edlines

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ECTODERMAL DYSPLASIA SOCIETY

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ED Christmas Party 2013

We are pleased to announce the

EDS Christmas Party will be held on

Saturday, 7th December 2013

12.30pm - 5.00pm

At the fully air-conditioned

"Thistle Cheltenham"

Children's Entertainment from Chris Brown and friends

Music and a Bubble Machine

Buffet and Father Christmas

Presentation from Prof. Angus Clarke

Tickets - Adult £12.50 ED children free, non-ED children £6.25

Family and friends welcome

Financial assistance is available towards travel and accommodation on application

The venue has ample parking and is close to the M5 motorway, about 2 miles from Cheltenham coach and rail stations and close to a range of hotels.

Please return the enclosed registration form by ***18th November 2013***

Teenagers welcome – we have had a few 13-15year old who have expressed an interest in coming to the party to meet with other teenagers - the more the merrier!

edlines

Medical Advisory Board Members

Prof. Angus Clarke	-	Clinical Genetics (MAB Chairman)
Prof. John Hobkirk	-	Prosthetic Dentistry (Implants)
Prof. John McGrath	-	Genetics, Molecular Dermatology
Prof. June Nunn	-	Paediatric Dental Surgery
Dr. Helen Stewart	-	Clinical Genetics (IP)
Mr. Colin Willoughby	-	Ophthalmology
Mr. Martin Bailey	-	ENT
Prof. Michael Tipton	-	Human Applied Physiology
Prof. Nichola Rumsey	-	Psychologist
Dr. S. Aylett	-	Paediatric Neurologist
Mr Paul King	-	Restorative Dentistry (incl. Implants)
Prof. John Harper	-	Paediatric Dermatology
Mr. Michael Kuo	-	Consultant Otolaryngologist
Dr. Fiona Browne		Dermatologist
Dr. L. Albery	-	Speech / Language Therapist

Trustees

Paul Collacott	-	Chairman
Alan Waller	-	Treasurer
Diana Perry	-	Secretary
Mandy White	-	Air-Conditioning / School Liaison
Melanie Davis		
Stephen Ayland		
Simon Lees-Jones		
David Wyatt		
Mark Macnair		
Liz Beckmann		
Sharon Cooper		

Staff

Sue Beard	-	Accounts / Website
Julie Cox	-	Administrator
Fergus Gordon	-	Scotland

Membership

A membership form is enclosed

Please remember membership runs from 1st January to 31st December

Please return your form as soon as possible so your membership does not lapse

Temperature Research

We are still trying to obtain funding to carry out this research; as soon as we are successful we will let you know and hopefully proceed to the next stage of the project.

I will send another email of invitation to everyone once we have funding.
Diana Perry

The Ectodermal Dysplasia Society Facebook page is buzzing with chat, lots of questions and answers, tips, support and much more.

Join us and make lots of new friends.

Disability Living Allowance And P.I.P

Contact a Family have a really helpful leaflet on how to apply for DLA and advice on how to complete the forms. You can find the leaflet at

http://www.cafamily.org.uk/media/379427/dla_factsheet.pdf

Diana is always happy to help complete the forms - it's best to get the forms right at the beginning in the hope it will not have to go to appeal or tribunal.

Christmas Party

Directions and accommodation

The venue is The Thistle Hotel, Gloucester Road, Cheltenham GL51 OTS

Directions by car…
Leave the M5 at junction 11 and take the A40 towards Cheltenham. At the next roundabout take the second exit; Thistle Cheltenham is immediately on the left. We have 220 parking spaces available on-site for a small additional charge for all overnight guests. To programme your sat-nav, please use the postcode GL51 0TS

Directions by train (Cheltenham railway station is 1.5 miles from the hotel)
There are taxis available at the station, but if you would prefer to pre-book then we have connections with a local taxi firm, contact us for details.

Directions by coach
Cheltenham coach station is in the town centre. There are taxis available at the station, but if you would prefer to pre-book then we have connections with a local taxi firm, contact us for details.

