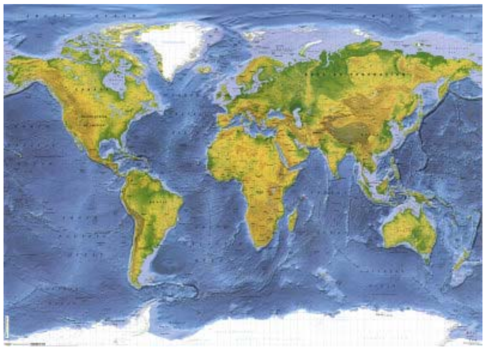
Internationl Ectodermal Dysplasia Newsletter



Summer 2013

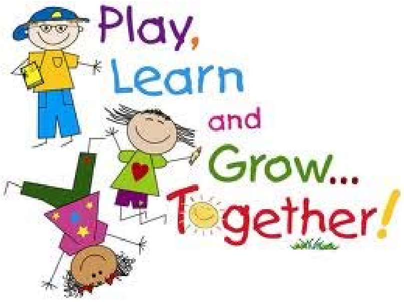
International ED Network
Unit 1 Maida Vale Business Centre, Leckhampton, Cheltenham, Glos. GL53 7ER England.
Tel: +44 (0) 1242 261332 Mobile: + 44 (0) 7774 465712
www.ectodermaldysplasia.org - Email: diana@ectodermaldysplasia.org

Here is our second IEDN newsletter!

It has not been easy collecting all the information for the newsletter, but with joined forces Diana and I made it! In addition to the result of our survey, there are several important and interesting articles you can read.

Please do not forget to save the date for our next meeting in Milan, Italy. We hope that as many as possible from the large ED family will be able to participate. Planning has now started in Italy and we will communicate more details during the course of the year. Any suggestions and proposals are most welcome.

Greetings from Vienna
Ulrike



International Group Leaders Meeting
29th March, 2014 Milan, Italy

Save the Date

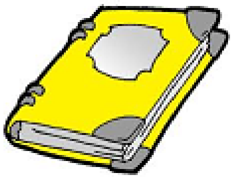
Giulia Fedele from ANDE and Salvatore Randazzo from XLPDR are busy organising the Group Leaders' meeting to be held in Milan next year.

More information will be available later this year.

Any suggestions regarding the agenda will be appreciated.

XLPDR International Association ONLUS

A.N.D.E.



International directory

The recent amended sheet has been distributed. Please inform Ulrike about every change of address or contact details. We'll discuss how to proceed in the future with the topic at our next meeting in Milan. Please ensure you use the correct email addresses.

The ED family is growing! We recently received the following contact details for new ED patient Associations or people who intend to found an organization.

Argentina: ADEA, Asociacion Displasia Ectodermica Argentina. President is Mr. Dario Corrao. Secretary Mrs. Valeria Vera. The Association have 145 members, 40 of them are affected. Mail: Valeria Vera [vera_valery@hotmail.com]

Hungaria: Mr. Gabor Kovacs intend to found a group and is looking for affected people in his country. Gábor Kovács [kgabor.biol@gmail.com]

Venezuela: Facebook group, Displasia Ectodermica Venezuela

National Foundation for Ectodermal Dysplasias Fall 2011 Update

Greetings Friends,

It was such a pleasure to see everyone in Erlangen, Germany last summer. It provided the opportunity to get to know each of you better and to share information about the Ectodermal Dysplasias with organizations around the globe. We look forward to meeting again in 2014 in Milan!

The National Foundation for Ectodermal Dysplasias is on track to have our most exciting year ever. There are new programs that will be rolled out, an outstanding National Family Conference, and a lot of fun with special events and promotions.

We began 2013 in our new offices, and we are so excited! We are now closer to St. Louis, Missouri, and the resources it can provide while being equidistant from two of our partner dental schools (that are also Dental Treatment Centres). It places us closer to several major universities that can provide student interns who can help us achieve our aggressive goals.

The NFED is gearing up for an absolutely fantastic National Family Conference. Our committee has created a conference that will have something for every age. ***Get Roped In*** will be held July 23 through 26 in Houston, Texas. Two days prior to the Conference, we are holding a research conference on Goltz Syndrome. What an exciting opportunity we will have with this rare syndrome – 15 families will gather to meet with researchers and clinicians to begin seeking treatments and ultimately a cure for Goltz Syndrome.

