The Yearly ED-Lines

October 2022

Your donations. Your Impact!

"I cannot tell you how many families we have helped since 1996, but here is a snippet of just the last year!" Diana

Donating enables us to support our ED children through their school life, to get them the correct care and attention they so desperately seek.

Sending your children to School should not be filled with dread and worry, it should be a safe place for them. With our intervention, more than 20 school care plans and EHCP's have been put in place this year, ensuring they are carefully monitored to prevent any unnecessary danger.

Money raised by you grants our ED families the financial benefits they so rightly deserve. The ED Society pour their time and expertise into guiding parents through DLA applications for their children. But it is a sad realisation that for most, Ectodermal Dysplasia is claimed to be "no different to the problems any other child of their age will face", and the applications are turned down.

But we step in to get those decisions overturned. We work tirelessly on supporting documentation to appeal and will travel to our families to be by their side at tribunals.

We are proud to have assisted and overturned more than 10 DLA applications and appeals over the last year.

Did you know – we have a 100% success rate for DLA?

Lack of temperature control is a high-risk symptom in most ED syndromes. Many homes are not equipped to deal with the difficulties this presents. Homes either being too hot during the summer months, not enough windows, flats in tower blocks, not enough bedrooms, no gardens to cool off, only got either a bath or shower. Homes that are faced with endless issues, not fit for purpose, and causing a threat to our ED families health and safety. Homes dealing with damp and mould, giving rise to respiratory problems.

This year The ED Society are grateful to have helped multiple families, by writing to their local councils to help them find a home safe and suitable.

Your ongoing financial gifts allow the ED Society to flourish, to thrive and to win these fights.

Your donations have grown our ED community, widened our presence and given Ectodermal Dysplasia the platform it deserves.

SET UP YOUR MONTHLY GIFT TODAY

WE struggle. WE fight. WE win!

We are stronger together!

EDELIFE Clinical Trial of Treatment in utero for X-linked hypohidrotic ectodermal dysplasia

Prof. Angus Clarke, Institute of Medical Genetics, University Hospital of Wales, Cardiff

I am delighted to tell you that we are almost ready to recruit to this long-awaited clinical trial of an intra-uterine treatment for X-linked hypohidrotic ectodermal dysplasia (XHED). It has been frustrating that it has taken so long to get through all the steps of the regulatory process and then the contract between the clinical trial organisers and the hospital, but we are now very close. I am hoping that it may all be underway by the time you read this.

For those unfamiliar with the background let me summarise the story. The gene – known as EDA - lies on the X chromosome, which has long been known from how the condition is inherited. Its location on the chromosome was gradually narrowed down during the 1980s and it was finally isolated and sequenced in 1996. When the gene is disrupted, steps in the development of several organ systems fail to occur correctly. These are organs that require cooperation between the outer layer of the embryo (the ectoderm) and the underlying mesodermal layer. The most affected organs are hair, teeth, and several types of glands (especially sweat glands, salivary and lacrimal glands, the breast, and Meibomian glands in the eye).

Two researchers in Lausanne developed a hybrid molecule, based on the protein produced by the EDA gene, but with another component added to help it be transported across the placenta into a pregnancy, and across membranes within the body. This was tested in mice (pregnant females carrying affected males, and also newborn affected males) and was found to correct many of the features of the animal conditions equivalent to human XHED. It was also tested in a type of dog affected by the condition and was successful in them as well.

The treatment then moved to testing in humans and it was found to be safe in both adults and infants, but ineffective in treating newborn affected infants. That it didn't work in newborns was established by the Edimer sponsored trial of treatment a few years ago. It was no surprise; being given to newborns was too late in development for it to work, as the body's process of laying down these structures would usually be completed before birth in humans. It worked in mice as they are much less mature at birth, after a pregnancy of only 20 days.

Prof. Holm Schneider in Erlangen, Germany, then gave the treatment to three affected fetuses/infants in two pregnant women from about 26 weeks into the pregnancies (they included one singleton and one set of twins). This does seem to have been effective, at least in allowing sweat glands to develop and function in these boys. This has led to a team being established to offer the treatment as part of a trial, to assess just how effective the treatment is for correcting various aspects of the condition and seeking to confirm its safety. The companies involved are both European, EspeRare and Pierre Fabre.