Accommodation There is wide range of hotels available including:-

Travelodge - Cheltenham 0871 984 6202 (recommended as it is just across the road from the party and has a Harvester and KFC)

Thistle Hotel - Cheltenham 0871 376 9013 (on-site convenient for the party but expensive)

Premier Inn - Cheltenham West 08701 977 055 (Tewkesbury Road, Uckington, Cheltenham, GL51 9SL)

Premier Inn - Cheltenham Central - 08701 977 056 (374 Gloucester Road Cheltenham, GL51 7AY)

Contact Number on the Day for directions – Sue 07885 507238 or **Diana** 07774 465712

If you get lost or need directions please call either Sue or Diana on the numbers above

NB. Please aim to be at the party for 12 O' clock to avoid seating problems and missing out on food.

Raffle Tickets

Please sell as many tickets as possible—the monies raised help fund the Christmas Party

Please ensure you return the Raffle tickets by 30th November

If you are attending the Christmas Party then you can bring them on the day

Update on the Edimer EDI 200 Trial

The team in Cardiff is becoming excited as the approval process to start this trial is moving along. We already have regulatory approval from the MHRA in London and we are being supported in the local research ethics approval process by the Research & Development Office of the local University Health Board. We will submit the final application by the end of October and expect (hope) that authorization will come through in December. We will then be able to start treatments for newborn affected males in Cardiff.

The first treatment to be given to an infant has already happened in Germany. This went smoothly - without any problems - but of course it will take a long time to see what benefits the child might have. We hope that there might be a number of benefits but we are not expecting very much as the ideal time to start treatment will probably turn out to be well before birth. We are hoping that infants treated in the newborn period may have more and/or better teeth in their milk or adult dentition but we don't know how this will work out. However, as long as the trial shows that the treatment is safe, we are planning to set up a follow-on study giving the treatment in pregnancy. The protein used in the treatment is designed so that it can be pumped across the placenta into the baby and this mechanism works well in the treatment of affected mice and dogs before they are born. We think that starting in pregnancy will enable the treatment to make a really major difference to the affected boys.

If anyone is interested in taking part in this study - any pregnant woman who is (or might be) a carrier of X-linked hypohidrotic ectodermal dysplasia - then she would be welcome to make contact with Diana Perry or me to discuss this. In practice, it will be really important for anyone who wants to do this to make arrangements with us well in advance of the baby being born. It involves moving to Cardiff for three weeks and for the baby to be monitored closely while the treatments are given. We will do all we can to support people in this - and Edimer will meet their travel and accommodation costs - but it will obviously be difficult and disruptive and not at all like the usual first three weeks with a baby. In fact, it is a big 'Ask'. I very much want anyone who is thinking about it to be fully aware of the drawbacks, so that the decision is made in a thoroughly realistic way.

I can be contacted by e-mail clarkeaj@cardiff.ac.uk or phone (029 2074 4865) and would be very happy to talk with anyone who is or might be interested. The best way to find out more, however, might be to attend the EDS Christmas Party on Saturday 7th December as I will be giving a talk about the treatment and the trial during that afternoon. So I hope to see you there!

Angus Clarke, Institute of Medical Genetics, Cardiff University, Wales

Edimer Initiates Phase 2 Trial of EDI200 in XLHED-Affected Male Newborns

-- First neonate subject with X-linked hypohidrotic ectodermal dysplasia completes dosing with the novel ectodysplasin replacement protein EDI200 --

Cambridge, Mass. – October 08, 2013 – Edimer Pharmaceuticals, a biotechnology company focused on developing an innovative therapy for the rare genetic disorder X-linked Hypohidrotic Ectodermal Dysplasia (XLHED), today announced the enrollment and completed dosing of the first XLHED-affected neonate in a Phase 2 trial of EDI200, the company's novel, proprietary, recombinant protein. XLHED is an ultra-rare orphan disease of ectoderm development associated with a lack of sweat glands, poor temperature control, respiratory problems, and hair and tooth malformations. Affected individuals are at risk for serious and potentially life-threatening hyperthermia and respiratory infections. EDI200 replaces EDA-A1, the protein missing in XLHED and a key regulator of skin and tooth development. If fully developed and approved, EDI200 will be the first protein therapeutic to provide a sustained correction of the symptoms of this disorder.

