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ECTODERMAL DYSPLASIA SOCIETY
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We are having a new Website!

We need your help!

Exciting news— The ED Society are in the process of having a brand new website, and we would like help from our members. We would be really grateful to have some fantastic pictures to be included on our site. If you have any lovely family photos, pictures of fundraising events you have taken part in or those wonderful new dentures, they would be great!

If you could please email them to danielle@edsociety.co.uk

We would also be interested to know our members' thoughts. Is there anything you would like to see new on our site? Or would like to know more about? Any ideas would be greatly received.

REMINDER

The Annual General Meeting of the Ectodermal Dysplasia Society will be held at 10 Newcourt Park Cheltenham Glos GL53 9AY, on Saturday 7th May 2016 at 10.30am. If any members are interested in standing as a Trustee, please call or email the office to find out more information about what is involved, meeting commitments, etc. and to request a nomination form.

AGM
Annual General Meeting

Medical Advisory Board Members

Prof. Angus Clarke	-	Clinical Genetics (MAB Chairman)
Prof. John Hobkirk	-	Prosthetic Dentistry (Implants)
Prof. John McGrath	-	Genetics, Molecular Dermatology
Prof. June Nunn	-	Paediatric Dental Surgery
Mr. Colin Willoughby	-	Ophthalmology
Mr. Martin Bailey	-	ENT
Prof. Michael Tipton	-	Human Applied Physiology
Prof. Nichola Rumsey	-	Psychologist
Mr. Mike Harrison	-	Paediatric Dentistry
Dr. Claire Forbes-Haley	-	Restorative Dentistry
Prof. John Harper	-	Paediatric Dermatology
Mrs. E. Howard	-	Clinical Fellow
Dr. Fiona Browne		Dermatologist
Dr. E. Jones	-	Clinical Genetics
Prof. Celia Moss	-	Paediatric Dermatologist

Trustees

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

Staff

Sue Beard	-	Accounts / Website
Julie Cox	-	Administrator
Danielle Gue	-	Administrator

Volunteers

Diana Perry	-	Chief Executive Officer
Fergus Gordon	-	Scotland
Elaine Aylward	-	Ireland
Kerry Russ	-	Fundraising Co-Ordinator
Stuart Atkiss	-	Fundraising Co-Ordinator

Membership

You will be unable to gain access to the Members' Area of our Website until we are in receipt of your 2016 membership form.

Please return your form as soon as possible to ensure you have access.

Blue Badge

To obtain a Blue Badge you can either apply online at **https://www.gov.uk/apply-blue-badge** or telephone your local Social Services Blue Badge Team Department for an application form.

Many of our families already have a Blue Badge; if you would like Diana to help you complete the forms or appeal if your application is turned down please email **diana@edsociety.co.uk**

Symptoms Questionnaire

Could you please return a Symptoms Questionnaire for each individual who has ED in your family? This document will hugely help us when answering any of your questions or assisting you with DLA, appeals, tribunals, schools, doctors, etc.

This is available for download from our website.

Disability Living Allowance, Carer's Allowance and P.I.P

Diana is always happy to help you complete the forms.

It's best if the forms are right from the beginning in the hope that the claim will not have to go to appeal or tribunal.

If you are unsuccessful, Diana is happy to write an appeal letter for you and if necessary, attend a tribunal with you.

Lets Get Fundraising!!

Fundraising season is here—the warmer months are a great time to hold and take part in events.

Fundraising events and activities are principally about raising much needed funds for the Ectodermal Dysplasia Society, to raise public and professional awareness of Ectodermal Dysplasia and to aid and support the work the ED Society provides to individuals and families affected by ED.

Cycle rides are becoming a firm favourite and they're so simple to organise. We can give you and your co-rider a free ED Society 'No Sweat' T-shirt.

Here are a few more ideas to get you on your way—

Another easy and simple one to organise is a coffee morning. Get all of your friends and family together, maybe everyone can make some cakes for a sale too?

