



October 2021



## From Diana Perry

**This year we should have been celebrating 21 years of being a charity and 25 years of voluntary work from myself with an Anniversary dinner and dance. However, sadly due to Covid, this couldn't take place. I am so proud of how far we have come, but we couldn't have done it without the help and support of our Trustees, Medical Advisory Board, Staff and all our ED community past and present.**

**So we want to share this celebratory edition with you all, to learn a little bit more about the Society and how it all began.....**

## For my family, it all started 30 years ago!



When our twins, Joseph and Philip, were 2 years old, they had a genetic test which confirmed Joseph is affected by Ectodermal Dysplasia (ED), but Philip wasn't. We were both relieved and devastated.

This was the beginning of a very long road. The Paediatrician and GPs knew nothing about ED, so it was down to my husband and I to find as much information as we could to help with the day to day management.



Where to go for this information – we really didn't know.

When the twins were four along came Jack. As soon as we saw him we knew he was affected by ED. However, the road this time was a lot easier, we knew more about ED and how to care for a child affected by ED. Or so we thought...

Living with ED is not easy. It is about how to ensure the child is safe, happy, has the same opportunities at school, is treated the same in social environments, etc. For all this to happen, you have to be prepared.

Once we had a diagnosis, I realised I had a lot to learn to help the boys throughout their life, and was lucky to have found a support group - the British Ectodermal Dysplasia Support Group. As I began to learn more I felt the need to pass this on to others and my passion sent me on my new journey.



## Thank you for joining me on my journey

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# Highlights From

**1996** - The original support group was set up in 1984 when, Denise Matthews and Jane Faherty, two mothers of children affected by ED met by chance, and with the help of Prof. Angus Clarke, arranged a meeting for the few families known to them. At that time there was no internet or emails, so running a support group and finding new families was extremely difficult. In 1992 Moira Hargreaves took over running the group. In 1996 I contacted her for support and to offer some help. Jane, Moira and Veronica Boyle are the longest standing members.

An international team of researchers, which included Prof. Angus Clarke (Geneticist and Chairman of our Medical Advisory Boards), identified the gene responsible for X-Linked Hypohidrotic ED and provided a drug target for scientists.

**1998** - The NFED in America, who have been a wonderful support to me over the years and helped me get the Society off the ground, mooted the idea of holding their annual family conference in the UK.

**2002** - David Wyatt, who is still a member today, created our first website and we created our first logo.

I gave my first presentation to the 1st International ED Conference in Sweden.

**2000** - The NFED held their first annual family conference outside the US here in the UK. We established a Steering Committee to take the Group forward to obtain charity status.

The gene responsible for Incontinentia Pigmenti was identified by an international team of researchers including Susan Kenrick (UK).



Charity No. 1089135

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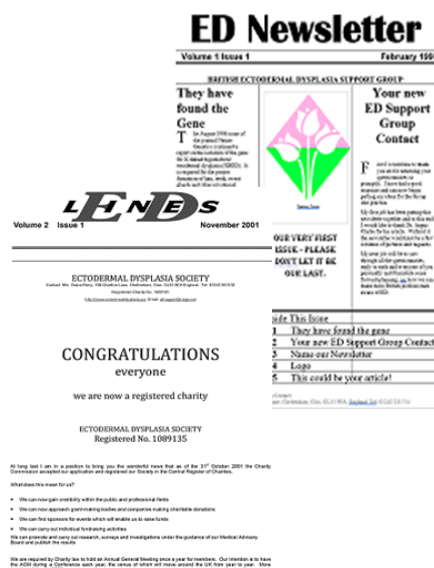
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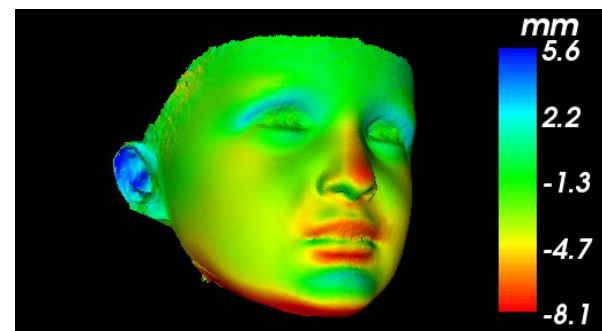
**1997** - The next I knew, I was sent the address book of 50 families in the UK to contact, but only 6 replied! I was given the Building Society book which held £200, and my journey began. I worked from home and produced our first newsletter in 1997, but, I needed a name for this newsletter. After putting it to our readers I was grateful to Peter Crawford, my boys dentist at the Bristol Dental Hospital, who came up with the name and EDlines was born!