Following are some of the highlights the NFED experienced this past year:

Support
We began the process of empowering our Family Support Council (FSC) to create a robust volunteer program that will attract families and friends who will engage our family members, care providers, and friends in the work of the NFED in order to identify and connect more individuals affected by Ectodermal Dysplasia.

- The National Family Conference in Orlando, Florida, drew 300 attendees; 87 families; 6 syndromes plus unknowns; people from 30 states and 6 countries; and 38 volunteers.
- 314 individuals contacted the NFED for the first time in 2012.
- 6 Webinars were offered and recorded for later use. More than 200 people accessed this service through our postings on YouTube.
- 10 students were awarded academic scholarships to assist with their college education. This totals 259 students over the Scholarship Program’s history.
- 20 Family Liaisons actively shared and connected with families in 8 Regions around the U.S.

Treatment

The NFED received a record number of applications for treatment this year, showing the need for continued financial assistance to our families for complex dental care, dentures, wigs, cooling vests, and air conditioners. In addition:

- The Treatment Assistance Program funded 4 complex dental treatments; 7 dentures; 5 cooling vests; and 1 wig.
- Five new Dental Treatments centres were added.

Professional Education

The NFED continued to educate and inform care providers and future care providers in the area of Ectodermal Dysplasias treatment.

- A Dental Symposium was held in Houston with 40 attendees.
- Educational workshops were provided to students at two local dental schools with approximately 25 students attending at each site.

Research

As Edimer Pharmaceuticals conducted adult clinical trials with their EDI200 treatment, the NFED continues to provide access to patients willing to participate in necessary research that allows researchers the ability to find cutting edge treatments. The NFED provided Edimer with a seed grant a number of years ago that allowed them to make this breakthrough discovery.

- NFED supported all Edimer Pharmaceuticals' projects by organizing and implementing research at our National Family Conference, marketed Edimer's efforts and recruited individuals for each of their projects.
- Funded 3 research projects.
- Held an Ectodermal Dysplasias Classification Conference: A Model for Integrations of Clinical Systems Biology and Bioinformatics October 18-20, 2012. Thirty experts from across the world participated in this landmark conference. *The American Journal of Medical Genetics* will publish all proceedings in their journal.

Warmest Regards,
Judy Woodruff, Executive Director

The ED Society are busier than ever with new families joining us from around the world every week!

We have been busy making contact with some of the overseas families and encouraging them to become a contact for ED families in their country.

Our Facebook page is very busy with many parents chatting to each other and we are now having new families join us from around the world, which is very exciting.

Our Christmas party in December was a huge success attended by many familiar faces but also many new families.

We are currently working on creating a network of specialist doctors around the UK who are willing to help us. This may be a topic to discuss at our next meeting for those countries who do not have centres of excellence, but need to know where doctors who have experience of ED are situated. A network of this kind would enable the groups to recommend doctors who work close to their families' homes?

Results of the Questionnaire

1 Cooling systems

ITALY
Companies providing cooling systems: Lydda Wear, http://www.lyddawear.com
Tecniche Italia : http://www.tecniche.it

GERMANY
The products has been presented at the International Conference in Erlangen
Pervormance International GmbH www.e-cooline.de

USA
See sheet attached

SPAIN
We only have cooling jackets in black, blue and grey. Contact: "jeronovi@gmail.com"

CANADA
The vest we test marketed, and loved, was the Arctic Heat cooling vest which can be custom-made to fit any size adult or child based on chest measurements. Not only is it lightweight (just over 2 lbs, making it great for even young children), it is very easy to use and maintain.

The vest contains crystals located in panels that you activate by soaking the vest in water for 10 minutes. You then put it in the fridge or freezer - on very hot days, we have used this vest when it is frozen solid but it takes a deep breath to put it on! On milder days, wear it cold from the fridge. It is easy to wash!

To order online, visit www.arcticheatusa.com

Our organization supporters get a discount. We have not yet found a similar product made in Canada.

