

DeBRA Currents

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

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New EB Classification

The following article is extremely technical. A number of families expressed interest in the re-classification of EB and as a result, Dr. Jo-David Fine provided the most up-to-date information. Dr. Fine's book is available for pre-order on Amazon.com. It can be found by typing his name into the search engine.

The Classification of Inherited Epidermolysis Bullosa (EB): Report on the Third International Consensus Meeting on Diagnosis and Classification of EB.

Jo-David Fine et al.

In the last eight years, the advances in science that address the spectrum of inherited Epidermolysis Bullosa (EB) have been substantial. On May 19, 2007, 18 leading authorities met to review the classification

system of EB and update it to reflect current knowledge. The findings of this meeting were published in the American Academy of Dermatology in June 2008.

This Consensus meeting resulted in an expansion of EB's classification to include other disorders associated with mechanical fragility of the skin. Kindler Syndrome is now classified as a fourth subtype. Kindler syndrome is an autosomal recessive disorder involving kindling, and the cleavage plan varies in these individuals.

The Junctional subtype was expanded to include laryngo-onycho-cutaneous syndrome

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Level of skin cleavage	Major EB type	Known targeted protein(s)
Intraepidermal ("epidermolytic")	EBS	Keratins 5 and 14; plectin; *6β4 integrin; plakophilin-1; desmoplakin
Intra-lamina lucida ("lamina lucidolytic")	JEB	Laminin-332 (laminin 5); type XVII collagen; *6β4 integrin
Sub-lamina densa ("dermolytic")	DEB	Type VII collagen
Mixed	Kindler syndrome	Kindlin-1

LETTER FROM THE EXECUTIVE DIRECTOR

Summer is usually the time of year when the pace slows. Not at DebRA!

At the end of June, DebRA held its biennial Patient Care Conference themed **“NEW HEIGHTS, NEW HOPE”** in Denver, Colorado. Through the gracious support of sponsors, volunteers and an anonymous angel, we welcomed 275 adults and 44 children and 23 speakers to the beautiful Denver Renaissance Hotel. Registration spiked once it was announced that Dr. John Wagner of the University of Minnesota would be a guest speaker.

DebRA's **PCC IS THE ONLY FREE NATIONAL CONFERENCE FOR EB FAMILIES**. Yet in these days of high gas prices, and a “tight” economy many families fund-raised in their local community to pay for travel expense. The opening reception was a great opportunity to see old friends and make new ones. Lunches each day reunited parents and kids who had to be pried from the fabulous Kids Room staffed by volunteer nurses, siblings and local Denver residents who just “wanted to help” make the experience special for our EB kids. Without strong sponsorships, this conference would not be possible. We are grateful to Hollister Woundcare, our Title Sponsor; Mölnlycke, our Gold Sponsor and National Rehab, this year's Silver Sponsor. We welcomed Byram Healthcare, Direct Medical and TTW as exhibitors.

DebRA provided a binder for each family with agenda, speaker biographies and topic information. The city provided a very complete guidebook—though

we kept everyone very busy during the day. Macy's provided discount coupons.

Conference Feedback is important to DebRA. This year we will use **SURVEY MONKEY (WWW.SURVEYMONKEY.COM)** for feedback, and to learn more about what is important to EB families. It will guide us in many ways beyond conference feedback. It will guide our work and better serve your needs. Please take time to complete it and return it to Survey Monkey. Individual responses are anonymous.

Our friends at National Rehab fully funded taping all the Conference sessions. The four DVD set will be available free through DebRA's website and National Rehab www.nationalrehab.com. We are looking at a six-week turnaround time. It is the first time the Conference will be available to everyone—indexed on the DVD set to take you to the information you need. Terrific!

MARK YOUR CALENDAR for **MONDAY, OCTOBER 20TH, 2008** for the **TENTH ANNUAL MATS WILANDER GOLF AND TENNIS EVENT** at the Westchester Country Club. It is Debra's major fund-raising event. There is always room for talent on the Event Committee to secure the perfect auction items, fill the foursomes, play tennis with the greats, all followed by a wonderful dinner and dancing. Roger Stern has graciously agreed to chair the event. Our corporate honoree is Ron Duckstein—a wonderful DebRA supporter. We are also delighted to have a Spirit Award winner we met at the Denver Conference. We actually modified the Conference schedule so everyone in Denver could meet Jassamine. She is a very special young woman. Watch for the invite.