**1999** - We created a leaflet and posted this to all NHS Trusts in the UK to bring awareness of ED.



**2001** - The Ectodermal Dysplasia Society became a registered charity. The Medical Advisory Board (MAB) was established.

We began assisting families wishing to obtain Disability Living Allowance grants for their children.



# Then To Now ...



**2006** - I gave my third presentation to the 3rd International ED Conference in Copenhagen.

**2004** - We saw the launch of the 2nd International ED Conference this time at the Eastman Dental Hospital in London, organised by Prof. John Hobkirk, who had become a member of our Medical Advisory Board in 2000. These conferences are now held every three years.

**2008** - We held our first annual ED Christmas Party which was a huge success, with 120 people attending. The ED Society Support Fund was established.

The Ectodermal Dysplasia classification studies began with a conference explaining the idea behind this event was that the advances in molecular genetics from the past two decades have given the opportunity to rethink the whole idea as to what is an ectodermal dysplasia (ED). We reviewed the types of ED for which the relevant genes are known and discussed how we could best group the EDs into different categories. Following this conference we held the International Group Leaders meeting. We updated the website and created a new logo.



**2003** - We employed our first member of staff, Sue.

**2007** - The International ED Support organisations came together for their first meeting and these meetings are now held every 18 months, which I co-chair.

**2005** - Our first conference was held at London Zoo. The children went off to the zoo while parents attended the conference. This was the first time we had been involved in facial 3D filming. Children and adults with ectodermal dysplasia have subtle differences in face shape when compared to the general population. These variations are attributable to the genetic coding of the individual. We hope that differences detected for ED might eventually assist in the diagnosis of individuals with a mild presentation of the condition. Our thermoregulation study began. The results showed many children have a low normal temperature.



**2009** - The Society connected with Amazon Smile and Easy Fundraising. We set up a closed and private Facebook Group. We made our first connection with Edimer, the organisation taking the treatment trials forward following the development of a recombinant protein (EDI200; formerly called APO200) as a potential therapy for XLHED.



**2013** - Edimer Pharmaceuticals' X-linked Hypohidrotic Ectodermal Dysplasia (XLHED) Newborn Clinical Trial which was the first to test a potential treatment for boys affected by XLHED.



**2015** - Danielle joined the office. We increased our office space by renting the office next door to ours and refurbished them into one larger office with a grant from the British Association of Dermatology. The XLHED clinical trial completed recruitment in 2015, sadly, they didn't see significant changes in sweat gland function and other early markers of biologic activity.

**2011** - Diana and Sue moved into our new office. Updated our website and established the website for the Ectodermal Dysplasia International Network (EDIN). We launched a Temperature Research programme in Portsmouth with Prof. Tipton. This is still ongoing today with interesting results along

**2014** - I attended the International ED conference and International Group Leaders meeting in Italy, when we were given a very detailed presentation by Edimer.

**2009**

**2010**

**2011**

**2012**

**2013**

**2014**

**2015**

**2010** - The research project "The Psychological Impact of ED" commenced - Heather Brant (PhD student). The Irish Branch of the Society was established by Elaine Alyward, Trustee who lives in Ireland. Gave a presentation to Dermatology student doctors at the Birmingham Children's Hospital to bring awareness of what it is like living with a genetic condition. This was so well received, it became an annual event.

**2016** - Research investigators involved in the XLHED trial concluded that the babies didn't receive the protein. This was a significant impact. One of the investigators, Dr. Holm, explored a way of earlier dosing that would work not only in affected humans. As a result of his research, we started with ER004 in utero in the spring of 2016 and a third child was born. All three treated children have since been able to sweat with no problems during summer time. They also show no tooth problems and clearly more tooth germs of the secondary dentition than the affected brothers. This was such a relief.



**2012** - The Womens Survey results were published by the NFED. This survey laid the groundwork to verify that breast and vaginal concerns are associated with the ectodermal dysplasias.





**2016** - The information, resources and work kept on growing, it was time we updated our website - again, and a new logo was created.