"The completed dosing of the first patient in the neonate study of EDI200 represents a significant milestone for Edimer and those affected with XLHED," said Neil Kirby, Ph.D., President and CEO of Edimer. "Today is the culmination of several years of dedicated and impassioned work by the Edimer team and our external collaborators. We celebrate the courage of conviction that supported the science at the foundation of EDI200's development and the selfless participation of those involved in the clinical trials who share our goal to create a clinically-significant, life-long health benefit for those affected with XLHED."
"The first research project the NFED funded was in 1989 for the gene identification of X-Linked Hypohidrotic Ectodermal Dysplasia, XLHED," said Anil Vora, President of the Board of Directors at the National Foundation for Ectodermal Dysplasias (NFED). "On that day, we started an amazing journey that lead to the development of EDI200. Along the way, NFED families stepped up to volunteer for every research project in the battle to find a cure. We are thrilled to have witnessed and supported the ongoing development of EDI200 and look forward to learning the outcomes of this clinical study."

About the Phase 2 Clinical Trial

The Phase 2 clinical trial is designed to evaluate the safety, pharmacokinetics, pharmacodynamics and efficacy of EDI200 in XLHED-affected male newborns in the first two weeks of life. EDI200 dosing will be initiated between the 2nd and 14th days of life, with each study subject receiving two doses per week for a total of five doses. For additional information on this clinical trial, please visit clinicaltrials.gov, identifier NCT01775462.

Phase 1 Clinical Trial Outcomes

The EDI200 Phase 1 trial was an open-label, multicenter study to evaluate the safety and pharmacokinetics of EDI200. XLHED-affected males and females were enrolled in anticipation of future studies dosing XLHED-affected neonates. Six adult subjects, four males and two females, were enrolled at two U.S. sites and all successfully completed the five dose course of EDI200 over two weeks. EDI200 was generally well tolerated at the doses to be studied in neonatal subjects. No SAE's were reported and the majority of AEs in this open-label study were mild and transient with full resolution. An independent Data Safety Monitoring Board reviewed all adult safety data and approved the dosing and monitoring protocol for the Phase 2 neonate

study. For a developmental disorder such as XLHED, there was no expectation of clinical benefit following EDI200 administration to adult subjects. However, several subjects administered the higher dose of EDI200 demonstrated a short-term improvement in hair growth, dry eye symptoms and lung inflammation. Larger studies and a longer follow-up period will be required to determine if these changes represent an unanticipated and possibly sustainable benefit related to EDI200 treatment.

About EDI200

EDI200 is an ectodysplasin-A1 (EDA-A1) replacement protein, representing the first of a new class of molecules rationally designed to correct a specific developmental disorder. EDI200 has been shown to bind specifically to the EDA-A1 receptor (EDAR), activating the signaling pathways that lead to normal ectoderm development. EDI200 has demonstrated substantial and durable efficacy in animal models of XLHED with notable reduction in mortality and morbidity. The U.S. Food and Drug Administration (FDA) granted Orphan Drug designation and Fast Track status to EDI200. EDI200 also has Orphan Drug designation in Europe.

About XLHED

XLHED (also known as Christ-Siemens-Touraine Syndrome) is a rare disorder of development resulting from genetic mutations in the ectodysplasin gene (EDA). Patients affected by XLHED are at risk for life-threatening hyperthermia based on their inability to regulate body temperature, and for clinically-significant pneumonias resulting from their abnormality in respiratory secretions. Cardinal signs and symptoms in XLHED include diminished/absent sweat, reduced and abnormal airway secretions, few and often misshapen teeth, and absent or early hair loss from face and scalp.

XLHED patients surviving infancy are predisposed to atopy presenting with eczema and asthma, chronic sinusitis, recurrent nose bleeds, and dry eye complications. Almost uniformly they require dental interventions including early prostheses and later implants. Their susceptibility to hyperthermia, may impact normal participation in outdoor activities, sports and school attendance. Both medical and self-esteem issues are life-long in this disorder. As is generally true with X-linked inheritance, males are fully affected while females are variably affected.