CONSIDER BECOMING A SPONSOR.



Mary Sprague

DONATE. VOLUNTEER. We need all of you to make this event the best ever. If you would like an online version of the invitation, visit our website at www.debra.org. We all have contacts, are members of social and professional groups. Copy the link and send it to your network.

We have been **SEARCHING FOR NEW OFFICE SPACE**. We have a lease in the works, so wish us luck! Other projects include re-branding DebRA to better illustrate the quality of the work we do in patient/family support and research. I believe we have a great story to tell, and should be proud of what we have accomplished and our vision for the future.

I am very happy to be a part of the DebRA family and the EB community. I welcome your ideas and input. My email is msprague@debra.org. Please stay in touch.

Warm regards,
Mary

• Ask the DebRA Nurse Educator •

Dear DebRA Nurse:

My firstborn child has a severe form of EB. Our genetic test results say it is a recessive form and both my husband and I carry the gene mutation. I have been told that there is a method, PGD, which can ensure I have an EB free baby when I have our next child. Can you tell me how it works?

New to EB

Dear New:

Having the genetic mutation of your child's EB identified is the first step in Preimplantation Genetic Diagnosis (PGD), improving your chances of having an EB free child.

PGD is accomplished with in-vitro fertilization, in which the mother takes hormones to increase the number of eggs she produces during her monthly cycle. These matured eggs are surgically removed and then fertilized with the father's sperm. When the

fertilized egg reaches the eight cell stage, one of the cells is removed and analyzed at a special laboratory to see if it carries the EB mutations that were identified in the family. The fertilized eggs that do not carry the mutations can then be implanted. If pregnancy results from the implantation process, the child will be EB free, but may also be a carrier like the parents.

This process has been used successfully in a number of EB families. However, it is very expensive and not always covered by standard health insurance. Successful pregnancy with live birth is achieved in approximately one third of PGD cycles. As with any medical procedure, you should discuss with your medical provider whether PGD is right for you.