Women will be eligible to take part in the trial if they are known carriers of a variant in EDA that causes the condition and are pregnant with a male foetus. They will then have an ultrasound scan in Cardiff to look specifically at tooth buds in the foetal jaw. If these are either absent or too few in number, and if they are prepared to go through with the trial protocol, they can then go ahead with the three sets of treatment from about 26 weeks through to about 32 weeks in the pregnancy.

However, I would like to emphasise that the trial protocol is quite burdensome, involving visits to Cardiff both during the pregnancy and afterwards, until the infant is 5 years old. We need to point out that, following the treatments, there may be quite a small risk of triggering preterm delivery of the pregnancy.

We would prefer the infants to be born in Cardiff, but realise that this will not always be possible. Where feasible, we also hope to arrange for an affected male relative of the treated infant to make one research visit to Cardiff.

Participation in the trial is not entirely straightforward and I am trying to make this clear without discouraging anyone. We do want everyone who joins to be thoroughly realistic about the demands it will place on the family and about the small element of risk.

If you are a female carrier of XHED, are pregnant or hoping to become pregnant, and would like to discuss this with me, then the best way is to make contact by emailing me at clarkeaj@cardiff.ac.uk

What have we been up to?

Rare Beacon #DrugRepo October 2022

I was intending to attend this conference to understand more about drug repurposing, however, I was very excited to learn that Beacon invited EspeRare, Pierre Fabre and myself to give a joint presentation. This meant all my expenses were paid for!

What is drug repurposing?

Drug repurposing is finding new uses for existing drugs.

Drugs are chemicals that modify the biochemical pathways in our cells. It is rare that a drug will only affect a single pathway, in a single way, with a single effect. This is why most drugs have side effects.

In drug repurposing, we use the side effects of a drug to treat a different condition.

The Covid vaccines are repurposed, which explains why they were rolled out so fast, and saved thousands of lives.

Working together with EspeRare (a not-for-profit biotech) and Pierre Fabre (a pharmaceutical company) we have repositioned and developed a landmark treatment for XLHED - the in utero Treatment Trials.

ICED June 2022

"Collectively, we are bringing awareness of ED around the world - and collectively, we are stronger."

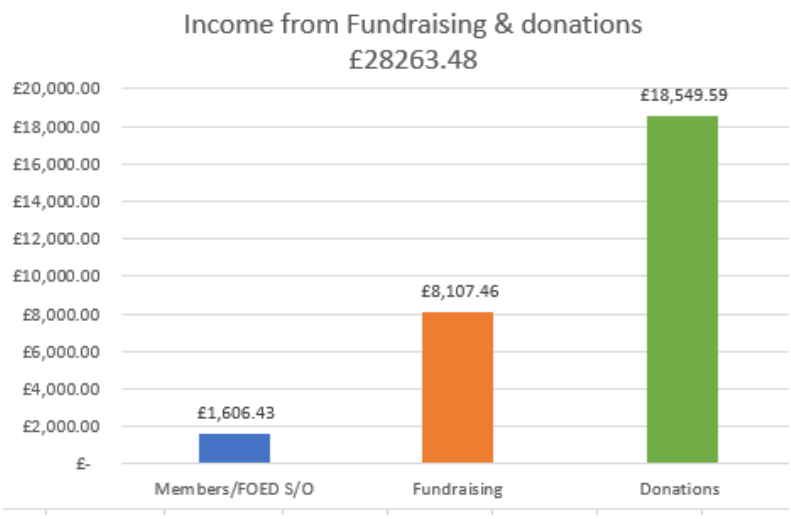
I attended the International Ectodermal Conference (ICED) in June this year. This was a 2-day conference with presentations on 'mucous membranes', 'Incontinentia Pigmenti', 'dental care', 'treatment trial updates', 'ED diagnosis and new genes', 'supporting parents of children with appearance affecting conditions', 'ocular manifestations and management update's, and some workshops.

It was a very interesting and informative conference, adding to the ED Society's knowledge. During the conference I was given the opportunity of presenting on film which you can watch now on our website. In this interview I talked about the ED Society, how it all began and how the importance of the XLHED trial is for us as a patient support organisation.