Chillow Pillow: http://www.soothsoft.ca/products/1-chillow

Cooling bandanas and neckties: We purchase these through Lee Valley tools, a Canadian based company. The bandanas and neckties work like a cooling vest with gel beads to help you stay cool.
www.leevalley.com

UK
As we are a notoriously cool country, there are only a couple of companies who provide jackets/vests for children.

http://disabledgear.com and www.jacksontechnical.co.uk

2 Pre-implantation Genetic Diagnosis (PGD) testing

USA
In the US PGD testing is possible for boys and girls.

Costs depend on the area of the country. PGD testing is expensive - costs are usually between $2200 and $5000 - in addition to all usual IVF and medication fees.

Average pre-implantation genetic testing costs in the US

- IVF costs - in the US the average cost for IVF with ICSI is about $12,000 to $13,000
- The average cost for in vitro fertilization medications is $3000 to $4000
- Embryo biopsy charges are about $1500 to $2200
- Aneuploidy testing (for chromosome normality) with PGD is $2200 to $5000
- For single gene defects (such as cystic fibrosis), the cost is about the same as for aneuploidy screening, $2200 to $5000
- IVF with pre-implantation genetic testing (with meds) usually costs $17,000 - $22,000

This is dependent on the cost of the in vitro fertilization and also on the amount of medications needed for IVF ovarian stimulation.

Some couples have insurance coverage for in vitro fertilization, but it is unlikely that insurance would cover chromosomal or genetic testing.

UK
There are about 8 clinics where PGD can be carried out. Not all genetic conditions are able to be tested. The sex selection procedure is only allowed to avoid having a child with a serious medical condition

The National Health Service (NHS) will provide funding for PGD testing, but it has to be approved via the local Primary Care Trust (PCT). If funding is not granted then families may pay at a cost of around £6,000 - £9,000 per treatment cycle.

Only two conditions are available to be tested at the moment, Ectodermal dysplasia (Hypohidrotic) and Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome (EEC)

GERMANY
PGD is only allowed to avoid having a child with a serious medical condition. Parents have to undergo before medical advice.

AUSTRIA
PGD is forbidden

ITALY
Genetic testing is for both men and women, at own cost. It depends from region to region, however the Tribunal established PGD available for diseases with high risk of mortality, not for Ectodermal dysplasia. Moreover PGD is forbidden by law number 40, overall in Italy.

SPAIN
In Spain there are family planning clinics of ectodermal dysplasia analysis.
This clinic is situated in Gerona but the price depends on the type of analysis of ED.

Laboratori de Reproducci Assistida, CENTRE DE GENETICA GIRONA, T. 972 217407
info@centredegeneticagirona.com

MEXICO
Unfortunately in México we don´t have genetics testing for ED or tests related with this. Most of Insurance Companies don´t cover genetic diseases.

CANADA
Yes, but until recently was only available to some Canadians at the McGill Reproductive Centre (MRC). It is now available to all Canadians through a transport PGD program. An incubator has now been engineered for use with the new Transport PGD program. By using this service, patients are able to have their eggs recovered and biopsied at IVF clinics across the country, with genetic material for diagnosis sent immediately to MRC in Montreal. By using this service, patients can have PGD without having to travel to another city.

It depends on the disease being diagnosed and which IVF clinic is doing the procedure and the province.

These are government regulated clinics but not publically funded – in other words, patients pay except in some circumstances (each province has its own eligibility requirements around IVF treatments of any kind) The McGill Reproductive Centre lists the following fees. Keeping in mind, depending on what IVF clinic is doing the procedure, there will be additional fees to these.

Each IVF clinic will have its now PGD fees plus what the McGill clinic charges.

McGill Reproductive Centre Rates 2013

PGD Consultation Fee (approximately one hour)	$250.00	$300.00
Sex-selection for X-linked diseases (X, Y, 15)	$2,000.00	$2,250.00
Aneuploidy Screening / PGS (8 chromosomes X, Y, 13, 15, 16, 18, 21, 22)	$2,450.00	$2,750.00
Maternal or paternal reciprocal translocation (without normal/balanced distinction)	$4,000.00	$4,500.00
Robertsonian translocation (without normal/balanced distinction)	$3,000.00	$3,400.00
Robertsonian translocation (without normal/balanced distinction) + PGS	$4,000.00	$4,500.00
Single gene defects / ICSI included (CF delta 508, SMA)	$3,450.00	$3,900.00
Other single gene defects (upon request)	$4,000.00	$4,500.00
Y Chromosome Deletion	$495.00	$590.00
Embryo biopsy for PGD (required for all patients undergoing PGD)	$500.00	$600.00

4 Student exchanges

As in Erlangen agreed we like to promote and support student exchanges like the one of Markus Kappen. The story of his stay in Singapore is attached.