About Edimer Pharmaceuticals

Edimer is a privately held biotechnology company based in Cambridge, Massachusetts dedicated to delivering a significant and durable improvement in the health and quality of life for future generations affected by XLHED. Edimer was established in 2009 with investment from Third Rock Ventures and VI Partners. NEA and Sanofi-Genzyme BioVentures joined the initial investors in a Series B round of equity financing that closed in July of 2013.

For further information on Edimer Pharmaceuticals, please visit www.edimerpharma.com. To receive regular updates about Edimer Pharmaceuticals' progress please join the XLHED network at www.xlhednetwork.com.

CONTACTS:

Edimer Pharmaceuticals	**National Foundation for Ectodermal Dysplasias**	Maureen L. Suda (Media)Neil
Kirby, Ph.D, President & CEO	Jodi Edgar Reinhardt	Suda Communications LLC
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Money Spent in the Last Quarter

From the Support Fund

1 Child's Wig

1 Air-conditioning Unit

2 School Support

Race Day 9 – Better Late than Never

By Dave WIllats

For the last eight years I have organised an Annual "Race Day" in aid of The ED Society. It's a typical fund raising evening where we play a DVD of horse racing, bets are placed on all the runners with the winners sharing a tote pot and a small percentage being donated to this wonderful charity of ours. It's very much a DIY event as we use our own TV and DVD player with the actual Race DVD and tote tickets being purchased on ebay.

Since we held the first day back in 2005 in my own back garden the event has been a regular fixture on the calendar of my friends and family. It is an eagerly anticipated event that we normally held on either the first or second weekend of July. However, this year was different, my wife Tracey and I have had incredibly busy years with both of us starting new jobs, so the event had to take a back seat. But friends and family refused to let it go and kept up the pressure and eventually we was able to find a free date in the diary and more importantly a hall that could hold it.

A hall was booked for 12th October, invitations were sent, a DVD of new races purchased and the planning began. We normally get between 60 and 70 people to our race day but this year our expectations were exceeded as over 100 people expressed an interest in supporting us.

We planned nine races, each with eight runners and I offered the ownership rights for sale at a bargain £3. All 72 horses sold very quickly bringing in £216. I requested sponsorships for every race for a minimum of £10 a time. I received enough offers of sponsorship for 3 people to sponsor each race and the total of sponsorship bought in an incredible £500. A sponsor doesn't get much other than their name in the program and the satisfaction that their hard earned money is going to a worthwhile cause. We even received sponsorship from Texas in the U.S.
The night itself was fabulous with everyone thoroughly enjoying themselves. It's not quite being at Ascot but it's a great opportunity for my friends and family to all get together and have some fun. I am an eight time veteran of hosting these nights but I handed the reins over to my youngest son Max to learn the ropes. He had a bit of a croaky voice but did a sterling job presenting all the races, controlling the tote and ensuring that everyone knew what was going on.

A raffle, run by my mother in law, Rene, bought in another £200 and we had enough prizes to keep us going for quite some time. It was a shame we ran out of tickets as people wanted more and we probably could have sold another £100 worth. Note to self for next time: buy more books of raffle tickets!

We counted up the total on the night and had raised a brilliant £1,285. With pledges of over £100 still to come in we reckon the final total will be about £1,400 which is a sum I could only dream of before the night but makes all the organisational effort well worth it. The feedback I got after was great, with regular guests as well as those coming for the first time all wanting to come back again. Our July event was delayed three months to October but as the saying goes, it's better late than never.

Honeymoon Around the World!