During the conference, the ED International Network Leaders had a meeting; this is an opportunity to discuss global events, research, trials, surveys, and more.

One of the big topics was of course, the XLHED clinical trials. I was interviewed whilst there and discussed the beginning of the ED Society, why we do what we do and the importance of the EDELIFE clinical trial for us as a patient support group. You can watch the interview on our website here - **Diana ICED 2022 Interview.**

Your Impact



We achieve great things because of you! The ED Society are in awe of everyone's time and efforts over this last year, despite the hard hitting effects of the pandemic and the cost of the living crisis we are facing.

In September 2022, we introduced the **"Friends of the ED Society".** It is great to see so many of you joining us and setting up your standing orders - thank you!

When you donate, your monthly gift can make a real difference in the lives of children and adults affected by Ectodermal Dysplasia. Funds raised by you, family and friends, immediately go to work providing support services and hope to our community, as we work towards helping those looking to find treatment for Ectodermal Dysplasia.

"The ED Society helped in finding probably the most knowledgeable dentist in whole of the UK for my son. I am more so grateful because then, I was new to the country and the whole medical system."

Priyanka, Mum to Aarush.

"Our son is thriving, in no small part thanks to the passion, dedication and knowledge of Diana and her team at the ED Society."

Lissa, Mum to Kaldi and Trustee for the ED Society.

"They were an incredible source of information and support. Especially at the time of the initial diagnosis, when we needed additional help and support. If you, or your loved ones, are affected by Ectodermal Dysplasia, I would urge you to check out, and support, the ED Society."

Scott, Dad to Ryan and Daniel

"I couldn't imagine a more caring and dedicated charity that are always there to support us and ensure our little boy lives life to the max. The Society is so caring, informative and in a word amazing"

Scott S, Dad to Austin

Achievements for the year

There are many areas in which Ectodermal Dysplasia affects our families lives, and the information and services we provide go above and beyond the general advice and support we give with the day to day management.

Over the past year, the ED team have helped with applications, appeals, supporting letters and we are happy to report we have been successful for our ED community in the following;

8 DLA applications and appeals

2 PIP applications and appeals

2 Blue badge appeals

5 Rehousing and accommodation needs

8 School letters/care plans

5 Theme parks "fast pass" for queuing

Multiple Covid related issues, such as letters for vaccination priority

Applications for Severn Trent Water scheme for families

Fundraising for Ectodermal Dysplasia

We want to give a BIG thank you to the following supporters, who have fundraised the following in the last 12 months;

- Chantelle Epton, for putting the ED Society forward to her employers at **Next PLC,** who have donated towards our Christmas Party
- Thomas Briggs, for raising £1095 during his **"Super Keeper"** fundraiser for ED Awareness Month
- Rachel Pearson, for nominating the ED Society to **Barratt Homes,** her employer, who donated £1,500
- **Leicester Rotary Club,** who raised £10,128 for the ED Society for the past year
- Paddy O'Reilly and our Trustee Lissa Pelham, for raising over £2,000 running this years **London Marathon**
- Julie Patterson at **Baildon Tangent Rotary Club**, donating £750
- Alice Todd (8 years old) for raising £300 by holding a **"guess the name of the bear"** fundraiser
- **5th Ormskirk Rainbows** for their sponsored walk, raising £157.50
- Adam French, running the **Manchester Marathon** and raising £1500
- Sarah James and family for raising £300 by completing **100 walks** during our May walking challenge

Do you want a simple, no effort way to donate to us? Have you tried easyfundraising?

Over 7,000 brands including eBay, John Lewis & Partners, Argos and M&S donate to The ED Society for FREE when you use easyfundraising online or in the app to shop with them.

Please remember to use easyfundraising when you do your Christmas online shopping this year.

This means you can raise donations for us when you buy anything from decorations and festive food shopping to gifts like toys, jewellery, tech, experiences and more!

These donations really help us and all you have to do is use easyfundraising when you're ordering the things you'll be buying anyway.....EASY!!

Since The ED Society signed up with easyfundraising, we have gained 98 supporters who have raised a whopping £1151, all for FREE!

Fundraising Story

Paddy O'Reilly

On Sunday 2nd October, I made my way up to Greenwich to attempt this year's London Marathon.

