“Strangers a few years ago – now a rock for my family”

The ED Society is the single, most helpful, kind, and caring Charity we have ever come across.

Alice was a "difficult" baby. She wouldn't feed, her temperature was always low, yet she was always bright red and fussy if we wrapped her up. She had numerous allergies and febrile seizures. She had rashes and skin issues galore! Alice didn't cry tears from one side, she was constantly congested, and she couldn't poo. We felt like failures because she was a "failure to thrive".

Alice was referred to Endocrinology at Evelina at three years old because she just wasn't growing. Initially, we were told it was likely she had skeletal dysplasia (dwarfism). It was a long road to rule out so many things, but in 2020 we were told Alice had a clinical diagnosis of ectodermal dysplasia.

All the years of working with children with additional needs, all the family history of health issues, and all the medical training my parents have didn't prepare anyone for this unheard-of diagnosis. I barely knew what the ectoderm was!

We were given the ED Society’s website by a genetics consultant, and we felt so out of our depth. We joined the private Facebook group and have regularly asked for advice on there. Sometimes trivial things, like hair care or holiday plans. Other times much more serious issues like raging fevers and endless vomiting.

When I put on a post, I know the other ED parents "just get it", and I also know the other adults with ED are on hand for support and advice to help Alice live her best life. I was blown away when I asked for advice on managing Alice's fever. I was contacted by the ED Society’s CEO, Diana, and told to call her!

This is so far beyond anything we ever expected from a Charity. Diana listened to my fears and tears at nine o'clock at night. She gave me helpful advice, telling me what to say to 111 when they tried to tell me a 40° temperature is normal for a fever in a child. We all know it is incredibly dangerous with ED, unfortunately, 111 really don't understand such rare conditions. Diana offered her phone number for me to pass on to doctors and told me to get Alice seen. Alice was admitted to the hospital and stayed in for a few days. Without Diana's help, she wouldn't have been there, and there is a high chance the febrile seizures would have returned.

We have learned enough about ED to know that our son, Alfred, also needed support. He is of average stature but shares many physical features with Alice, if anything his hair and teeth are more typical of ED. Without the ED Society and the information they share on their website, we wouldn't have been so confident taking him to see his pediatrician.

We attended our first ED Society Christmas Party in December 2023, and I wish Christmas happened more often! All the children made new friends, friends who understood when Alice needed to strip off her jumper, tights, and vest to run around. Parents who understood why we didn't make our children wear coats and scarves outside! Most of all, parents understood the fear and the pride that comes with raising an ED family.

Supporting a normal lifestyle

Alice has been following the ED Society's Ambassador, Hannah Harpin, for some time. When she finally met Hannah in real life at the ED party, Alice was nothing short of starstruck. Knowing Hannah is there raising awareness in the wider world, but also here helping us all raise happier, more confident children means the world to us.

As for me, I had a bit of a cry upon meeting Diana in person. A stranger only a few years ago who is now a rock for my family. Continuing the crying, poor Danielle and Jaye also had to suffer my sobs, but I felt so understood and so supported! They were happy tears, I promise!

At the party we were lucky enough to have a chat with Angus Clarke, who is a Professor in clinical genetics, and what a wonderful man! Angus gave us some excellent advice and reassured us Alice's lead geneticist is excellent. He, without overstepping his medical boundaries, confirmed our suspicions that ED is likely on my side of the family. We are still waiting for our trisomy results, but Alice has a functional clinical diagnosis for now and that is enough to reassure us that we are in the right place.

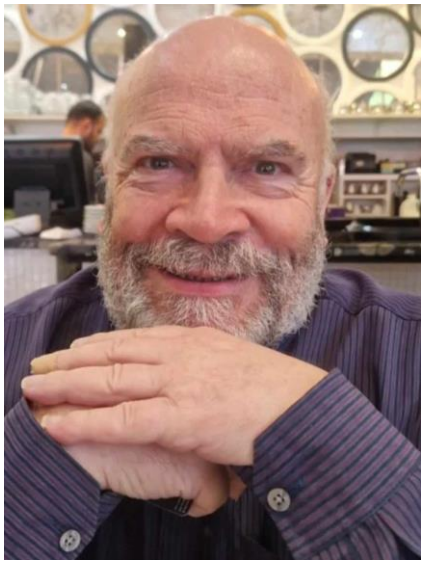
Following on from the party and using our knowledge from the ED Society's website and Facebook group, we have been able to suggest ED to my dad's doctors whilst he has been very unwell with kidney issues. Dad now has a clinical diagnosis and is waiting for genetic testing. He cannot wait to meet everyone at the 2024 party.

For anyone feeling unsure if their child is "clearly affected enough" for a diagnosis - join the group and ask questions. You can arm yourself for appointments and most of all you can become an advocate for your babies.

Finally, to the ED Society, I am so incredibly grateful you exist. We are so fortunate to have found you and we are eternally thankful for all you do for our family.

Supporting a normal lifestyle

ectodermal dysplasia society



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