, as well as fundraising through bake sales and EB awareness band and butterfly balloon sales.



Julian Jackson with Trey

FAIRLEIGH DICKINSON UNIVERSITY

The Campus at Florham Senior Business Organization fundraising project was a cocktail reception and Chinese auction at the historic Mansion, the former Vanderbilt family home that is the centerpiece of the New Jersey campus. Tom Murphy, a business student, had the winning project proposal which enabled him to host the event to benefit DebRA. Tom's cousin Robbie Twible has Recessive Dystrophic EB.

Wilander Fall Event A Success

A glorious October day provided the perfect backdrop for the 8th Annual Mats Wilander Celebrity Tennis and Golf Classic in Rye, NY. The well-attended day-long event kicked off with tennis enthusiasts participating in a clinic with pro legends, Mats Wilander, Jim Courier and Tim Mayotte and being treated to an exhibition match between Wilander and Courier. The afternoon saw golfers hit the links for a round of 18 holes on the famed PGA West Course of Westchester Country Club.

In the evening, guests donned their finest attire for a dinner, silent and live auctions, and dancing. With several EB families on hand, Karen Peshkur of Long Island spoke on behalf of her family and movingly shared what it is like to be the parent of a child with EB.

"Over the past eight years, this event has raised a total of nearly \$2 million for DebRA programs and services," Executive Director Suzanne Cohen said. "Sonya and Mats' involvement have made it a marquee event for the organization and the results have enabled DebRA to continue to keep pace

with the growing needs of the EB community."

The event was made possible through the hard work of DebRA staff and the event committee, chaired by Thomas Misco, with assistance from committee members Faith

great success -- Gold Sponsors: Molnlycke Healthcare, National Rehab and Brad Rasmussen; Silver Sponsors: Cambrex, Direct Medical; Hole-In-One Contest Sponsor: Tom & Kathy Shannon & Family and the T-Bird Restaurant Group; Bloomin' Hole: Outback

Advisors. In addition, Curry Automotive donated a 2007 Acura as the prize for the hole-in-one contest and Southeby's auctioneer Jamie Ritchie donated his time for running the live auction. For a full list of event supporters see the Mats Wilander event section of the DebRA website at www.debra.org.



Mats Wilander

Daniels, Cindy Henderson, Jeanne Humphrey, Frank Kacmarsky, Karen Misco, Ruth and Paul Reimer, Mark Vereby and Mats and Sonya Wilander.

The generous support of these event sponsors (as of press time) helped make it a

Restaurant; Tournament Bag Sponsor: Loews Corporation; Cocktail Reception Sponsor: Tom & Kathy Shannon & Family and the T-Bird Restaurant Group; Tee Sponsors: Allcare Medical, Atlantic Business Products, Gus' Franklin Park Restaurant and Nottingham

The volunteers who helped the event run smoothly included Jody Carfi, Janet Chan, Lisa Disa, Kathleen Jay, Linda Kacmarsky, Geri Kelly-Mancuso, Jackie Medina, Charlee Miller, Leslie Rader, Pat RonRon, Bonnie Veilleux, Lisa Stumpf, Matt Polchinski, Nick Guido, Collette Hall, Ian Morgan, Scott Reimer, and Madeline Weiner.

The day's events were made complete by EJ Carfi, a young man with EB, who proved to be a tireless and effective salesman making his way on the golf course and through the dinner crowds to sell an impressive number of mulligans and raffle tickets.



PROFILES IN EB

Anna Dittman—Born A Fighter



Anna (3rd from left) with family

At the tender age of four Anna Dittman has had experiences that most people wait a lifetime to fulfill. In her role as Ohio's representative in Champions Across America, a national Children's Miracle

Network program, Anna has met such diverse VIPs as President Bush, retired San Francisco 49ers Quarterback Steve Young, Marie Osmond and The Wiggles.

The Hudson, Ohio native has EB Simplex-Dowling Meara and was chosen to serve as an ambassador for the millions of kids treated each year at children's hospitals across the country. Anna is an energetic and outgoing girl and her participation in the program helped the public learn about EB and its challenges.

Anna was one of 50 children who made special visits to

Capitol Hill and The White House. She participated in segments pre-taped at Walt Disney World in Orlando for the internationally televised "Children's Miracle Network Celebration". Back home in Ohio Anna's story was chronicled in the Hospital's video called "Born A Fighter" and she also was presented with a medal for her courage in dealing with EB. The 2005 Columbus area Children's Miracle Network telethon raised a record amount in excess of \$7 million.

Anna's story helped highlight the important work of children's hospitals as she was

first treated at Columbus Children's Hospital. At the time, she had open wounds and blisters over 90% of her body. Anna's mother, Wendy, recalled recently that "seeing Anna stronger and healthier today and watching her receive that medal gave us perspective on everything we've gone through and where we are today."

Children's Miracle Network is an alliance of premier hospitals for children that is a non-profit organization dedicated to saving and improving the lives of children by raising funds for children's hospitals across North America.

Tailormade wound care



Mölnlycke Health Care dressings with **Safetac®** soft silicone technology do not adhere to the moist wound, yet adhere gently to the surrounding skin and therefore minimize trauma and pain at removal. **Safetac®** is a patented soft silicone adhesive technology designed specifically for wound management dressings.

Mölnlycke Health Care products featuring Safetac® soft silicone technology include:

- Mepitel®**
- Mepilex®**
- Mepilex® Lite**
- Mepilex® Border**
- Mepilex® Border Lite**
- Mepilex® Transfer**

Mölnlycke Health Care

EB Research Grant to Explore Wound Healing



DebRA of America takes several avenues to foster research into the causes and treatments for EB. While it supports the extensive international portfolio of research grants given through DebRA International, it also supports an EB-focused Dermatology Foundation Research Grant to a U.S. scientist.