Everybody seemed to be very excited about it; the problem is how to implement the idea into reality. **Italy** already placed an announcement on their website for student exchange, www.assoande.it , mail to: segreteria@assoande.it and is waiting for participants.

Mexico will have the 12th, 13th and 14th of July their 6th Family Reunion. Karla will collect the names and addressed of the people interested in participating.

Germany will have the annual family conference on 3rd to 5th May 2013. We will present the project to our members and publish addresses on the IEDN website.

UK do not have student exchanges, but are happy to support your country via our newsletter. The IEDN website should be the appropriate place to offer and to search for addresses.

5 International Registry

ITALY

Dr. Gianluca Tadini takes care of the International Registry, for the National one every hospital recognized as Centre for Diagnosis and Therapy of ED should provide o insert patients data. It is not accessible for patients users but only for the ISS (Istituto Superiore di Sanità, Superior Institute of Health)

GERMANY
All members has been informed many times by newsletter or during the family conference about the importance to register.

MEXICO
Karla wish Mary to send her the information to have access to the international registry, so she can be able to make the comments about it.

UK
Our members have been informed by letter and via the newsletter many times, many have registered with the XLHED network.

6 National Health systems

SPAIN
The National Health System has the same treatment region to region, but some regions are paying implants and prosthetics and others not. The health system treatment usually is same with the age. c) There are no pharmaceutical protocols.

ITALY

a) Unfortunately we do have discrepancies outcomes from region to region, sometimes even in the same region. The explanation: ASL (Azienda Sanitaria Locale, Presidium of Local Health) “interprete” in their way the National Law (104) about the right to have or not the “invalidity” and then the therapeutic protocol.

b) Passing from adolescence to 18 years old, the percentage of “invalidity (0 to 100%), mostly changes, lots of rights are lost, therapeutical protocol remains equal.

c) Unfortunately in Italy there is no unique centre for children and adults, mostly the paediatricians keep on following the patients all lifelong.

UK
All health costs are covered under our National Health System, although prescription drugs have to be paid for those between 19 years and 65 years of age. The cost of a prescription is £7.85 per item. A prepayment certificate can be bought: a 3 monthly prescription is £29.10 and a 12 month certificate is £104.00 – making a huge saving if you have to have many medicines each month. Low income individuals can apply for assistance with prescription charges. All operations and emergency treatment are covered under the NHS.

CANADA
No, emphatically not. While we have a national health care system, it is managed by provincial territorial governments. We have 10 provinces and three territories, and all handle what is covered or not, differently. So doctor’s appointments, genetics, and anything medically related is generally covered across the country but with huge waiting lists (months to years for genetics).

Most drugs are only partially covered or not at all so you have to rely on our own drug plan or pay out of pocket. The dental component is also an issue. Two provinces, Ontario and Quebec, have some coverage for children with ectodermal dysplasia to help with dental costs, we have heard that Saskatchewan may be implementing one too, but all the other provinces, you are on your own for all dental-related surgeries, implants, repairs, bridges, etc.

Provincial governments have all instituted programs to cover the costs of drugs. BC, Manitoba, Québec and Saskatchewan have universal coverage. The other provinces provide drugs to senior citizens and those on social assistance. About 44% of Canadians are covered under these programs with a similar number belonging to private insurance plans. Provincial governments attempt to limit the cost of their programs by permitting pharmacists to product select; that is, to dispense a lower price generic drug in place of a more costly brand name one.

Pharmacists: In Canada, education and licensure for health professionals, including pharmacists and pharmacy technicians, falls under the responsibility of the provinces and territories. The Canadian Council for Accreditation of Pharmacy Programs (CCAPP) is responsible accreditation and continuous program improvement of professional degree programs in Canadian universities, as well as pharmacy technician programs in the college-level system.

