# Caring for a child with xeroderma pigmentosum (XP)

Nicola Miller

**Nicola Miller recounts the shock and confusion of discovering her three-month-old baby suffered severe, sunburn-like reactions to daylight; the frightening and frustrating journey to diagnosis and the terrifying weeks spent straight afterwards in near-darkness. She describes the UV-protected life her family has learned to adapt to and the challenges of giving her young son as 'normal' and carefree a life as possible.**

January 2011 blessed us with the birth of our first child. It was a time of great celebration for us and we were blissful at the arrival of our perfect little baby son, Eddison. Eddison was just three months old when we had the first indication that something unusual was occurring. Our new neighbours invited us to their barbeque. It was a pleasant spring day, warm but nothing to note. I dressed Eddison in a long-sleeved top, lightweight dungarees and a sun hat and applied SPF30 baby sunscreen before walking next door. Due to nap time we didn't stay long, just enjoyed 20-30 minutes chatting, shaded under a large parasol, before heading home.

Later that afternoon Eddison looked flushed, but by the next morning his hands appeared puffy and swollen and his nose and forehead red. My initial reaction was he had experienced some kind of allergic reaction to the sunscreen. We saw our GP who diagnosed infected eczema and prescribed liquid Piriton along with Cetraben lotion.

Thinking it a one-off we set about applying lotion morning and night to avoid future flare-ups. Over subsequent months similar instances followed with reddening and swelling to face and hands; some more acute episodes required visits to the GP where Piriton was prescribed. I wasn't happy with the Piriton; firstly, it made him extremely drowsy and, secondly, it didn't seem to have any effect on outcome. In every case the cycle seemed to be the same: blushed appearance and swelling, reddening with some blisters, then peeling skin slowly returning to normal over a period of 10 days. My instinct was it had some relation to sun exposure, as the reddening started within 2-12 hours of being outside. Eddison had the most gorgeous soft skin with no patches of dryness so a diagnosis of eczema didn't ring true.

**Eddison, pictured here in his protective visor with mother Nicola, was 13 months old when he was diagnosed with XP.**

One day in early summer we took Eddison for his first visit to our leisure centre to go swimming. We walked the short distance from the car to the centre and enjoyed our swim. Later Eddison again appeared a little pink and I wondered if the chlorine might have affected his skin. The next morning, I went into Eddison's bedroom. He looked almost unrecognisable; his face was swollen, his eyes were almost closed and his hands were twice the normal size!

I screamed through to my husband. I genuinely thought he had experienced some kind of brain swell. On closer inspection, his face seemed badly burnt and most peculiarly his hands, arms and legs were bright red with a very distinct cut-off line at the edge of his vest. This made no sense; he'd been outside in a vest with a long-sleeved top, so it seemed impossible that he could be affected by the sun in such an extreme way. (We have since found out the swimming pool is UV-protected, so his sensitivity is such that this level of burning was sustained during the brief walk to and from the car.)

We revisited the GP, who said it appeared to be sunburn, which seemed extraordinary to me. I felt sure that they did not believe our account of his limited time in the sun. Later that day, still very anxious, we took Eddison to A&E. He was immediately rushed through to see a consultant. During the examination I

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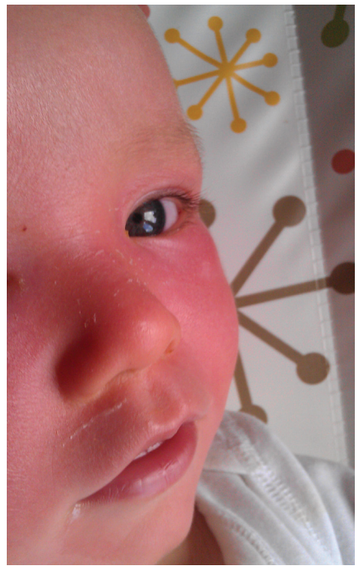
kept repeating that his appearance was worsening, and in fact the whites of his eyes had started to turn blood red (we now know this was a result of being examined directly under a fluorescent light). The consultant was confused and sent an urgent fax for a referral to a local dermatologist. At last we thought we were going to get some answers.