We were all extremely saddened to hear of the passing of Fergus Gordon who was our ED Scottish representative. Fergus had the AEC syndrome of ED.



**2017** - We welcomed Jaye to the office. Both she and her son are affected by Ectodermal Dysplasia.



**2019** - We created an Educational Health Care Plan sample document relating specifically to ED.

In July and September Diana attended the Communique and BMA Patient Information awards in London. The Society were awarded for "Excellence in Writing", highly commended and runner up for "Equality, Diversity, and Inclusion"

*"Everybody's Different was a slam dunk clear winner! It was inclusive, easy to use and written with great empathy for children with this rare condition. It is easy to imagine this being used as a teaching aid in schools or with a parent. Simple but vivid explanations will help increase understanding and empathy among children's classmates, and potentially reduce bullying."*

*"The book the ED Society have written is precisely what kids need to refer to at a young age - reminding any child they should dream big and live the life they desire. I can only support this piece of work and encourage more."*

**2020** - The Covid-19 pandemic began and the UK was placed into lockdown with everyone working from home. As if the pandemic wasn't enough we had a computer crisis where two of our computers ceased working. The enthusiasm and support from our ED community was amazing sending in donations to get us up and running again. All meetings were held virtually which Diana attended, keeping our international and national presence. Sadly, the Christmas party was cancelled but we managed to hold some events online for our families.

Zoom was the word for 2020!

**"EspeRare partners its XLHED program with Pierre Fabre"**

In December 2020, after a couple of years of set backs and dashed hopes with the clinical trial, EspeRare entered into a partnership with the Pierre Fabre group to develop ER004, a novel in-utero protein replacement therapy for the treatment of X-Linked Hypohidrotic Ectodermal Dysplasia (XLHED).

2015

2016

2017

2018

2019

2020

2021

ED clinical trial came to the fore early enough to have a team Schneider, had successfully not only in animal models, but findings, he dosed twin boys and infant a few months later. sweat normally and have had normal saliva production better than their untreated exciting news!



**2018** - We created our 'Guide to Ectodermal Dysplasia' and also an ED children's book.



**2021** - Last year we were very fortunate and delighted to have gained celebrity endorsement and support for the Society from Francesca Jones. Francesca has competed at Wimbledon and the Australian & USA Open playing tennis. She was born with Ectrodactyly Ectodermal Dysplasia (EEC), has three fingers and a thumb on each hand, three toes on her right foot and four toes on her left. She also started life with webbed fingers.

**"the greatest thing in life is to do what people say you can't do".**

Still working from home, a lot more online activities took place, but as the year went on we, once again, cancelled the Christmas party but plans are being put in place to hold this virtually. Watch this space!

The ER004 XLHED program received the necessary green lights and is well on the way to commencing in the winter 2021/spring 2022.

It is very exciting that during the second half of 2021 both EspeRare and Pierre Fabre aim to start enrolling patients into the clinical trial.

The Ectodermal Dysplasia International Network became a legal entity and I am on the Managing Board.

## Testimonials to the ED Society



The Board of Trustees of the ED Society would like to add our congratulations to the EDS on this significant milestone. We offer our heartfelt thanks to Di for starting this journey and for her passionate determination to take forward the cause of Ectodermal Dysplasia. The very existence and continuation of the EDS is a tribute to her vision and leadership. The timeline in this newsletter reflects its growth from a support group comprising an informal handful of parents with children diagnosed with ED, to a fully-fledged organisation. It may be a small organisation but the EDS informs and influences the NHS and other medical practitioners, supports pioneering research, advocates worldwide to improve understanding and recognition of ED, and promotes new member organisations to the ED network. However, the core mission of the EDS is still to work with families affected by ED on a daily basis, from responding to Facebook enquiries and providing a virtual shoulder at the end of a telephone to helping families submit and argue their cases for financial support and recognition to ensure individuals affected by ED can lead active, fulfilled lives. The stories in this newsletter illustrate all aspects of this organisation so well. Thank you Di, to you and your stellar team, Sue, Danielle and Jaye, for your relentless dedication and energy to a community of people who really need you. Thank you too, to our Medical Advisory Board who give their time to guiding the organisation. We look forward to the day when we can all meet again more easily - staff, medics, supporters, friends and trustees.

