Dear friends, chers amis,

I am very happy that after some initial difficulties our common project “the International ED newsletter “can now be launched.

It is important that we keep in mind everybody’s contribution will make the newsletter interesting and helpful.

Many thanks indeed to Diana who has taken over the job of collecting and distributing the information. As we discussed in Vienna in October 2010 I suggest we publish the next newsletter in March/April 2012. Could you please aim to send your next article to Diana in January 2012.

Greetings from Vienna,
Ulrike
I would like to share with you a project that means a lot to me, as I really think it helps to improve life for ED persons in France:

I give a lecture on ED in most of the dentistry schools in France every year. This year, one of the examinations in Paris was on my course and I had to put the notes to 80 students! This lecture is of course on ED syndromes:

I explain that they need to be vigilant when they see a girl with agenesis as it could be a girl with ED. I advise them of what actions they should take (where to ask for the molecular testing, what other symptoms they should search for…) I insist on the importance to diagnose the little girl or woman in order that the family know what is going on.

I explain to them the importance of the molecular testing as it provides information on who is or who could be a carrier; also that in order to decide or plan the therapeutics, because an agenesis isn’t like another agenesis, they have to know what gene is involve: ED bones are more fragile and osteointegration should be complete before putting teeth on the implant.

I also explain to them that most doctors say to wait for the kids to be ready for having dentures, but I don’t agree as kids are never ready to live with such a thing. They want teeth, but not this thing in their mouth. The more we wait, the less children are compliant, so we have to ensure that they have their dentures by 3 years old.

I tell them that the patient isn’t only the kid in the dentist chair but that it’s the whole family. So dentists need to explain to the parents on and on, they need to gain the parent’s trust, because if the parents aren’t sure about what is going on, the kid won’t wear his denture.

I tell them that I don’t understand why it’s so difficult to find dentists who are willing to take care of our kids; our kids should be their most rewarding challenge and that there is nothing better than to put a smile where there was none.

I explain to them what parents have go through, having to face the unknown and even worse the lack of consensus on medical issues. For example, there isn’t a consensus here in France about using composite or an over denture, if the denture should be put by night or no, where to take bone in order to practice a bone augmentation...

I tell them how I have managed to obtain an official decision about implants in youths. However, before the official decision, nobody wanted to put implants in children here in France, they were really against it, but since I obtained the official statement the government now pays for the treatment in kids from 6 to 18 years. In 6 months time, I should also get reimbursement for implants on adults.

Olivia
Selbsthilfegruppe Ektodermale Dysplasie e.V.

Here is the news of the German speaking Patient Association Selbsthilfegruppe Ektodermale Dysplasie e.V.

1) Our annual family conference was held on the 8th to 10th April at Löwenstein, Germany. 160 families attended the meeting, among them about 50 people, who had never attended a meeting before.

2) During the meeting several medical examinations took place. All of them especially for ED1 affected people.
   - Examination of the eyes with infrared photography
   - Examination of sweat glands with a confocal microscope (by Edimer)
   - Testing of the lungs and of breathing

   The minutes of the meeting will be printed and sent to all members and to others if interested.

3) We actually have 205 member families.

4) We would like to encourage the International ED families to use our International Board at www.ektodermale-dysplasie.de

ozED Australian Ectodermal Dysplasia Support Group Inc

ozED are currently in the process of finalizing details for our annual camp/conference (Campherence) to take place 7-9 of October in the Blue Mountains NSW.

We have been fundraising madly to accommodate activities such as archery, high ropes, BMX, swimming, abseiling and leap of faith. These activities come under the banner of adventure therapy and do wonders to build our children’s self esteem and have a lot of fun.

The campherence is so important to all our members as it enables our members from all over Australia to meet, reunite, provide valuable information and education and gives the children an opportunity to meet other children who look like them and have the same daily struggles.

We allocate a great deal of time of parents to mix and mingle and share hints and tips and we find the information coming out of the campherence is invaluable.

I am looking forward to sharing some photos with you after the camp – watch out for this in the next newsletter.

Jenny Boss, President
Ireland Group

An Irish branch of the Ectodermal Dysplasia Society was established in April 2010.

Our main aim is to provide moral support and advice to parents and families affected. We also provide a network for sharing ideas and experiences.

We currently provide support for 21 families affected with ED in Ireland.

The establishment of the Irish society is also important to raise awareness of ED which we feel is an important role.

We had our first get together last October 2010 in Dublin. 12 families attended and many lifelong friends were made. We hope to make this an annual event and are planning another get together this year.

Ectodermal Dysplasia Society Ireland can be contacted on elaine@ectodermaldysplasia.org or 00353876951979

In January this year the ED Society moved to its own office. For the past 14 years the Society has been fortunate enough to run from the my home. However, we are thinking of moving to smaller accommodation, and the Society therefore took this opportunity to move to new premises and become financially independent as we take the Society forward.

As a consequence there is much work to do as we have taken the opportunity of giving the Society a different look by creating a new logo. This means all our literature will be revised over the coming months.