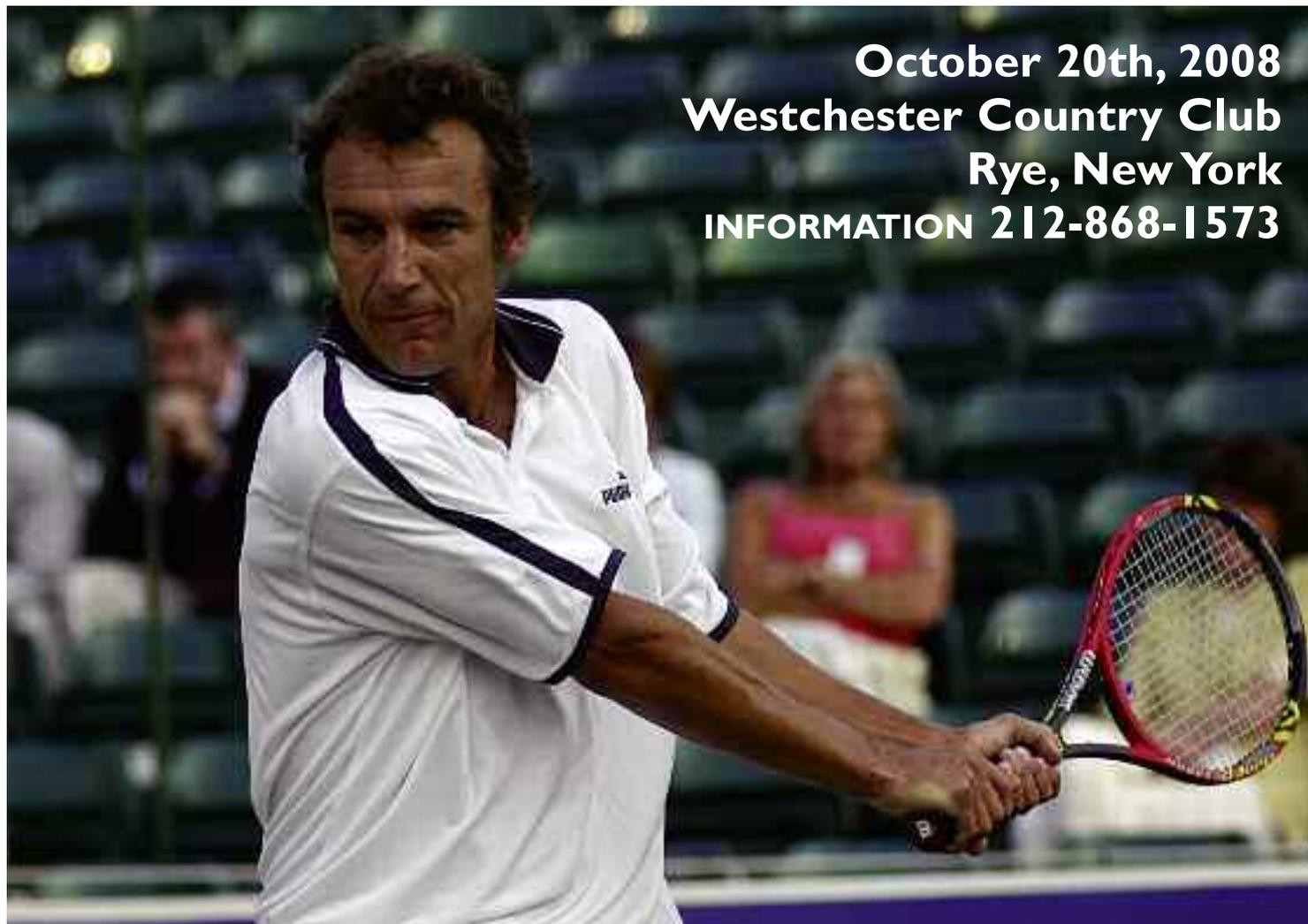
Geraldine Kelly-Mancuso, RN

For more information on Genetics and EB, you can go to www.genetests.com and at GeneReview, search for Epidermolysis Bullosa. Currently there are articles by Ellen G Pfendner, PhD and Anne W Lucky, MD on EBS, DEB, JEB and EB-PA available.

EB Tips for the Summer Months

- Sun-protective clothing is important for all family members, both with EB and EB-free.
- Ocean water and pool water swimming is okay for those with EB.
- Spray on sunscreen is great because it is easy to use. Pick a brand intended for sensitive skin.
- Use very light-weight sunglasses that block UVA/UVB—thin strips of a non-stick dressing can be used behind the ears and at the bridge of the nose if necessary.
- Many families have success with water shoes, salt water sandals and Croc's—find the ones that fit your needs.
- Heat can cause increased blistering—many folks with EB opt for air conditioning where available.

10th Annual Mats Wilander Celebrity Tennis & Golf Classic



October 20th, 2008
Westchester Country Club
Rye, New York
INFORMATION 212-868-1573



The day begins with a delicious continental breakfast in the early morning followed by a tennis clinic with Mats Wilander, John McEnroe and a host of internationally-known tennis champions.

Mats and John will play an exhibition match befitting their world famous status in the tennis arena. There are also fast serve and shot-making competitions for tennis participants, as well as an afternoon tournament.

Following an elaborate brunch and a mid-morning shotgun start, golfers join celebrities on the famous West Course for some of the best 18 holes on the PGA Tour.

After a full day on the courts and greens, our guests slip into evening wear for an elegant cocktail reception with a silent auction before heading inside to a three-course dinner and dancing to live music by New York City's hottest dance band, The Stingers. This year promises to be as fun and exciting as ever.

[continued on next page](#)



Tennis & Golf Classic continued from page 4



Ticket Prices

Golf and tennis tickets include all day dining, reception and dinner

Golf Foursome	\$2,750
Golf Individual	\$750
Tennis	\$400
Reception/Dinner only	\$250
Corporate Table of 10 guests	\$2,500

EVENT JOURNAL ADVERTISING

Advertising opportunities range from \$250 for a business card size ad to \$3,000 for a 4-color cover.

SPONSORSHIP OPPORTUNITIES

Sponsorship opportunities range from \$350 for the sponsorship of a tee or green to Platinum sponsorship at \$30,000.

FOR MORE INFORMATION, PLEASE CONTACT:

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5 WEST 36TH STREET, SUITE 404
NEW YORK, NEW YORK 10018
212.868.1573 PHONE • 212.868.9296 FAX

Reserve and register at www.debra.org



Mark Your Calendar

Tenth Annual Mats Wilander Celebrity Tennis and Golf Challenge

Monday, October 20th, 2008

Westchester Country Club

Rye, New York

National EB Awareness Week

October 25th–October 31st, 2008

Watch for updates on our website!

