

## The History of the ED Society!

Hi, I am Diana Perry and work on a voluntary basis for the ED Society. For my family, it all started 30 years ago. The twins, Joseph and Philip, were born at 32 weeks gestation.

### When the boys arrived -

The boys spent 6 weeks in hospital during which time the doctors knew there was something wrong with Joseph but had no idea what it could be. The Special Care Baby Unit Sister described him as '26-week retarded development' – not what you want to hear about your beautiful newborn baby. Joseph could not suck and had to be tube fed; his nose was always blocked with thick mucus and had to be extracted using a suction machine – boy I could have done with that machine at home over the next few years until he learned to blow his nose! His twin, Philip, was normal in development – he does not have ED.



There were lots of questions but no answers. So, after 6 weeks we were discharged from the hospital. Then the fun began.

Joseph was affected by failure to thrive. He vomited at every feed, struggled to breathe, and took about an hour to feed, whereas Philip took his feed in 10 minutes. I now know the warm milk was making him hot, which stopped him from wanting to feed and caused the vomiting! Easy when you know.

The doctors tried to help and changed his feed to soya, thinking he was allergic to milk. This made him worse, but due to the care and understanding of the hospital dietician, Joseph was put on a special diet from Great Ormond Street; I had to put 6 different ingredients into each bottle – no fun when you are already making 14 bottles a day!

During the first year we saw the Paediatrician every month; he was extremely helpful and supportive. He was happy to refer us to any specialists needed to help with the problems we encountered with Joseph. These were many as he was covered in eczema from head to toe; had oesophageal reflux; inability to control his temperature; feeding problems; constipation; a blocked, foul smelling nose; no teeth; no body hair; thin, sparse hair; puffy eyes; toenails which grew round the end of his toes; no eyebrows; recurrent ear infections, etc.

This meant an excessive number of trips to the doctors, dermatologist, ENT, etc.; monthly visits to the paediatrician; regularly visits to the optician and several stays in hospital.

Overheating due to non-functioning or lack of sweat glands has been the hardest to deal with.

### What was wrong?

Philip, however, was my sanity. The Health Visitor said I was neurotic and over-protective. I fed, changed, and bathed them both at the same time. I therefore worked out that it could not be anything I was doing that was wrong, because Philip was fine, but there had to be something wrong with Joseph and as it turned out I was right!

Joseph developed mentally and physically at the same pace as his twin Philip, except for his teeth.

Having successfully got through the first two years of the twins' lives, we had absolutely no idea of what lay ahead.

## Supporting a normal lifestyle

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## Teething troubles

When they were 2 ½ years old, we went to the dentist as Joseph had two very pointed top front teeth and a matching pair at the bottom. My Dentist recognised something was wrong and suggested it may be Ectodermal Dysplasia. We were sent off to see a Geneticist.

This was the beginning of a very long road.

We soon realised there were no alternatives and had to learn to live with it. 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 did not know.

When the twins were four along came Jack.



As soon as we saw him, we knew he had ED. However, the road this time was a lot easier, we knew more about ED and how to care for a child with ED to prevent overheating, etc., our GP believed in us, and we knew what dental work awaited him.

What we were not prepared for was a total absence of milk teeth. Jack's X-ray at age 2½ showed only three adult teeth, two at the top, one at the bottom.

When he was 3 ½ he was fitted with upper dentures, which did not fit very well; consequently, Jack wouldn't wear them. One day an insensitive adult visitor asked why Jack did not have any teeth; he promptly ran upstairs and returned with them in his hand saying, "here they are".

Both Joe and Jack had dentures from the age of 3.5. Joe is happy with his dentures and Jack now has lower implants and having had bone reconstruction last year he is now in the process of having upper implants.

Living with ED is not easy. It is life threatening. 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 must be prepared.

## Where it all began

When I had lived with ED for 2 years, I joined a support group which had been established by two mothers of children with ED in 1984, for mutual support. I took over running the group in 1996 when it had grown to a membership of 50.

During 2000, the name of the organisation was changed to the Ectodermal Dysplasia Society and a Steering Committee, and an eminent Medical Advisory Board had been established with a view to moving the group to charitable status, which was finally achieved in October 2001. Membership now stands at over 700.

As with many charities we have a wealth of experience and information which not only helps individuals and families, but also helps the medical profession.

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## Research

In 1996, a team of international researchers identified the first genes responsible for Hypohidrotic Ectodermal Dysplasia (HED).

By 2003, two doctors in Switzerland characterized the protein produced from the EDA1 gene and designed a therapeutic molecule, which was called EDI200.

During the period 2003 to 2013 many trials took place using tabby mice, dogs, primates and adult humans before moving to neonates when the first baby received treatment in Germany.

Since then, 10 more were treated in America, Germany, France and Cardiff, UK which showed no signs of any adverse effects.

However, the results were not as good as hoped for and the funding sadly ceased.

This was hugely disappointing news to the ED support organisations around the world and more particularly to the researchers.

Professor Holm Schneider from Germany and Professor Angus Clarke, two of the primary investigators in the clinical trial, felt that the research had been stopped too early and that there would be far better impact on the development of sweat glands and other key markers if the babies were dosed by injecting EDI200 into the mother's amniotic fluid, prior to birth.

In 2016, we were very excited to learn that Prof. Schneider engaged in a "Trial to Cure" which is a legal possibility under the German law. He administered EDI200 to 3 boys in utero who were affected by XLHED.

The early results are very encouraging. Male twins were treated in utero at 26 weeks and again at 31 weeks. An impressive number of tooth buds were noted after treatment. It is too early to know how many teeth will develop, but the number of tooth buds is encouraging.

The sweat glands have appeared to develop normally! Prof. Schneider noted that it was a very hot summer in Germany and the twins did fine regulating their temperature.

There is no doubt that having a treatment to correct some of the symptoms would alleviate all those agonising questions.

Having gone from impossible times of lack of information, little or no support, high mortality rates, etc., to in utero treatment is beyond our wildest dreams.

Once successful in XLHED boys it is hoped the research will move to treating girls and from there onto treating the different syndromes of Ectodermal Dysplasia.

It is also hoped that the success of these trials will, one day, help other debilitating conditions where they experience similar symptoms.

## What's Next?

During the second half of 2021 and in view of discussions with the EU and US regulatory agencies, both partners (Esperare and Pierre Fabre) aim to start enrolling patients into a clinical trial geared towards marketing approval.

<https://esperare.org/en/news/esperare-partners-its-xlhed-program-pierre-fabre>

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