We recently created a document ‘Ectodermal Dysplasias International Registry’ which was published in April to help our members understand the difference between the NFED Registry and the Edimer Network. We have also created a document ‘Neonates and Infants with Ectodermal Dysplasia’.

A research project was carried out in 2010 ‘The Psychological Impact of Ectodermal Dysplasia’ and a paper published in October.

A further research project was carried out by a PhD student, from the University of Bristol, who conducted research into the experience of living with ED, in particular, how people with ED feel about their appearance and whether their experiences and feelings are affected by the knowledge that this is an inherited condition. Having completed this project she is now embarking on a second phase of the study, exploring the role of the health professional in the challenges that individuals with ED face.

Fundraising is going well for us and we are holding our first Charity Golf Day at the end of June.

Diana Perry
Norsk ED-forening

The Norwegian Support Group is ‘sleeping’ at the moment, but would like to keep up to date with the other Groups and be involved in the IEDN.

AADE Asociacion de Afectados por Displasia ectodermica

In 2010, Geneticist and Paediatrician Ms. Encarna Guillen, founder of the AADE, the Spanish Association, wrote an interesting article on Hypohidrotic Ectodermal Dysplasia.

In this article, Dr. Guillen said that the DEH is a genetically heterogeneous disease, since there are three genes involved, in Xq12-q13.1 EDA, in 2q11-q13 EDAR and in 1q42.2-q43 EDARADD, and three different inheritance patterns, X-linked, autosomal dominant and autosomal recessive.

The autosomal recessive EDARADD and EDAR are clinically indistinguishable.

The autosomal dominant, EDARADD and EDAR have a milder symptoms.

In the study by Dr. Encarna Guillén, 95% of the patients were randomly selected had EDA, and the remaining 5% had the autosomal dominant or recessive.

Social Administration in Spain, IMSERSO, has made a brochure on the ectodermal dysplasia, which includes the study by Dr. Guillon and further notes that the EDA gene on chromosome X, consists of 12 exons, eight of them in charge of encoding a transmembrane protein, involved in ectodermal development, whose mutations give rise to the pathology that characterizes this disease.

As genetic transmission occurs as x-linked recessive, the full syndrome occurs mostly in men carrying the gene responsible for women up to 60% of them have certain features usually incomplete, depending on the abnormal gene expression.

Gema Chicano
President From A.A.D.E. (ASOCIACIÓN DE AFECTADOS DE DISPLASIA ECTODÉRMICA)
What’s new at CEDSA — Summer 2011

It’s been a very productive summer here at the Canadian Ectodermal Dysplasia Syndromes Association. Our main focus has been on obtaining our charitable status, which we’re happy to say is progressing nicely. We hope to have crossed the final legal hurdles and be recognized as an official charity by the fall/winter of this year.

As a young organization, another of our primary objectives has been to fundraise. Our efforts this summer have been concentrated on raising money to help the families of diagnosed children purchase cooling vests and to help both adults and children with other financial challenges around dental and medical bills. We are excited to report that we have a fundraising Dance for Cool Kids coming up in September — an event that will take place in Renfrew, Ontario. Ticket proceeds along with monies raised from our silent auction will go directly towards supporting our members. We’d like to thank NFED and the Ectodermal Dysplasia Society in the U.K. for helping with the fundraiser and sending us beautiful photos to use.

And speaking of cooling vests, we were touched by the generosity of Dean Sainsbury of ARCTIC HEAT USA, whose company donated an Arctic Heat Body Cooling Vest, valued at $220 CDN, for our membership draw. We are currently in discussions with another manufacturer of cooling garments, and we soon hope to have a vest “demo” section on our website.

Finally, we have devoted much time into research, particularly focused on how the Canadian health system (which is regulated provincially) handles cases of ectodermal dysplasia. It has been rather discouraging to note that, in particular, diagnosed adults receive little to no financial assistance, a trend we hope to change in the future.

Looking ahead, CEDSA’s goals are many: to increase our membership base as well as our fundraising initiatives, which, in turn, will allow us to better serve our members through research, financial support and advocacy.
Greetings from the United States! I would like to introduce myself. My name is Judy Woodruff and I have served as the new executive director at the NFED for about eight weeks now, and I already have had the pleasure of meeting many of our international partners at our National Family Conference in July. What an honor it is to be part of such an outstanding group of organizations dedicated to serving those affected by ectodermal dysplasias.

The NFED has had a busy year, and we would like to share with you the following information from our organization.

**Support**

- **Save the date!** The 2012 National Family Conference is scheduled for **July 19-21, 2012**, Buena Vista Palace and Spa, **Orlando, Florida**.

- **Education Scholarship Program** – We are helping six students attend college this fall. **We granted them scholarships totaling $10,000.**

**Research**

- **Ectodermal Dysplasias International Registry** – We are pleased that 1,247 people have completed a profile in the Registry but know that we still have progress to make. Please encourage all of your members to go to [https://nfed.patientcrossroads.org/](https://nfed.patientcrossroads.org/) and complete a profile for every affected individual and carrier in their family. The Registry is available in English, French, Italian and Spanish. Members can also visit the Registry to easily access de-identified information to learn more about the ectodermal dysplasias. For example, you can query to learn how many people on the Registry with HED experience nail issues. The Registry is a key tool in linking families with interested researchers and further describing the syndromes.