In the interim, waiting for the appointment, we searched the internet endlessly and started to wonder if perhaps it was a photosensitivity resulting from an allergy. We cut out soap products, stopped using deodorant, make-up, perfume etc. We insisted that anyone that held him did the same.

Soon a referral appointment arrived and we went armed with our photographic records. After endless questioning, the consultant declared it to be sunburn and gave us guidance on sun protection, reminding us how delicate a baby's skin is. We were horrified and felt judged as being incompetent and failing in his care. Around four months later we returned for a follow-up appointment and were aghast when the consultant simply repeated her guidance on sun protection and seemed sceptical to our suggestions of some form of photosensitivity.

I cried all the way home from that appointment. We still weren't being taken seriously. Feeling distressed and frustrated my husband called the GP to seek action. She was fantastic and recommended a private specialist for a second opinion. During this appointment I was immensely relieved when she said the simple words: "I believe what you say"! She agreed with the likelihood of a photosensitivity and suggested a referral to Guy's & St Thomas' Hospital, London, to be seen by a paediatric specialist.

For Eddison this marked a real turning point. We took him to London, again with our photos. After a brief chat with Dr Hiva Fassihi (who, it turned out, was the Lead in the UK's only XP clinic), and a visit from fellow XP consultant Robert Sarkany, it was decided that a skin biopsy would be done there and then.



**At first Eddison's flare-ups were diagnosed as eczema.**

We returned for his follow-up appointment on February 14, 2012, expecting to hear the biopsy was clear. Immediately we entered the room I sensed the results were going to be involved. There was a nurse (who we now know to be Sally Turner, the wonderful XP Outreach Nurse) on hand to entertain Eddison, while Hiva delivered the diagnosis. We were advised that Eddison had an extremely rare and incurable condition called xeroderma pigmentosum. The effect being that he has no ability to repair damage caused by exposure to UV light; furthermore, being a complementation group XP-D, he bore a 20-30% chance of developing neurologist complications possibly in his late teens/early adulthood.

To say this was a shock is an understatement. I recall my husband's first question: Is it hereditary? "Yes" came the response. XP is a recessive genetic condition and both my husband and I are carriers. We were advised that we could speak with the team geneticist to discuss matters of future family planning. However, I was already 15 weeks pregnant with our second child. While at the time this seemed like a double shock, and caused increased anxiety during the pregnancy, I often think what a blessing it is. Had we waited just a few months, until after the diagnosis, there is a very good chance that our gorgeous son Raife would not be here. The relationship between our two boys is simply magical and the obvious benefit to Eddison of having a sibling immeasurable. Our second son, though a carrier, does not have the XP gene.

Following Eddison's diagnosis the drive home was terrifying, knowing that every second of exposure was causing irreparable damage. Even once home we knew that every window and many lights were unsafe, so our former haven seemed alien. The first days were spent searching the internet, phoning family and doing a lot of crying. The ensuing weeks were spent in almost complete darkness, curtains closed, lights off and not being able to leave the house. My husband going to work seemed like a big expedition. Our weekly baby groups and visits with friends stopped and the day was spent keeping my then-crawling toddler away from the curtains, where he tried to play peek-a-boo.

At around six weeks things started to improve. Thanks to the kindness of a construction company our neighbours worked for, our house and car were treated with DermaGard film, meaning that the curtains could be reopened. The XP Support Group provided film so we could make Eddison his first visor, and our new best friend the light meter arrived. All of this meant that we had the basic tools to allow us to leave the house in safety. During the interim we were relieved to have regular contact with XP nurse Sally to answer our many questions and guide us through the intricacies of managing Eddison's condition.

At this point, we breathed deeply and decided two things. One: we were not going to accept any tolerance level on behalf of Eddison. We would adapt our entire lives to ensure that he could live a completely UV-protected existence. Two: there would not be anything that he couldn't do; we would find a way to give him and brother Raife all the childhood experiences and opportunities possible.