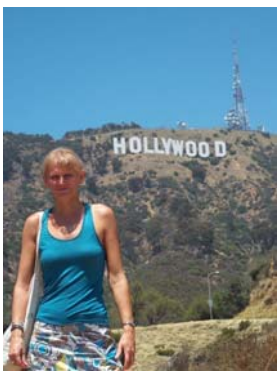
My last article for the ED newsletter was mainly about my own denture experience which spans over 40 years, from the age of two. I'm hoping that I gave some hope and confidence to all those that suffer from ED and who may find themselves in a similar situation. I also mentioned that my last dentures were replaced partly due to general wear and tear, and also in preparation of my wedding day, and my honeymoon which I would like to now share with you.

Our aim/dream was to make our honeymoon last for 2 years, and travel around the world visiting family and friends on the way. I also had it in mind to hit as many summers as possible, this was for 2 reasons - (1) Once my body has acclimatized, I find warm temperature more comfortable – and even better, if I am near a beach, shade and have plenty of water. And (2) Thin small clothes meant I could fit more into my backpack, as opposed to just a few warm thick ones! I'm sure women reading will understand where I'm coming from.

I've always had the travel bug, and found that living the outdoor life suits me very well, my body stays at roughly the same temperature, as does my mood . Living in cooler climates mean that wrapping up warm to go out and then unwrapping to stay in has always caused my temperature to rise and fall so rapidly that my body and brain just can't keep up with it, which can lead to me not only being very uncomfortable, but quite irrational and sometimes act out of character. This has been very frustrating at times as people just can't seem to understand this ED trait.

So, when deciding our locations to visit and where friends and relatives lived around the world that we also wanted to see on route, the weather played a bigger part than I realised. Having lived in the south of Spain for 7 years, we decided that we would start at the North, beautiful Bilbao. The Basque country is truly unique for its food, coast line and various historic attractions. The temperature back in March 2012 when we arrived was only a few degrees warmer than the UK, so it was a good place to start our honeymoon and allow my body to slowly start adjusting. We travelled through Portugal and down to Malaga in the South. This was also what I called our honeymoon Part 1, when travelling for a long time there are so many little things that are forgotten about but that are so important. So after spending 9 weeks away it was time to briefly head back to the UK and repack our backpacks and make sure that we had as much as we needed to continue our travels, including our Factor 50 sun cream which was essential and not always easy to buy in the UK.

Our next location was Toronto, another beautiful city, although I wasn't quite prepared for their heatwave. One thing that was always included in our itinerary was a beach. I know you're probably thinking that I just want to lay and cook in the sun, but this is not the case. If I am within walking distance of the sea I know for a fact that I can keep cool, keep my circulation going and not suffer any side effects from the heat. So when we headed to Lake Ontario it was a vision that I was dreaming of. Toronto is actually nowhere near the coast, however it does have a lake so big that it actually looks like the sea. Our next stop was via Los Angeles, Anchorage in Alaska. As well as having relatives there my husband is also a keen fisherman, so fishing for the famous Alaskan Salmon was a must, eating included which was delicious!. We hired a car and camped all around the South Peninsular of Alaska, making the most of the 23hours of day light which gave us extra time to take in the beautiful sights. It was then back to LA for a few days where we walked down Hollywood boulevard, had our photos taken by the Hollywood sign and saw the tourist attractions. This was also a break to prepare us for our 14 hour flight to Brisbane in Australia, we spent the next 3 months travelling up and down the East Coast. I had ideas that while my husband was fishing for our breakfast, lunch and dinner, I would relax in a comfy chair in the shade reading a book..however, I soon became



bored of this, and decided to start fishing myself. We caught fish we'd never seen, or eaten before and spent many hours in amazingly breath taking locations around the coast, as well as rivers inland that we could get to.

So after 3 fantastic months and just before our tourist visa expired, we then flew to Auckland, New Zealand and spent the next 3 months travelling slowly south to Queenstown ,just in time to spend Christmas and New Year with family. Although it was summer, New Zealand has very little ozone, which meant you couldn't really spend too much time in the sun,15 minutes maximum, but all cafes, restaurants and site seeing locations provide excellent shade wherever you go, so a real plus for me.