In 2006, Fatima Rouan, PhD, Assistant Professor of Dermatology and Cutaneous Biology at Thomas Jefferson University in Philadelphia was awarded a research grant to explore the role bone marrow and cord blood mesenchymal stem cells play in skin wound healing. Dr. Rouan hopes to establish a novel therapy approach using these cells as a tool to cure skin blisters among other affected tissues with EB. Dr. Rouan chose this approach as cord blood stem cells have been used to treat other heritable diseases.

"I chose this approach for EB as it has proven to be effective in wound healing for other diseases," Dr. Rouan said. "What we are exploring is just the first step in a series of questions we need to answer about wound healing in EB. And we must always consider that what is promising in a mouse model may not always translate to diminishing or preventing EB in people, but this project will give us new insights that are valuable."

Dr. Rouan is hoping to expand the role these cells can play in different forms of EB, as they are already known to play a role in normal wound healing and regeneration. The project will be conducted with normal and immunodeficient mice afflicted with skin wounds or skin blisters that will be injected with umbilical cord blood-mesenchymal stem cells (UCB-MSK) either directly into wounded areas or systemically via tail vein. Non-injected

Dr. Rouan chose this approach as cord blood stem cells have been used to treat other heritable diseases.

mice will serve as controls.

Once injected, the mice will be monitored to see if the newly introduced UCB-MSK leads to skin repair. Skin biopsies of wounded areas will be collected and assessed. At the same time, Dr. Rouan will extend her studies to transgenic mice that the Jefferson investigators have developed

to mimic dystrophic and junctional forms of EB.

To determine the role of UCB-MSK in EB skin blistering repair at early stage of development and whether they promote longevity of EB mice after birth, she will transplant mesenchymal stem cells alone or expressing type 7 collagen (for COL7A1^{-/-} mice) or laminin 5 proteins (for LAMB3^{-/-} and LAMC2^{-/-} mice) into mouse embryos. Collected skin samples before and after birth will be used to detect integration and stability of the UCB-MSK in mouse epidermis and the outcome of these two approaches will be compared in terms of skin repair and mice longevity.

Dr. Rouan joined Dr. Jouni Uitto's laboratory at Thomas Jefferson University in the Fall of 1997 because of her interest in gaining expertise in the molecular biology and genetics of inherited skin diseases. In addition, she wanted to pursue a career path that had close ties to patients. She began working in EB doing mutation analysis, particularly in the variant that manifests with skin blistering and later on with muscle weakness. She has also worked on various aspects of Junctional forms of EB.

Dr. Rouan completed her undergraduate degree in her native country Morocco before earning a Master's degree and PhD in biology from Temple University in Philadelphia. She then completed post doctoral training at the University of Pennsylvania before joining Dr. Uitto's laboratory and the EB field at Thomas Jefferson University.

Have you remembered
DebRA in your will?



NR National Rehab

Specialists in Healthcare Products and Services

The Leading Provider in Wound Care For:

- Brand Neutral Product Selection
- Direct Shipping to Patients' Homes Nationwide
- Direct Billing to all Major Insurers And Government Programs
- Fast/Reliable/Knowledgeable Service

Customer Service:

Phone: 1-800-451-6510

Fax: 1-800-749-0711

www.nationalrehab.com

National Rehab Corporate Headquarters

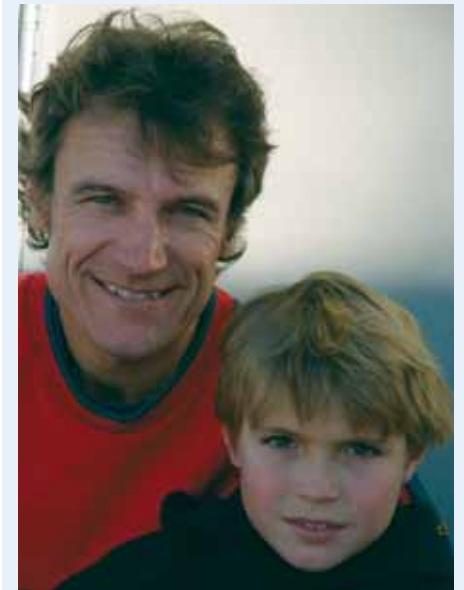
Airside Business Park

540 Lindbergh Drive

Moon Township, PA 15108

Serving the Nation!

Shop the DebRA online store for EB bears and other awareness items at www.debra.org



Mats Wilander appeared on NBC's Today show to discuss his work on behalf of DebRA and to inform the public about EB. He also described the Mats Wilander Celebrity Tennis & Golf Classic.

www.debra.org



Direct Medical Inc. specializes in a full range of medical products specially developed for patients with Epidermolysis Bullosa

www.ebdressings.com

(800) 659 8037

**Your One Stop
for EB Supplies**

Mepitel Silicone contact layer
Mepilex foam dressing
Mepilex Transfer
Roll Gauze Bandages
Healing ointment
Sterile Gauze
Acticoat Absorbent Dressing

- Direct Medical, Inc. offers special discount prices for EB Families.
- Direct Medical can bill Medicare, Medicaid and Private Insurance.
- Please call for more information. 1 (800) 659 8037

DebRA

DebRA of America, Inc.
5 West 36th Street, Suite 404
New York, New York 10018

PRESORTED
STANDARD MAIL
U.S. POSTAGE
PAID
BENSALEM, PA
PERMIT #118