- **Ectodermal Dysplasias Classification: A Model for Integrations of Clinical, Systems Biology and Bioinformatics, October 2012, Medical University of South Carolina, Charleston, South Carolina.** The goal of this conference is to classify the ectodermal dysplasias using an interactive internet based database that can be used by affected individuals, clinicians and scientists. This system can be used to help with diagnosis and to locate the potential involved genes and molecular pathways. The significance of the conference results is twofold: a new classification approach will foster a better understanding of ectodermal dysplasias and will open new field of research. This conference is a follow-up to the event held in 2008.

- **Ectodermal Dysplasias Research:** We funded the following projects in 2011.

  - **Telomere Defects in the Poikiloderma with Neutropenia Syndrome**, Alison A. Bertuch, M.D. Ph.D., Lisa L. Wang, M.D., Baylor College of Medicine.
  - **Toward A Cure for Skin Erosions in AEC Patients**, Maranke Koster, Ph.D., Peter J. Koch, Ph.D., University of Colorado, Denver. We will publish a special research report in November showcasing 30 years of progress and the latest developments in projects we have supported.
Treatment

- **Treatment Assistance Program** – We have helped three individuals with $45,000 to help pay for their complex care; $4,000 to pay for wigs for two individuals and $490 to help one individual get an air conditioner.

- **Dental Treatment Centers** – Families can now choose from 14 different Dental Treatment Centres throughout the United States to receive oral health care from a team of experienced providers. It’s also been a great opportunity for dental students at the centres to learn about the syndromes. Our goal is to increase families’ access to quality and affordable oral health care.

These are just a few of the key highlights from the 30th Anniversary year of the NFED. We look forward to continuing the work that Mary Kaye Richter began all those years ago, and we look forward to continuing our mission of serving the individuals and families affected by ectodermal dysplasias. Please feel free to be in touch with our office if you have any questions or need anything. I look forward to meeting many of you in the future and to working with you.

Warmest regards,
Judy Woodruff
Executive Director

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*Please don’t forget the “International Conference on ED” next year (1st to 3rd June 2012) in Erlangen, Germany.*

*Stay informed and register for the newsletter at [www.ed2012.org](http://www.ed2012.org).*

*Would you please inform all professionals from all the different medical fields related to Ectodermal Dysplasia of this meeting and please encourage them to participate. It is very important that as many professionals as possible attend this meeting to make it a success.*

*Ulrike*
HELLO FROM POLAND

My name is Elizabeth Blum, I am a mother of a wonderful seven-year-old boy with Hypohidrotic Ectodermal Dysplasia. My son was diagnosed at the age of 5. It was the first contact with the National Foundation for Ectodermal Dysplasias and The Ectodermal Dysplasia Society.

One day my son found a NFED newsletter. When he was looking at the photos all of a sudden he said "Mom, they look just like me." I said that these people also have ED. My son smiled and replied, "Mom, call them and let’s make picnic together". That was the moment I understood that contact with other people with ED is very important for our children. I started thinking about founding an association.

I owe so much to the National Foundation for Ectodermal Dysplasias, Mary K. Richter, and The ED Society, Diana Perry. The first e-mails, information, support, the joy of our accession - priceless.

I was watching with a great joy as our Association was being formed thanks to the involvement of unselfish help of my friends, absolutely not related to ED - Iwona Miedzinski, Magdalena Lietzau and Wladyslaw Harasimowicz. I can honestly say that without their persistence, a great heart, good organization and knowledge our association would not have been established.

That way STOWARZYSZENIE NA RZECZ DZIECI I MŁODZIEŻY Z DysPLAZJĄ EKTODERMALNĄ ORAZ ALEGIĄ "JESTEŚMY" was established in Poznan. Our goal is to seek contact with other families, offer them support from our group, provide all necessary information and willingness to cooperate for the good of our children and youth. We are committed to leading them towards an active social life as well as boosting public awareness of the needs of people with ED.

The cooperation of our families consists in providing a variety of information but also supporting them at difficult moments. Unfortunately, our children are confronted every day with the lack of acceptance and understanding. The big problem is an access to specialists and even obtaining a correct diagnosis or attendance to schools and kindergartens. Unfortunately, some families do not want to reveal their problems and isolate themselves from the rest of society. I’m hoping that we can persuade them to act because we are a strong and organized team!

There are a lot of tasks ahead of us to be accomplished. Currently, our Association has joined the National Forum for the treatment of rare diseases, which was founded in 2005. The Forum brings together all the associations, which seek care and treatment for people with rare diseases. Soon the talks with the Ministry of Health on the establishment of the register of rare diseases in Poland will be held.

Thank you for such a warm welcome. We are very pleased that we belong to such a great family. The ED Society gives us support but you also inspire us to act - we received the wonderful news of George Keeble. Our congratulations!

Best regards,
Elizabeth Blum