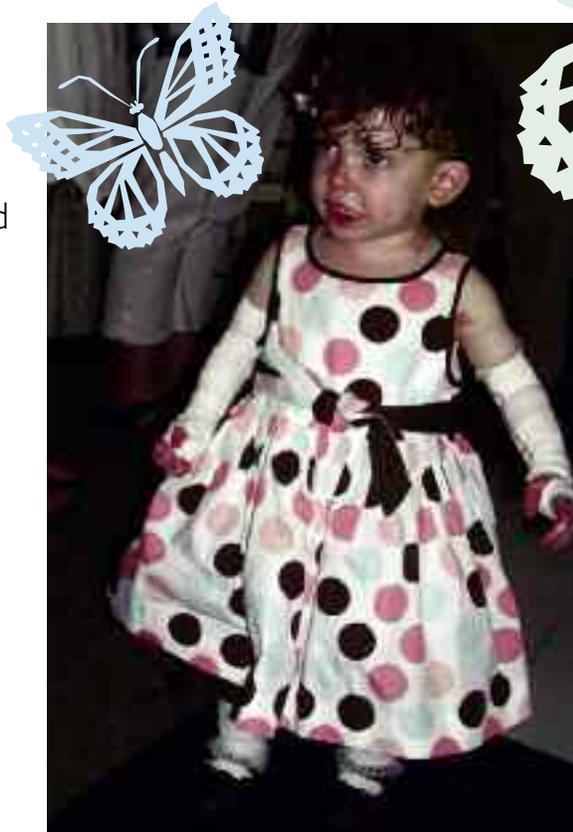


Jassamine Domino has been selected as the 2008 Spirit Award Winner and will be honored at the Mats Wilander Event.

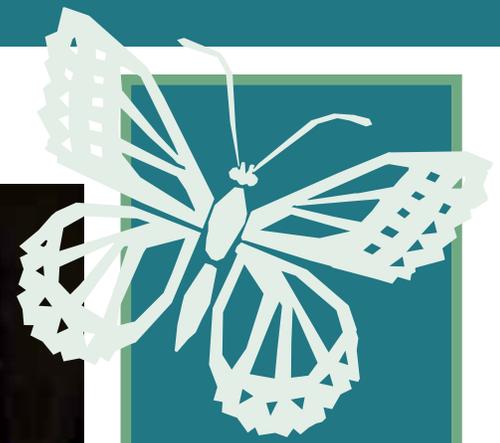
Butterfly Wishes for Ellie

The 2nd annual "Butterfly Wishes for Ellie" event on May 8th, 2008 was a special evening for the EB Community. We had a record turnout of over 150 people, raised over \$30,000, and spread awareness. State Representatives Sharron Cooper and Michele Henson were our honored guests and our champions in trying to promote an EB awareness throughout the State of Georgia. The funds raised for this event benefit DebRA of America and are being used for research and the nurse educator program. We look forward to continuing this event in the years to come.

—ANDREW, SHAWN AND ELLIE TAVANI



Ellie Tavani



DebRA would like to thank the generous sponsors of Butterfly Wishes for Ellie

Allstate Insurance Company

Doctors Marianne and Stephen Garber

John and Dean Phillips

Stuart Shapiro

The Goldstein Family



The Atlanta Swing Orchestra Inc. performs, much to the delight of the guests.



New EB Classification continued from page 1

(LOC or Shabbir's syndrome). LOC is an autosomal recessive disorder seen with increased incidence in Punjab and involves Laminin 332 (formerly Laminin 5).

As stated by the Consensus group, "The rationale for our inclusion of selected new entities as types of EB is (1) that they have friction-induced blisters and other clinical features in common with more established forms of EB; (2) that they are hereditary; and (3) that, from a practical perspective, the patients, especially neonates, and their families, will potentially benefit from the growing resources already available for EB patients." Page 941

The Consensus group also reviewed the testing methods and findings for EB within each. In regard to DNA testing, the group stated "mutational analysis remains a superb research tool. In addition, the eventual application of gene therapy to EB patients will be dependent on the determination of the specific mutations present. It is also the recommended technique whereby prenatal and preimplantation diagnosis can be performed. At the present time, however, it is not considered to be a first-line diagnostic test for EB."

Additionally, the nomenclature has been adjusted in order to ensure uniformity in the diagnosis of EB. The changes are

shown in the chart below.

The outcomes of the reclassifications of other disorders as part of the EB spectrum along with clarification in the nomenclature of EB diagnoses will result in more consistency across medical disciplines.