The reason that I wanted to do it?.....I have lived with ED without realising until my daughter was diagnosed when she was 19 (I was 45). My grandson is also affected.

I was feeling apprehensive as my training hadn't gone as well as I had hoped. Due, in part to to the fact that as result of living with ED I am unable to sweat, meant I struggled to regulate my temperature.

As I travelled up the weather was quite bad, and I envisaged 26 miles of being rained upon. Once I arrived at London Bridge station, the skies cleared, and the sun came out.

The walk from the station to the start area was full of nervous anticipation. The crowds of runners increasing the closer I got.

I have run marathons previously..... and one thing that never seems to change is the queue for the toilets

We were marshalled into our start pens and within a few minutes we were underway. I resisted the temptation to set off too quickly and fell into a comfortable rhythm.

I last completed London in 2010. I had forgotten how amazing the crowds were. There were points where, although I was never going to give up, I certainly questioned my sanity.

It's difficult to take in all the sights, but running around Cutty Sark, past Big Ben and Buckingham Palace was amazing.

As I turned in to the mall, and the finish came into sight, the realisation of what I had achieved was almost overwhelming.

I would like to thank Danielle, Jaye and the ED Society for all of their support and for giving me the Charity place.

I'm hoping to raise a little more before my fundraising page closes, so I would be massively grateful for any donations

https://tcslondonmarathon.enthuse.com/pf/paddy-o-reilly-8db3c

I have always believed that you can achieve anything you set your mind to, regardless of any condition or barriers. Believe in yourself.

Welcome and thanks

Many of you will be pleased as we welcome Jaye back to work after her maternity leave. Needless to say we have all missed her and it has been hard for us keeping on top of her work.

We welcome Kelly to the ED Society. Kelly began working on a voluntary basis at the beginning of the year and began as an employee in August. She is working tirelessly in digitising our work, bringing the database up to date following the Covid lockdowns; under GDPR this couldn't be taken off the premises.

We thank the Trustees for working hard looking at the future and where we want the Society to be in 20 years time. We have updated the website to make donating a lot easier and are working to modernise the site. They made the decision earlier this year to cease the membership and create 'Friends of the ED Society' so all your family and friends can set up standing orders to donate as much as they are comfortable with and as often as they like.

We thank the Medical Advisory Board for always being there to answer our questions and support the work we do for you. They will be working hard over the next couple of years putting the ICED25 conference programme together, giving their time voluntarily.

ED Events to look forward to:

ED Christmas Party 3rd December 2022

It is not long until the ED Christmas party—YIPPEE! It has been 3 years since the last, and we cannot wait to see familiar faces and some new families!

We have chosen a new venue for this years Christmas party - the Hatherley Manor Hotel in Cheltenham. A bigger, brighter room with outdoor space.

Juggling John will be joining us this year to provide some fun filled and daring entertainment for the children (us adults too!).

John will be showcasing many different talents such as a magic and circus show, fire juggling (weather permitting), juggling and magic on stilts, close-up/table magic, circus show and an interactive circus skills workshop.

We are excited to be welcoming Mad Hattie back for her fabulous face painting.

On the day there will be a buffet lunch, members of our Medical Advisory Board will be on hand to talk with our families, the winners of our raffle will be announced and there will be a visit and gifts for the children from the main man—Santa!

Agnes Jaulent, from Esperare, will be talking alongside Diana to share their amazing work on research in developing an in utero therapy for XLHED.

Rising star and model Hannah Harpin, who is affected by Haywells, will be joining us at the party to give an update on her adventures, her career and how she has overcome her battles.

Time is running out to get your tickets—you can purchase your tickets via our website; www.edsociety.co.uk/support/events/

Or, complete the enclosed registration form and return to us as soon as possible. Payment can be made online using our new easy to use donation page.

ED Coffee Morning Saturday 19th November 2022

Sharing a cuppa and a slice of cake is a great way to bring people together, so why not make Time for a Cuppa this November and help support the ED Society and families affected by Ectodermal Dysplasia? Get in touch with us today to share your plans and for your fundraising packs.

ICED25—International Ectodermal Dysplasia Conference 2025

We will be hosting the International ED 3-day conference in 2025. The last time we hosted this conference was in 2004 in London. In 2025, it will be held at the Austin Court Conference Centre in Birmingham in June 2025.