Schools often donate to local charities, why not ask at your children's school?

Sponsored swimming and running events?

If you are running in a marathon or participating/organising a big event, a t-shirt will help you to stand out and bring more awareness of ED. If you would like any please get in touch—and don't forget to have a photo wearing it!

Do you need a clear out? Hold a garage or car boot sale?

How about sending the Society some stamps? Postage is one of our biggest expenses so this would help us out a lot!

Any good ideas are welcome—so please let us know and we can add them to the suggestions.

Some companies annually select a charity to benefit from their fundraising once a year, maybe ask your employer to put the ED Society forward? If you need any leaflets to spread awareness please let us know.

We are registered with Virgin Money Giving for our fundraising events. Go to their website **www.virginmoneygiving.com**, which is extremely easy to use, and create your own page. Let all of your friends know and the rest is done for you.

Sponsor forms are available for events and we would be grateful if you could please ensure the forms are filled in correctly using full names, house number/name or postcode. Without this information we are unable to claim gift aid.

Fundraising—Our Grateful Thanks

We have received many donations over the past 3 months which amount to over £3600. Thank you to everyone who sent in donations with their membership form and to all those who pay regular donations by standing order.

Our huge thanks ……

once again to Richard Pettigrew's employers, Bottomline Technologies, for sending in a further £100.

The Gallacher family for donating £550

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Mark and Vicky Macnair—for raising another £324. This was £124 from their stall and £200 profit from a meal they did for 40 people from Chudleigh Twinning Society – Well done!

Mark Boyden—who is about to run the Marathon and has raised £867 so far.

VWR International Limited—for £108.77; a number of their associates donate their pennies from their salaries on a monthly basis to a nominated charity of choice.

Sarah Hathurusinghe—donated £32 from the sale of Christmas cards.

Support for Fathers

Having a child with a disability affects all members of a family. No matter if you are a new parent, or an experienced one—everyone can react in different ways to the news that their child has a disability or medical condition. Contact a Family's Fathers' Guide aims to help dads with practical information and insights into how other fathers have experienced having a child with a disability.

We hope it can help you! The guide has information on:

- Dealing with other peoples reactions
- Relationships
- Dealing with employers and your rights in work
- Money advice
- When you don't live with your child
- Further places to go for information, advice and support.

Contact a family have many guides, all of which are free to family members who can call the helpline **0808 808 3555.** Also, they are free to download from **www.cafamily.org.uk/publicationlist.**

We Want to Hear Your Stories—Articles Needed

We are always in need of articles for our newsletter and, along with other members, we enjoy reading your stories. Members of the ED Society take comfort from reading individual or family stories as it helps to make them feel they are not alone. Please send in to us any useful tips you have, personal stories or experiences. The article can be as long or short as you like and include photographs.

- Positive stories of treatments and operations
- Your biggest problems and how you deal with ED
- Coping in School
- Dealing with ED when on holiday
- Fundraising events you have taken part in

Uniformed Services Diary

(By Daniel Sanchez)

This is my eighth diary entry for my Uniformed Services course. It's not long now until I finish it.

This term I started unit four, which was about working in a team. We all voted to pick two team leaders and I was one of the leaders chosen. Our team's goal was to raise £100 for the Army Cadets. We started by selling tickets for a basketball game at school. It was pupils against staff and the pupils won. We also organised a car wash for teachers' cars. I had to organise my team and make sure they knew what tasks they were doing and what items they had to bring along to the car wash. We had a very successful day and met our targets, and all of the teachers were pleased with their shiny clean cars. The Sergeant Major also got his car cleaned for free.

At the local detachment we have been doing a lot of field craft, both practical and theory. When I use camouflage cream I take most of it off with baby wipes and then wash my face well. It's not drying so it's ok. We have been using the nearby woodland for field craft, but it has still been quite dark in the evening. Last week we left someone behind by mistake and only noticed when we had got back to the detachment!