This is a review of only a small portion of the Classification review. The paper has summary tables about the relative findings, both cutaneous and extracutaneous, for each subtype listed within the new classification scheme. It is highly recommended that those with an interest in EB review the full article.

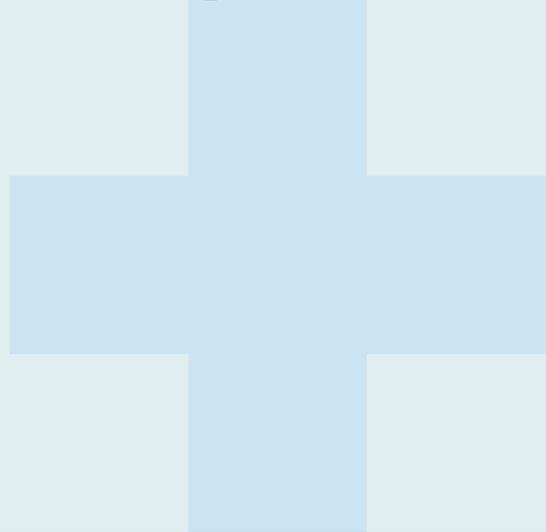
Name	Recommendation	Reason(s)
EBS, Weber-Cockayne	Change to "EBS, localized"	Lack of uniformity in original descriptions; new name has more immediate visual impact
EBS, Koebner	Change to "EBS, generalized other"	Inconsistency in definition even among EB experts; not associated with Koebner phenomenon
EB with pyloric atresia	Separate into "EBS-PA" and "JEB-PA" subtypes	Pyloric atresia may occur rarely with EBS, as well as with JEB
Hemidesmosomal EB	Eliminate this term	Includes only one of two JEB-nH subtypes having identical EM and clinical findings; distinction is based solely on targeted protein
RDEB, Hallopeau-Siemens	Change to "RDEB, severe generalized"	New term has more immediate utility for clinicians
RDEB, non-Hallopeau-Siemens	Change to "RDEB, generalized other"	Consistency in nomenclature
Transient bullous dermolysis of the newborn	Change to "bullous dermolysis of the newborn"	Not always transient; rare patients continue to blister beyond the newborn period or infancy

DebRA staff would like to thank Dr. Jo-David Fine for his assistance in the preparation of this article.

Supplies to SHARE

DeBRA's Wound Care Clearinghouse accepts donations of wound care products which are passed on, free of charge, to people in need. These products are distributed on a first-come-first-serve basis, and supplies vary over time.

Contact DeBRA at 212-868-1573 or send an email to nurse@debra.org. Remember, if you have supplies you no longer need and want to help others, please let DeBRA know. We're always happy to receive donations of non-expired, unopened, non-prescription products to help those in need.



Cards can be purchased by calling the DeBRA office at 212-868-1573 and mailed to you at no additional cost.

SUPPORT OUR ORGANIZATION BY PURCHASING A \$5 MACY'S SHOPPING PASS. WE KEEP 100% OF THE PROCEEDS!

On Saturday, September 20th your exclusive shopping pass entitles you to save 10-20%* on most items including many of Macy's best apparel and accessory brands.

Plus, enjoy a full day of special events, entertainment and a chance to win a \$500 Macy's shopping spree!



*20% regular, sale & clearance women's, men's & kids' apparel & accessories, fine, bridge & fashion jewelry, bed & bath items, housewares, frames, luggage, china, crystal & silver, all kitchen & personal care electrics & technology items
10% regular & sale furniture, mattresses & rugs.

EXCLUDES: Cosmetics and fragrances, fine and fashion watches, designer and bridge handbags (including Coach, Dooney & Bourke, Juicy Couture and Marc by Marc Jacobs), designer and bridge shoes, Louis Vuitton, Polo/Ralph Lauren, Lauren, Tommy Bahama, Lacoste, 28 Shop, Oval Room, Martha Stewart Furniture, Lauren Ralph Lauren Furniture, Waterford, Lalique, Baccarat, Tempur-Pedic, All Clad, Henckels, and Wusthof. Not valid on EDV, Macy's Gift Cards and Gift Certificates, special orders, previous purchases, restaurants and nonmerchandise-related services, Gift Registry kiosks, gift wrap, selected licensed departments or as payment on credit accounts. Discount will be deducted from the current price (regular, sale or clearance, as applicable). Cannot be combined with another savings pass or discount offer, except for new accounts (subject to credit approval). Valid Saturday, September 20, 2008.