In 2000, all provinces, except Quebec, signed the National Association of Pharmacy Regulatory Authorities (NAPRA) Mutual Recognition Agreement requiring Pharmacy Examining Board of Canada registration as a prerequisite for licensure of in- and out-of-province graduates and out-of-country applicants. You can't practice pharmacy in Canada without a license.

Sale of Pharmaceuticals: The sale of drugs in Canada must be approved by Heath Canada, through the Therapeutic Products Directorate (TPD) a national government agency that reviews safety, efficacy and quality both non-prescription and prescription drugs, and disinfectants and sanitizers. Companies or persons researching and developing new drugs must do preclinical tests to indicate a substance produces the desired result and is not toxic and must apply to the TDP for authorization to conduct a clinical trial. If the trial proves it has potential therapeutic value that outweighs risk, the sponsor can file a New Drug Submission with the TPD.

The review process is lengthy with multiple control points before a drug is approved for market.

All drugs must meet the requirements of the Food and Drugs Act and Regulations. All drugs on the market are monitored with strict regulatory controls. Additional guidance on pharmaceutical quality is provided in Health Canada guidance documents and the ICH guidelines.

The Food and Drug Act is currently being updated and modernized to take into account International Alignment and Best Practices, emerging public health threats, new and emerging science, medicines and technologies and globalization, amongst others.

Canada follows the ICH Guidelines on pharmaceutical quality.

MEXICO

a) No. A very short group of doctors know about ED Syndromes, therefore they don´t have the tools to treat patients. In many cases they come to the association to receive information or to contact them with doctor with experience in that subject.

b) Karla is convinced that in childhood is very important to invest in all kind of care, that's the way to strength the weak points of ectodermal dysplasia and have a better quality of life in youth and adulthood.

c) No pharmaceutical protocols.

DIA Meeting Amsterdam
4-6 March 2013

Thanks to EURORDIS, the Asociación de Afectados por Displasia Ectodermica (A.A.D.E.) from Spain, attended the DIA 2013 49th Annual Meeting, this meeting around Advancing Therapeutic Innovation and Regulatory Sciences is the largest multidisciplinary event that brings together a global network of professionals to foster innovation that will lead to the development of safe and effective medical products and therapies to patients.

DIAs comprehensive education and training opportunities are designed by and for a global audience of regulatory, academic, industry, and patient advocate professionals and experts working throughout the medical product industry.

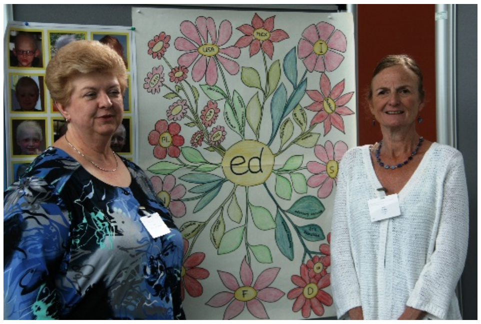
A.A.D.E showed a poster about ectodermal dysplasia, we saw this as a very good way to promote and raise awareness of ectodermal dysplasia within this community of professionals.

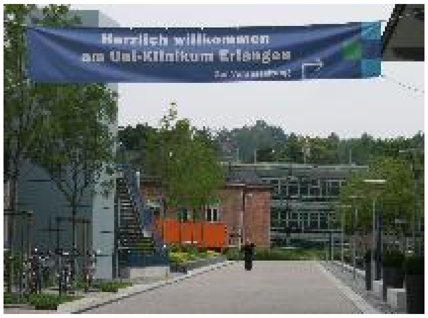
We have several goals and one of these is to engage researchers and pharmaceutical companies to research the different types of Ectodermal Dysplasias.

Gema Chicano, President A.A.D.E."

Remember Erlangen 2012

Some nice souvenirs of the International Conference June 2012 in Erlangen!





“Asia meets Germany” at 35 degrees in Singapore

I have always been curious, how living in a constant hot and humid climate might feel for people without sweat glands. Knowing that ED (Ectodermal Dysplasia) exists in hotter continent I wondered how their daily life would be. I am a German guy who has a Hypochondriac Ectodermal Dysplasia condition. Hence, I can only judge for European standards. In Berlin, only for a month or two within a year, the temperature soars above 30 degrees Celsius. Not even during my travels through southern Europe the high temperatures were comparable to those in Australia, parts of Asia or southern America. Fortunately, I was given a chance to participate in study exchange program for a semester, and I want to take this opportunity to experience first hand what I have been so curious about for so long.