Where do I begin....after having to fight with every doctor I'd seen for over a year when I had my first child, they made me believe I was over reacting and I was terrified they would take my baby away to finally finding the ED Society. I will never forget that phone call with Diana Perry she changed my life, I stopped living in fear that my son may die everyday and she set me on my journey for a diagnosis. Without the ED Society I would hate to think where I would be. From the support to being my biggest cheerleaders I owe them everything, the community has given me friends for life who are more like family and I know if I need any advice or support someone will always be there, You will always hold a special place in my heart! Love you all so much xxx

**Zoe McDonald**



**Richard , Ben, Cameron & Lennon**

Without you my granddaughter wouldn't have been awarded Disability Living Allowance which has made my daughters life so much easier. 18 months of fighting it, we walked into the tribunal and were awarded it straight away plus, as a bonus, it was backdated to the first claim date. Diana came down to Kent to help us in our cause. Without the Society we would have given up. We are so grateful to you all. **Mandy Hayes**



**Taylor Atkiss**

Where to start really? When we found out Taylor had Ectodermal Dysplasia we were like, "What is this? How? Why?" The head dental consultant at Birmingham Children's Hospital told us not to read everything on Google, and said there is a charity in Cheltenham The EDS, speak to them and they will help and give you info. Are we glad we did contact The EDS? 110% is the answer. Diana was amazing and helped us to understand what ED is and has always been there to help with any questions we have had. Now I'm a Trustee and Fundraising Coordinator, I am happier to help in anyway I can. So thank you to all the staff at the EDS Diana, Sue, Danielle, Jaye and you guys are great. **Stuart Atkiss**

The ED Society is a Society driven by the passion to help people, especially children. That passion is personified in Diana and the team. It was meeting Diana and attending an ED Society Christmas party that made me want to help the Society as best I can. After 25 years, Diana's desire to help remains as strong as ever, and her ability to pull together a first class team has helped many, many families. Congratulations!

**Prof. Mike Tipton (Medical Advisory Board)**



**Simon, Chrissy and Tessa  
Lees-Jones**

Very lovely, truly inspirational dedication & progress. We have all had so many shared challenges of the un-known which you & the docs have done so much to help for so many families. For us, the moment of truth came when we met Prof. McGrath at St Thomas' in London who was able to relate my & daughter Tessa's entire personal lifetime of health issues without having met us ever before that day. Like so many Mothers, my darling Chrissy had to summon all her courage to treat Tessa's scalp erosion with the conclusive solution that salt water treatment was the only thing that would help, but not anything at all easy to do – they are both heroes like so many other families. **Simon Lees-Jones (Trustee)**

I just want to say finding the ED Society has literally changed my life. It has given me hope and made me feel like I could cope with the overwhelming lost and confused feeling that I and my family had when we first had a diagnosis over a year ago, just before lockdown. I can't thank you all enough. I think you do an amazing job in emotionally supporting, educating and providing support with all aspects of medical, school, work and benefits. In three words - amazing, supporting, uplifting. I literally can't wait to meet people when Covid restrictions allow. Many thanks for all you help xxx congratulations on your 21 years xxx **Rachel Coombes**

Since I first encountered the ED Society some 20 years ago, it has transformed beyond all recognition to become the most progressive, responsive and holistic support group I know. A model for others to follow and a resounding success for Diana and her ever growing team. **Mike Harrison (Medical Advisory Board)**

## The Dentist's Tale

Long-standing members of the Ectodermal Dysplasia Society's Medical Advisory Board (MAB) were recently invited to comment on the 21<sup>st</sup> Anniversary of the Society, and Diana Perry's work in this field for over quarter of a century and this is the dentist's tale.

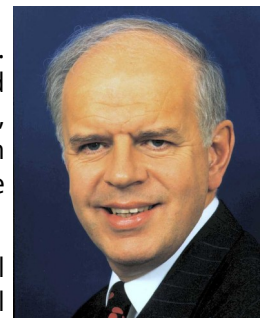
In 1977 three young consultants from different specialities, working at the Eastman Dental Hospital in London, established what is believed to be the UK's first multidisciplinary hypodontia clinic. Their proposals emerged after each had individually received several referrals of patients with ED and hypodontia. They initially met every three months to plan and coordinate treatment. Demand for the service soon grew, fueled by presentations at national and later international meetings. As a result, the team was expanded, and the clinic frequency eventually increased to fortnightly. The historic significance of this development is reflected in the time capsule in the new Royal National ENT and Eastman Dental Hospitals building which includes material relating to the treatment of hypodontia.