Need more up-to-date and accurate information?

Visit our website at www.debra.org

Have you remembered DeBRA in your will?

Jackson Gabriel Silver Fund



Jackson Gabriel Silver

DebRA recently had the opportunity to speak with Alex and Jamie Silver, parents of 9 month old Jackson Gabriel Silver (RDEB). The Silver/Grossman family started a fund in conjunction with DebRA and the Dermatology Foundation that is devoted to providing career development support for a junior investigator pursuing research in the realm of EB, funding additional EB research and helping to secure top notch patient care services for those in need, including support of the DebRA Nurse Educator Program.

How did you become involved with DebRA of America?

JAMIE: We became involved with DebRA of America when Jackson was born. The hospital had never had a child with EB before, and we were told by some doctors to go to the DebRA website and print out materials for them so that they could follow the protocol while he was there.

ALEX: We then reached out to the Executive Director and Board President of DebRA who were both incredibly receptive and got us a new parent kit...DebRA helped us make sense of the world that is EB which was new to us. We have found DebRA to be a very supportive organization which is what has led to our wanting to get involved on the fundraising side and helping out with events as well as my joining the board. It's an organization we want to be a part of, not just a recipient of its services.

JAMIE: DebRA sent a wonderful box of supplies. Some of the materials that they supplied for Jackson...were things that we didn't know of and proved to be very helpful to us those first few weeks we were home. I also send things to the WoundCare Clearinghouse. The things that we received were so helpful.

Have you had the opportunity to take part in any of the services offered by DebRA?

JAMIE: We are in frequent contact with Geri Kelly-Mancuso (DebRA Nurse Educator), who has been a lifesaver. She calls to check in to see how I'm doing and Alex calls her often with questions. What has been so nice is that Geri really seems to understand what it's like to parent a child with these needs and does not say that there is one definitive right way. She's very open to our need to do what feels comfortable and what's best for our child.

ALEX: The other service I've used are the DebRA chatboards - there is a wealth of information that people share as well as updates that I've found helpful especially about practical solutions to everyday complications.

JAMIE: The Occupational Therapist who works with Jackson went to the Patient Care Conference (PCC). It was great for Jackson that his OT learned from the doctors and families there. It really increased this understanding of current practices especially the newest research and different products.

Regarding your strong interest in research: are there any current research projects that have stood out to you recently in research?

ALEX: Overall, it's really wonderful as a parent to see the amount of research going on right now. It gives one a lot of hope that there is light at the end of the tunnel. We are grateful to those who spend their time trying to cure and manage EB. While we are clearly in the learning stages about various research projects, one of the more recent projects we've looked into is Dr. David Woodley's and Mei Chen's project at USC which focuses on replacing the missing collagen VII in someone suffering from DEB with an injectable form of Collagen VII. It has the potential to increase the quality of life greatly. Clearly the stem cell work being pioneered by Dr. Christiano at Columbia sounds very promising as does the work Dr. Wagner is leading at the University of Minnesota on bone marrow transplants though it is in its early stages. Stanford has been focused on gene transfer, and we eagerly await learning more about their trial. Basically we are encouraged that some of the smartest people we have met are trying to cure this disease.

We know that you funded the Jackson Gabriel Silver Fund; can you tell us about its purpose?

ALEX: When we established the JGS fund in collaboration with Edward and Ronnie Grossman (Jackson's grandparents), we did it for two reasons; the first is to fund research to the best of our ability. Given EB affects a relatively small community, it is upon us and all of the other people affected by EB to push to find an ultimate cure, or treatment. The JGS fund will fund a Career Development Award via the Dermatology Foundation. This award will go to a top junior transitioning to senior researcher for three years specifically to work on a cure or treatment for

FREE VIDEO NOW AVAILABLE... *More than Skin Deep*

VHS Tape Format Only, 1999

This 10-minute video features people with EB and parents of children with EB sharing their experiences, as well as brief EB information. \$3.00 shipping and handling to US addresses. Contact staff@debra.org for more information.



COMING SOON... *The Boy Whose Skin Fell Off DVD*

To add your name to the waiting list please go to www.debra.org and click on the Order Video link under Jonny Kennedy's photo.