I went for a practice expedition for my Duke of Edinburgh award at Almondell country park. It was nice weather but not too hot so I enjoyed it. We were given a map and had to follow the route our Sergeant Major had planned for us. My team arrived 20 minutes before the others as they had got lost. I have ordered all of my kit for the real expedition which will take place in May. I am looking forward to camping outside overnight and taking nature photos to make a calendar, which is my personal expedition target.

In March I went on a weekend cadet camp. We went to the Pentland Hills near Edinburgh, and set up a harbour for shelter. We set off on a long hike, and practiced camouflage and concealment. One team would hide and take notes on the other team who were reacting to enemy fire drills. It was a very sunny weekend, but still quite chilly. It was good fun and I was really tired when I got home.

I have completed my skill section of the Duke of Edinburgh award, which was skill at arms. I was also awarded my Youth First Aid certificate from St John Ambulance.

The Long Awaited Journey

If like Katie, you are currently going through the long process of many dental appointments and procedures, head on over to **www.thelongawaitedjourney.wordpress.com** and have a look at her insightful, witty and relatable blog. We think it is fantastic!!

Ectodermal Dysplasia and WNT10A

Chao-Kai Hsu, John McGrath
St John's Institute of Dermatology, King's College London, Guy's Hospital, London, UK

As we know, Ectodermal Dysplasia (ED) comes in all shapes and sizes, with a search through the medical literature indicating over 150 possible different conditions. For many years, the cause of these various forms of ED remained a mystery, although with recent advances in genetic medicine and DNA sequencing, we are now gradually beginning to understand the true nature of at least some types of ED.

By the start of 2016, the molecular basis of ED had been discovered in about a quarter of all the different types, but significantly these discoveries also included the more common forms of ED, such that it is possible to work out the genetic faults present in well over half of people living with ED.

One of the recent genetic discoveries includes a gene called *WNT10A* (pronounced WINT-TEN-AY, although its full name is wingless-type MMTV integration site family member 10A). So, what is WNT10A and what does it do? Well, it turns out that WNT10A is a rather important protein that acts as the conductor of an orchestra, directing several other proteins that play roles in constructing and maintaining numerous body tissues, particularly those from the ectoderm – such as skin, hair, nails, sweat glands, teeth, etc.

If a gene mutation occurs in *WNT10A*, then ectodermal development is no longer in tune, and the result is a form of ED. In fact, genetic abnormalities in *WNT10A* result in two forms of ED, neither straightforward to pronounce, called Schöpf–Schulz–Passarge syndrome (SSPS) and odonto-onycho-dermal dysplasia (OODD).

SSPS was first described in Germany in 1971 by Dr Erwin Schöpf, Dr Johann Schulz and Dr Eberhard Passarge. Clinically, SSPS is characterized by eyelid cysts (apocrine hidrocystomas), increased thickness of skin on palms and soles (palmoplantar keratoderma), missing teeth (hypodontia), excessive sweating (hyperhidrosis), loss or reduction of hair (hypotrichosis), and abnormal nails (onychodystrophy), sometimes with other ectodermal developmental anomalies. Many of the features only manifest or worsen during adulthood. This late appearance of problems means that SSPS can be difficult for physicians or geneticists to diagnose – some people may be in their 50's before the diagnostic penny drops.

OODD was first described by Dr. Mahmoud Fadhil and colleagues from Lebanon in 1983. OODD shows clinical overlap with SSPS, including abnormal nails and misshapen teeth, along with skin thickening of the palms and soles and variable sweating problems.