It was at a conference in Copenhagen that the whole Eastman team first met Diana Perry. This was a relationship which proved pivotal in building links between clinical teams and patient support groups. Diana had a very effective and positive attitude to the ED cause, and she and the UK dental team agreed that, if possible, the next conference should be in London. Her diplomatic support helped in the birth of the ECT04 meeting held at the Eastman in 2004.

At that time, it was uncommon to hold conferences attended by both lay and professional individuals in significant numbers, and it was a tribute to Diana and other national delegates that they were able to foster this relationship. Articulation of the challenges faced both by individuals with ED and their carers helped to formulate professional responses at a time when this did not always have strong collective professional support.

The ECT04 conference was attended by a wide range of international delegates, representing both clinical and patient support groups. Many recall Diana's vigorous questioning of a very senior dental civil servant, after his presentation, in relation to NHS funding of implant treatment for patients with hypodontia.

The meeting included group sessions on Dental MDTs (Multi-disciplinary Teams) from which emerged a consensus statement on their structure and roles, which was subsequently published.



**John A Hobkirk**

The Society's Medical Advisory Board) (MAB) includes representation from a wide range of medical and dental specialities. Its responses to the sometimes-challenging questions fielded by Society members is, we hope, as of much of help to them, as it is to us in increasing our knowledge of the condition.

The Eastman Clinic began to be copied nationally and the Society took on the role of maintaining and publishing a data base of clinics in the UK. This is coordinated by a dental member of the MAB.

As readers will know one of the highlights of the Society is its Christmas party, to which members of the MAB are invited. It is always an extremely enjoyable family occasion enabling people to mingle. For the clinicians attending, it is an opportunity to meet former patients. In the case of senior members this may span two or three generations of the same family!

My congratulations to Diana for 25 years of sterling work and to the Society on its 21<sup>st</sup> birthday.

**John A. Hobkirk (Medical Advisory Board)**

As a doctor I regret how little I have to offer my patients with genetic skin disorders. Gene therapy is becoming a reality but we are a long way from a cure. I know too that there is a huge difference between the "snapshot" of a clinic visit and everyday life. So I value patient support groups, such as EDS, who can fill the gap, provide accessible information and put people in touch with others who live with the condition and truly sympathise.

Diana has set up and nurtured a highly effective organisation meeting the needs of people with ED and their families. She is a fantastic advocate for ED in international as well as local forums and is involved with ground-breaking research. She uses her own experience to teach others including health professionals about ED. For over 10 years she has helped me deliver national teaching, reaching hundreds of dermatologists, explaining not only the medical facts but also the personal and social realities.

Diana was quick to recognise the importance of different specialists working together, resulting in her multidisciplinary Medical Advisory Board. Diana's caring personality is always apparent at the Christmas parties which I and fellow MAB members very much enjoy.

The one useful thing I can always offer my patients is contact details for the EDS. Congratulations to Diana and colleagues on all your achievements and very best wishes for the next 25 years of the EDS!

**Prof. Celia Moss (Medical Advisory Board)**



**Prof. Celia Moss**

## **25 YEARS ON, OUR MISSION REMAINS—WE THANK YOU!!**

Our mission remains that every individual and family affected by Ectodermal Dysplasia is equipped with the knowledge needed to manage the condition effectively and live life to the full; and that medical professionals have the information and understanding they need to support those affected.

We continuously aim to increase marketing and fundraising for the ED Society. We want to do more with you all, together! We loved the success of our ED family quiz and are hoping to plan more of these through the year.

For the future, we can only continue to grow and bring Ectodermal Dysplasia to the forefront. Please continue to support us by setting up monthly or yearly standing orders and funding through your donations and fundraising events. Share ED far and wide to help make our small charity that little bit bigger. It has been extremely difficult during this past year, but we have been in awe and inspired by how many of you have still come together and done some incredible work for the Society. You have certainly been there for us in our time of need too.

**We thank each and everyone of you, for YOUR support, YOUR fundraising, YOUR volunteered time, YOUR encouraging words and YOUR invaluable donations.**

**We would love for you to share your stories and memories of the ED Society over the last 25 years, share your photos, share your stories, share your memories..... #Happy25EDSociety**