Our next long flight after the new year was Singapore, very hot and humid, but by now my body had adjusted well to temperature change, thanks to a little air con now and again. After seeing the sights in Singapore we travelled in a VIP coach, apparently the way to travel north according to the hotel manager, and how right he was. After a very luxurious 5 hour coach journey (costing around £17 each, including lunch) we arrived in Kuala Lumpur, where there were many sites to see, including the famous Petronas Towers. Next stop was Langkawi, a small Malaysian Tax free island, we decided we liked it after a couple of days and extended our stay by 3 weeks, just to make sure we could visit all parts of the island, as well as surrounding islands. We were planning to go by boat and road to Thailand, but found that visa requirements had changed since our last visit and we would only get a 15 day visa if we travelled by land, unless we went to a Thai embassy and applied for a 3 month visa. This would give us the time we needed to travel to the places that we wanted to go. So, for the first time we had to go back on ourselves and travel south to Penang, another Malaysian island. After getting our visa, we bumped into a English backpacker who we'd met in Australia 5 months before. He recommended that we visit the west part of the Island, Teluk Behang, a small fishing village off the tourist trail. As we didn't have any definite plans we decided to see it for ourselves and caught the bus a few days later. This was the most surreal part of our trip, the village was very small and after trying to find a place to have breakfast one morning, we heard about the Prime Minster of Tourism & Finance and his wife were visiting the area to promote tourism to this little village. We had no idea that we would become part of this tour, when the Prime Minister saw us (while being taken around in a rickshaw) he and his wife insisted that we joined them, and being the only westerners around we were good publicity for them. So all of a sudden we turned into celebrities for the next couple of hours, both wanted to know what we were doing and how we found Malaysia and were extremely kind to us. Although we missed breakfast, it was without a doubt an experience that we couldn't have planned and will remember for ever. Just like when one afternoon, the lady in her restaurant turned on the tap to her tin roof so that we could cool down…maybe because I had my hands in small buckets of ice at the time, but it certainly cooled me down. We later continued our travels to Krabi, Phuket, Sura tani where we got a train to Bangkok then bus north to Korat.

So after a quite a few weeks, we then travelled back to Bangkok our last Thai destination before flying to Malaga, via Moscow, which was supposed to be for just a few hours, but turned into over 24 hours as we were delayed due to snow blizzards and under hotel arrest as we didn't have visa's, and then back to Malaga were we spent another month before heading north by bus to Bilbao and then back to sunny Gatwick when the UK summer started some 14 months later, having travelled roughly 45,000 miles, visited 10 very different countries and more locations than I can remember, but after having had a wonderful and memorable honeymoon. The website about our travels that I tried to keep up to date was www.johndenisechurch.co.uk

I will never forget my Doctor telling me at the age of 14 that I should keep out of the sun and never go to hot places…I wish he could see me know!

Fundraising Our Grateful Thanks

The fundraising events which have been organised and the many donations that have been coming in over the past 3 months amount to £3525.

Many thanks to the following families who sent in donations; Cunningham, Harding, Dewsbury, Creron -Jones, Pennington, Shortman, Beeson, Burden, Woodburn, Willats and Andrews.

Huge thanks to Kelly Fitton for holding a hugely successful charity dinner - the total raised so far is £1526.

Huge thanks to Mark Macnair (Trustee) for selling his own produce on his stall throughout the Summer and raised another £400 for the Charity.

Many thanks to Ryan Watkins for doing a sponsored 10k run in Cheltenham and raising £70 - well done Ryan.

Grateful thanks to Dave Willats and family who recently held their Annual Race Day and raised around another whopping £1400. Thanks to you all on this your 8th year of this successful fundraiser!

Keeping Warm in Winter

Individuals who have ED and lack of temperature control caused by malfunctioning sweat glands suffer in the winter months as well as the summer months.

Do you suffer from the cold in the winter months?

What methods do you put in place to prevent getting too cold?

How does the cold affect you?

How do you cope going from the cold outside elements to hot inside environments?

How does your child cope in school i.e. going from a warm classroom to the cold playground?

I will be writing an article for the January newsletter and would appreciate any tips you have for controlling your temperature.

Please email me diana@ectodermaldysplasia.org