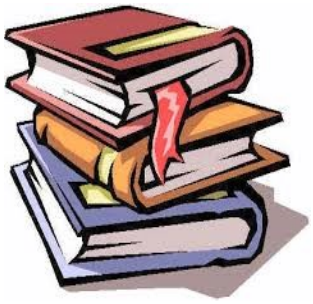
The main difference between SSPS and OODD appears to be the occurrence of eyelid cysts in SSPS, although analysis of the specific changes in the *WNT10A* gene has shown identical findings in some cases of both conditions, and therefore SSPS and OODD might be considered as the same clinical entity. SSPS and OODD, even when taken together as a single form of ED, are rare with perhaps less than 100 people with this type of ED appearing in the medical literature

The inheritance of SSPS and OODD is autosomal recessive. This means someone with the condition has to inherit two faulty copies of *WNT10A*, one from each parent. But what has become apparent is that some people who just have one abnormal copy of *WNT10A* can actually have some ectodermal abnormalities too – perhaps not enough to merit a diagnosis of ED, but nevertheless single mutation carriers can have changes in their hair, nails, teeth or sweating which demonstrate that even a conductor with one arm cannot keep the orchestra playing fully in the right key or with the correct tempo.

It is estimated that about 1 in every 200 people is a carrier for one faulty copy of *WNT10A* and that about half of these individuals will have clinical symptoms of signs of ectodermal anomalies. This means that about 1 in every 400 people in the world will have something wrong with part of their ectoderm – often hair or nails for females, teeth for males (although this varies a lot). So, the reality is that far from *WNT10A* only being relevant to a few people with rare forms of ED, it is in fact part of the daily lives of millions of people across the world, although most will have no idea that *WNT10A* is responsible.

At the moment there is no treatment available to correct the primary *WNT10A* conducting problem, although in the future it is hoped that recombinant protein therapy (as for ectodysplasin) might enter clinical trials. That said, however, it is clear that WNT10A signalling in the body is extremely complex and being able to restore exactly the right tune for a healthy ectoderm may not be straightforward. For people living with SSPS or OODD, as with most other forms of ED, the best clinical care should be focused on treating troublesome symptoms through medical practitioners and other healthcare personnel.

Exams and Overheating



As spring and summer approach many parents begin to worry about their child overheating in school, college or university and not focusing or concentrating properly, especially when it comes to the exam period.

Obviously children are not allowed to have a fan in the exam room as this may be a distraction to other children, making it near impossible for children with ED to complete their exam papers to their maximum ability.

We have a Health Care Plan which explains fully the difficulties children with ED have and how they are affected by overheating. Producing a document such as this to the school will enable them to fully appreciate the difficulties with overheating and how it affects the child's ability to focus and concentrate.

An exam room full of children on a warm day is very worrying. Provision should be put in place whereby the child with ED can take their exam in a different room where a fan can be used. During exam time it is possible for the school to have an invigilator on standby; if the child has begun the exam along with all their peers and discover they are struggling due to the heat, the clock can be stopped, the child taken to a different room and the clock restarted. This has happened for both my children several times over the years.

Looking to the future, if your child is heading off to university or higher education in September it is time to begin thinking of all the equipment they may need, such as air-conditioning, humidifier, fan, etc. You may like to apply for a Disability Student Allowance (DSA) which will help towards the purchase of such equipment. DSAs are grants to help meet the extra course costs students can face as a direct result of a disability, ongoing health condition, mental health condition or specific learning difficulty.

If you would like to discuss this further, request a Health Care Plan or for more information about DSAs please contact diana@edsociety.co.uk or call 01242 261332.

'Awesome!'

The following is an extract from an email as a result of Diana Perry helping this young man obtain his ambition to study dentistry, by putting him in contact with dental Consultants who helped him obtain a week's experience in a teaching hospital dental clinic together with future access.

When my son, who is 20 years old, and I received the news neither of us had ever envisaged, that he could not have implants, Diana and the ED Society were there to help.

This scenario had never been voiced by anyone, so we never managed my son's expectations any other way than "implants when you are older". My son, as he has always done, brushed it off and got on dealing with it bit by bit, devastated, but dealing with it. He channelled his energies by continuing to try and realise his ambition which is to study dentistry in 2016. He had sorted work experience near his University in London.