It is now two years since Eddison's diagnosis and we have found our new 'normal' to settle into. Eddison has continued to thrive and, thanks to measures in place, has had no further episodes of skin reaction since we left





**Eddison and little brother Raife, who is a carrier but does not have XP. The siblings have a "simply magical" relationship.**

the consultant's room. Living a life around avoiding daylight and unsafe artificial lighting is not easy and can be very isolating; every time the front door opens and you step outside you have to be in a heightened state of alert and think through every scenario and potential risk for him. The logistics of never opening a window, ensuring he is in a safe space to simply open the door to a friend or postman, adjusting to the confinement and lack of activity during warm months, and the difficulty of spending time with friends and family in environments outside of your safe bubble all mean that life requires a lot of planning and your every move becomes governed by the weather, seasons and time of day.

Finding new safe ways to have fun is a constant challenge, as is keeping up with appropriate clothing to allow him to enjoy the outside world. Every day brings new issues to overcome with his evolving development and abilities. These include finding creative ways to make a baby wear a full face visor and gloves, making the cream application tolerable, challenges of potty training when out of the home, teaching him the difference between day and night restrictions and adapting a tiny cycle helmet to incorporate a face visor. And all to be done without introducing any element of fear into his perception of these measures…!

Invites from friends become reduced and attending simple family events and everyday activities like shopping are difficult, but we have found that our incredible family and the few good friends who followed us through the process, as well as new ones we have made along the way, have been an immense support. Inevitably the warmer months are the most challenging when outside time is very limited and this year we will have the new dilemma of managing Eddison's restricted time outside versus his brother's natural desire to be in the sunshine.

In a few months Eddison will start nursery, which, after much work with various agencies, will be a local mainstream nursery setting with his peers. Alongside the anxiety of his protective care, we will then be faced with the reality of him having to deal with reactions from others, which to this point he has been blissfully unaware of; too little to pick up on the looks, nudges and comments from people when out and about.

## The Teddington Trust

After discussions with local social services, charities and various agencies it became very clear that XP simply does not fit their prescriptive criteria so, to put it bluntly, with regard to cost and funding to meet his needs we were on our own! Returning to work wasn't an option for me due to the issues of finding appropriate childcare, and so this has had a huge impact on our financial situation and family security.

With the challenge of keeping a roof over our heads we simply did not have the means to provide Eddison with the essential items he needed, and so the Teddington Trust was born. Within months fundraising was in full swing, which allowed us to start purchasing the best UV-protective clothing we could, which has greatly increased Eddison's time outside and the range of activities he enjoys. The support just snowballed and before long our essential list was taken care of — window film for car and home, light meter, creams, vitamins, air conditioning for the home, protective clothing, etc — and we turned our thoughts to achieving the dream of creating a 'safe garden' for him. Incredibly it wasn't long before works started and, with assistance from our local community, in November 2013 Eddison enjoyed his first slide, swing and climb in his safe indoor garden.

Having experienced barriers with regard to funding and support, we hope to build on the great following that the Teddington Trust has had and turn our attention to helping others affected by XP.

We feel sad when we think that Eddison will never walk barefoot through sand on a warm beach, paddle in the sea, run through a grassy field or enjoy the many pleasures of life that we all take for granted. But we have watched him seek so much enjoyment from exploring his world and even at three years old his appreciation of nature is inspiring. The first time, when playing out after dark, it rained, he lifted his head in wonder to enjoy the rain falling and the breeze on his face. Equally, his excitement when he runs his toes and hands through his indoor sand pit, or when he first stroked an animal with bare hands at a special after-dark visit to a farm, are all moments we treasure.

There are going to be many difficulties ahead, and there is always the underlying worry of future neurological complications. Despite these, we hope to encourage him to believe that all is possible and that he just happens to be a boy with xeroderma pigmentosum. XP may be a big part of him, but it doesn't define who he is.

*Read more about the issues of living with XP at www.teddingtontrust.wordpress.com* DN