Luckily, my university maintains a good partnership with education institutions in Taiwan. My decision was made and my target was Southeast Asia. Honestly, I was a little fearful of the climate. To seek answers for all my questions and doubts, I contacted Janice Chuah who is listed online as the Asian contact for Ectodermal Dysplasia. Janice is a teacher and lives with her husband and her three sons in Singapore. The 11-year old Yihhang, her oldest son, is also affected by ED. Patiently; Janice clarified all my questions via email in details. With her help, I was looking forward to a bright trip to Taiwan. After I booked my flights Janice immediately invited me to Singapore.

35 degrees, high humidity and a brightly shining sun. Caused by the close proximity to the equator, Singapore doesn’t have any season and constantly experience high temperatures. Fortunately, Janice recommended a nice hostel, and even organized a gondola ride plus reserved a table in a nice restaurant. Hence, I was fully treated to good entertainment and delicious foods.

Apparently, it is hard to find people with ED condition in Asia. After Yihhang was born in 2001, Janice began to search for fellows/Asians having the same condition. She wasn’t successful though. One day, due to his extraordinary engagement in school, a newspaper devoted a whole article to Yihhang’s school career. When Wee Tien a 30-year old Singaporean read this article, he immediately contacted Janice asking for a meet-up. Subsequently, they developed a good friendship and see each other once in a while. Relieved, Janice recounted, “The moment we met Wee Tien, our lives changed. People are always stronger in teams.”

When I met up with Janice and her family, Wee Tien and his mother joined us for dinner as well. Together, we had a great time talking about anything and every thing. Of course we also talked about the topics related to ED and it turned out that Asian dental care insurances do not cover implant surgeries. The self-support group in Germany fought many years to get such support from the government. Unfortunately Yihhang and Wee Tien face a significantly different situation. The Singaporean state doesn’t make exceptions for them. Hence, both of them work a lot on their education to finance medical care themselves.

I learned a lot from this meeting. Not only I experienced life in the heat and survived easily, but also, learned how important it is to forge good relationships with those having the same conditions. Having the German support group was always natural for me. Now as I know Yihhang and Wee Tien, I value this support even more than before. People are always stronger when they band together as a team!

I thank all my Singaporean friends for a great time in Asia and fully encourage encounters like this to all the international people with Ectodermal Dysplasia. As a globalized world is getting closer together you should stop on one of your international trips for a coffee with a fellow ED. I bet you will have a great time sharing and exchanging stories!

Markus Kappen markus.kappen@googlemail.com

---

edimer
Bringing Hope to XLHED

Edimer would like to thank the XLHED community for their support over the past years, but especially in the last few months as we began our clinical trials!

We had an overwhelming response from volunteers for our adult clinical study. It's not a small request to be the first group to receive a novel drug, especially when we do not expect to see personal benefit in those volunteers. We received inquiries from all over the world and were able to quickly enroll the trial. We are pleased to say this trial is now complete and we are finalizing our data report now.

Our next step in this journey is to offer EDI200 to newborn baby boys who are affected by XLHED. We are currently working through regulatory process and ethics committee approvals at 6 international clinical sites to make this happen. The sites we are working with are located in:

- Washington, DC, US
- St. Louis, MO, US
- San Francisco, CA, US
- Cardiff, UK
- Erlangen, Germany
- Paris, France

While there are many factors involved our goal is to have the first site open by this summer.

If you, or a member of your family, are an XLHED affected/carrier woman who is pregnant, or considering pregnancy, and would like more specific information about this clinical trial in newborn XLHED affected boys please be in touch with Edimer.

- Tessa Field, Manager of Clinical Outreach
  tessa@edimerpharma.com
  +1-617-758-4308

- Ramsey Johnson, Sr Director of Clinical Operations
  ramsey@edimerpharma.com
  +1-617-758-4305

International Ectodermal Dysplasia Network

The IEDN website is now ready to be used, but we need to input all the data. If you have any articles, newsletters or other information you feel will be helpful to have on the site please send them to Diana.