Once my son contacted you, well, you performed miracles. You know what you did practically for him – thank you! But I want to acknowledge also what you did personally for him at one of his lowest points. He is a very pragmatic young lad, which I am so proud of, but this implant scenario was a new ball game all together. With the combination of his determination, and the Society's contacts, you personally helped him at a most difficult time.

My son has now put in his applications, fingers crossed! I know he will do it one way or another, nothing has ever come easy for him and he is a determined young lad, never expecting anything. But I want you to know that your pro activeness with him and quick thinking, gave him the access to clinics in the near future that he could never have hoped to be involved with, senior people emailing him – priceless.

Diana, from the bottom of my heart – Thank You, and those words just don't seem sufficient. You are always so approachable, you helped myself years ago too. From a mum, who wishes I could give you a hug for what you have done, you would be able to see how much your kindness has meant to all of our family. In my son's words, what you did for him is 'AWESOME!'

Since receiving this email this ambitious young man has been offered a place at University!

If you or anyone in your family are having any troubles or have experienced any problems, please do not hesitate to get in touch with us at the Society. We are more than happy to help and support you the best way we can.

Help Keep Us Updated—Members New and Old!

We would like to keep our members' records as up to date as we can to help you. If it has been a while since you last completed a symptoms questionnaire for you and/or your child, and there have been significant changes to hair, teeth , skin etc. we would be grateful if you could please complete a new one. You can request one of these from us via email or by downloading it from our website.

Having a symptoms questionnaire which is current is a big help and makes it so much easier when answering your questions and providing support with DLA, Blue Badge applications and individual letters etc.

What We Do In The Office

When I meet some of our ED families I often receive the comment, "Oh I didn't know you did things like that" - so I thought I would like to write an article explaining what we do in the office.

There are four of us, myself, Sue, Julie and Danielle. I work completely voluntarily and the girls are employed part time, Monday, Tuesday and Thursday mornings 8:00 am to 1:30 pm.

Sue has been with us since 2003 and deals primarily with the financial side of the Charity, IT and management of the website, UK Support Fund applications for individuals wishing to obtain equipment such as air-con units, fans, Chillowpillows, wigs, etc., and the organisation of the Christmas get together.

Julie has been with us since 2008 and is responsible for the database of all our ED contacts and members, general administration duties and the 'follow-up' of families to see how they are and if there is anything we can help them with.

Danielle has been with us since 2015 and is responsible for producing our newsletters, checking through Disability Living Allowance and Personal Independence Payments forms, corresponding with families, helping Sue with our website along with the EDIN (Ectodermal Dysplasias International Network) website, and other general administration duties.

I, Diana, have been with the group since 1996 and have many duties; I attend conferences; make presentations to medical professionals and medical students; co-chair the international ED Leaders meetings and conferences whereby we are able to help our ED communities around the world; attend school meetings, tribunals for DLA or PIP with you, proof read patient information leaflets for the British Association of Dermatology, assist with research into aspects of ED, liaise with families and individuals, write articles for our newsletter and medical journals, write information leaflets and the content of the website, liaise with medical professionals on your behalf, attend Trustee and Medical Advisory Board meetings,

We have a Board of Trustees who meet three times a year and a Medical Advisory Board who meet twice a year— who they are and what they do is on the second page of this Newsletter. The Trustees, who are all voluntary, aim to keep the ED Society moving forward, ensuring their work, including the finances and policies, are in line with the Charity Commission rules, and provide advice and support to the Office; The Medical Advisory Board, who are also voluntary, help tremendously by writing articles, checking all the information I write, recommend medical professionals when families are struggling to find someone who has experience of ED, carry out research such as the Temperature Control project and have created and maintain a network of dental professionals around the UK.

What Can We Do For You?