What is ICED?

Medical professionals, ED Society families, international leaders and patient groups are invited to a 3-day conference, where they will meet medical teams, scientists and patient representatives for presentations and workshops relating to the different aspects of ED.

The first two days are for medical professionals and the third day is for individuals and parents to learn more about ED from the medical professionals and to have the opportunity to have your questions answered. It will also be a great time to meet and mingle with our ED community.

Planning has already begun, and we will keep you all posted as we go! Do you think you will come? You can register your interest now by emailing info@edsociety.co.uk.

What's new in dentistry for children?

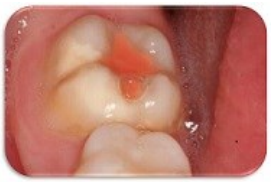
Kirsten Fitzgerald

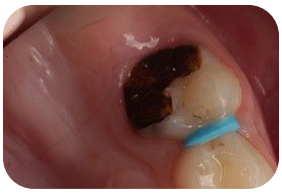
Pictures provided by Rona Leith and Isabel Olegario da Costa

The last few years have brought some really helpful techniques and treatments to children's dentistry. More and more dental teams are focusing on being able to provide the best care in the most child-friendly way. The days of numbing and drilling are not completely gone, but we have been able to take advantage of a range of techniques that help children receive the best care.

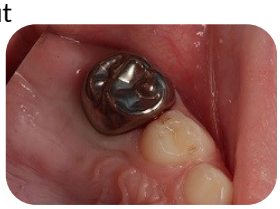
Of course, prevention of dental disease is still our number one goal, and your dentist can help you to achieve this. There is great value in regular visits to the dental team for examination and application of simple treatments such as fluoride varnish and fissure sealants.

Sometimes even with the best efforts, children develop some decay. Rather than "drill and fill", we can now use materials that help to stop the decay from progressing and allow the tooth to recover and heal a little. These techniques are useful for most children, but especially for children with ED as they help to maintain a positive attitude to dentistry by avoiding difficult procedures. So what are these new techniques?



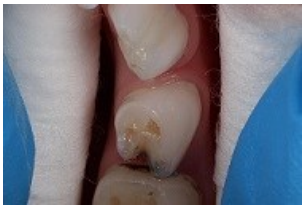


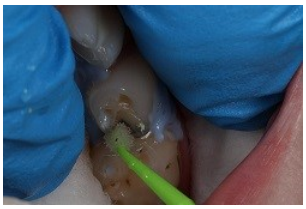
Silver diamine fluoride: this is a liquid that we paint on the teeth in tiny amounts in order to arrest tooth decay. It is a bit like a fire-extinguisher – it stops further spread of the decay, but it does not rebuild tooth structure. The downside is that it turns the decayed part of the tooth black, so you will need to talk to your dentist about that especially if it is a visible area.

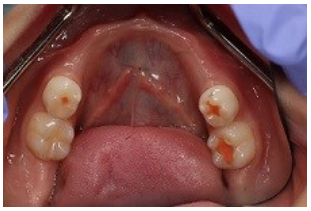


Hall crown technique: this is a method of placing a little silver crown onto a baby molar without having to numb or drill the tooth. It is generally well tolerated by children. Sometimes it requires a preparatory visit to create a little space for the crown by placing a small elastic band between the teeth. A crown is selected that fits the tooth and is then glued on with a special cement.

Glass ionomer sealants: This is a type of sealant that can be applied a little more easily than the traditional "resin" sealant that takes several steps including the use of a blue light to set it. For a glass ionomer sealant, the tooth does not have to be so dry, and we simply press the paste-like material into the grooves of the back teeth with our finger. It's very quick! Another advantage is that the material itself absorbs and releases fluoride over time, so it has an anti-cavity effect.







Articaine: This is an anaesthetic (numbing) medicine that dentist have begun to use more for children when we are trying to avoid a painful injection at the back of the mouth. Because it moves across bone a bit better than our usual anaesthetic medicine it can be helpful to numb a molar tooth in the lower jaw. It does still need to be injected into the gum tissue, but often we find that it is easier than our older "block" technique.

If you have questions about whether these techniques are suitable for your child, it is best to ask your dentist who knows the child best.