This award-winning documentary featured on TLC follows Jonny Kennedy, a man who had Recessive Dystrophic EB and died in 2003 age 36 after losing his battle against skin cancer. In his last months, Jonny decided to work with filmmaker Patrick Collerton to document his life and death, and the result was a film that is an uplifting and provocatively humorous story of a singular man. Not shying away from the grim reality of EB, the film was also a celebration of a life lived to the fullest.

FREE DVD AVAILABLE... *What is EB and Your Welcoming Classroom*

A guide for making the school experience supportive and positive for students with Epidermolysis Bullosa.

JGS Fund continued from page 11

EB. The JSG fund will also fund other EB Research. And second, it will fund patient care. We have been fortunate to have access to very good care with our son from the beginning and that's something that should be open to every parent who has a child with EB and being able to do that is very important to us. DebRA has been extremely helpful to us in administering the fund and making it possible for people to donate online directly to it.

As trendsetters highlighting a different kind of organized giving and using this opportunity to inspire others to follow your example, if someone comes to you with an interest in establishing the same type of fund, what advice would you share with them?

Alex: Number one, you just have to do it. It's actually much easier than it seems. We were helped greatly by DebRA who couldn't have made it easier for us. Number two, keep an open mind. Some of the best ideas we've heard have been

from other people...we listen and speak to everyone about research and patient care. And number three; accept the support that people give to you. This wouldn't be possible without the support of our family, loved ones and friends. It gives us a way to show that this is money that is going to go for two clear purposes, patient care and research, and it gets people interested when you can specifically point to something that you have tangibly done. Number one, have a clear goal and also I suggest doing it with an established organization like DebRA giving you instant credibility.

When people find out about the JGS fund, we hope they donate and tell other people about it.

We think it's also important to personalize it. The reason we called it the Jackson Gabriel Silver Fund is to show that there is a living breathing person suffering from this disease. We hope it provides a sense of urgency to push ahead with fundraising and research in real time.

Overall, it's really wonderful as a parent to see the amount of research going on right now. It gives one a lot of hope that there is light at the end of the tunnel

JAMIE: This is a disease that affects such a small group of people that if we don't take it upon ourselves to find treatment and a cure then who is going to?

Is there anything else you would like to share with the EB community?

JAMIE: For me, knowing that there is an organization like DebRA was such a huge relief when Jackson was diagnosed; that there was a resource out there for us to utilize and that there were people out there who would have knowledge about this disease that we had never heard of. I think that for new families it's a really hard adjustment. But I think that most families would agree how grateful they are to have an organization such as DebRA.

Families Reach New Height and New Hopes in Denver



More than 250 people visited Denver for DebRA's 2008 Patient Care Conference (PCC) entitled "New Heights, New Hopes". Denver, the capital of Colorado, played host to the free biennial conference that featured presentations from leading authorities on Epidermolysis Bullosa (EB). Denver was chosen as the 2008 venue due to its strong connection to the EB community through the EB Clinic at Colorado Children's Hospital.

Mary Sprague, the Executive Director of DebRA, opened the conference with a welcome to all families and medical professionals that traveled to attend this one-of-a-kind meeting of families and doctors from around the nation. The four-day conference provided the unique opportunity for experts in the field and families to come together to share information and an open forum to ask questions. It also allows families to meet in person and share their experience and suggestions with each other.

The agenda covered topics including wound care, nutrition, research, physical therapy, entering schools, stem cell treatment, dental care and pain management among many others. National Rehab generously offered to record the sessions and have made them available to the EB community.

While the adults were immersed in the sessions, the children had the opportunity to spend time together working on projects and playing in the Kid's Room. It is due to the amazing volunteers. Their time and seemingly limitless amounts of energy kept the room filled with laughter and the children entertained.

The PCC is free to all attendees and was supported in part, through the generosity of Title Sponsor Hollister Woundcare. Support provided by Gold Sponsor Molnlycke Healthcare and Silver Sponsor National Rehab also helped to make this conference possible. In addition, the conference received general support from Byram Healthcare, Direct Medical, Mr. & Mrs. Mark McCaughtry, Pedors, Smith & Nephew, The Brotman Foundation and Wayne Westland Friends of EB.

The next Patient Care Conference will be held in 2010 with the date and location to be determined. Details will be shared as they are available.

This Conference would not have been possible without all of the Doctors, Nurses, and other wonderful medical professionals and volunteers who worked so closely with us to make this year's Patient Care Conference happen. We will never be able to thank you enough.



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