- write individual personal reports for your doctors and schools etc.
- assist by writing letters and reports to help regarding rehousing
- write appeal letters for DLA or PIP
- assist when applying for a Blue Badge
- assist when applying for an Education Health Care Plan (EHCP)

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- provide a School Health Plan specific to ED
- ensure schools are providing the correct care for your child such as a fan, drinking water at all times, etc., by providing letters, and liaising with SENCO and the school
- attend tribunals and school meetings with you
- liaise with dentists or provide you with information to ensure you are not continually made to wait until your child is older before any treatment is carried out
- ensure you have been given all the dental options for you or your child allowing you to make informed decisions; we have a dental network around the UK
- talk with you via telephone or email regarding the symptoms of ED
- provide t-shirts and support for fundraising activities you may be carrying out for us
- liaise with medical professionals for assistance and advice on your or your child's care
- discuss genetics with you or your adult children to help informed decisions to be made
- provide an annual get together such as the Christmas party for families to get to know each other and to have the opportunity of speaking with the Medical Advisory Board on a personal basis
- assist with the transition to University and Student Disability Finance
- we have a Facebook page to help you speak with others who have experience of ED
- talk with you regarding lack of temperature control and behavioural problems which, for many individuals with ED, are connected

The list goes on …… if we do not have the answers for you we will find them!

As a parent of two boys I have chatted with many parents and often receive the comment "oh my goodness you are describing my child" - I have many years experience of my own children and those of our ED families and individuals. Any questions or worries you may have will never be too small or 'silly' for me to answer—I have been there too!

Don't struggle on your own—We are here to help

As we only work three mornings a week and you need to speak to me urgently, I am happy for you to call my mobile 07774 465712

If you ever have any troubles or queries, we are always here to help our ED Families. Please do not hesitate to get in touch with us – **info@edsociety.co.uk.**

Part of the ED temperature study is planned to run during the summer this year. The ED Society will pay for accommodation in Portsmouth and traveling expenses.

Heather Massey is in the process of putting together a short video to explain the study and to show you some of the facilities.

The study is planning to run during 28th May - 5th June and Mid July to the end of July. Some dates in August too.

If you and your family would be interested in visiting Portsmouth to participate, please contact Heather Massey heather.massey@port.ac.uk. She will be more than happy to chat with you on the phone to explain the study and send a short video too.

Rare Diseases campaign

The 2016 Rare Diseases campaign was launched on 29th February through every copy of The Independent newspaper for Rare Disease Day! The campaign will be presented at leading events and conferences worldwide throughout the year and the dedicated website is now live. In order to further raise awareness and support for Rare Diseases, you could join in sharing the campaign through your social media channels. You can find Mediaplanet on Facebook, LinkedIn, Twitter and Instagram.

Thanks for spreading the news!!

"Above everything else we need to recognise that every life has worth" **Yann Le Cam, CEO of EURORDIS**

Help Get The Right Support—With Contact A Family

Contact a Family is a national charity that exists to support the families of children with a disability whatever their disability may be. They were founded in the 1970's by families of children with a disability, who recognised that even if their child's conditions were different, they shared a common experience—of being a family with a child affected by a disability. They understood how important it is to support each other.

The challenges families face today—battling attitudes and inadequate services, trying to stay in work and stay together, or just trying to get a decent nights sleep, are all too familiar.

Everything Contact a Family do is about improving the lives of families with children affected by a disability. This is achieved by working directly with parents and carers.

Their vision is that families with children affected by a disability feel valued and are strong, confident and able to make the decisions that are right for them. They aim to ensure:

- Families with children affected by a disability know how to get the right support, for themselves and their families
- Families with children affected by a disability are confident to deal with the challenges they face
- The financial disadvantage that families with children affected by a disability face has been alleviated
- Families with children affected by a disability are understood, valued and included as equal participants in their communities and in society.

Contact a Family will help direct families to appropriate support and services provided by others, and campaign to ensure those services are there. They will maintain a connection with families through their networks and by being there for you whenever you need them.

You can get in touch with Contact a Family by calling their helpline **0808 808 3555**. They can also be found via **www.cafamily.org.uk, Twitter** and **Facebook**.