

BMJ Xeroderma pigmentosum

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BMJ 2008;336:444-446
doi:10.1136/bmj.39485.698356.AD

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A PATIENT'S JOURNEY

Xeroderma pigmentosum

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BMJ 2008;336:444-6
doi:10.1136/bmj.39485.698356.AD

Alex Webb was 4 years old when he was diagnosed with xeroderma pigmentosum nine years ago. This is his story as told by his mother. His parents established a support group that is now widely recommended by consultant dermatologists

My son Alex was diagnosed with xeroderma pigmentosum at the age of 4 years. He was born in Germany and lived there for the first two years of his life. Before the diagnosis, he had had a series of severe sunburns, the first at the age of 7 weeks through a car window, and then the worst when he was 11 months old on a winter holiday in Austria. After this burn Alex was referred to a dermatologist, who dismissed it as "just sunburn" and said, "You mothers just don't know what the sun can do."

Alex is a mixed race child, the second of two children. I had noticed that his cheeks turned red very quickly whenever he was exposed to the sun, so I made sure he always wore a factor 30 sunscreen. Yet he still managed to get burnt, usually in about 15 minutes. His sister did not burn in the same circumstances, so I knew there was something wrong.

I contacted our general practitioner in the United Kingdom, who suggested that Alex might have some kind of sun allergy and advised us to apply sunscreen, and to make sure he was always well covered up and wore a hat whenever possible. With this advice, we managed to avoid any burns until Alex was 3 years old, when he received another very severe sunburn. At this point he was again referred to a dermatologist, who, when I showed him photographs of the previous burns, understood my concerns and started his investigations.

The first series of tests Alex had were to detect which wavelengths of light he was sensitive to. The tests showed that he was most sensitive to ultraviolet A light and then ultraviolet B. Then a series of blood tests showed that he didn't have erythropoietic protoporphyria. As all other avenues had been exhausted, Alex then had a biopsy to test for xeroderma pigmentosum. His condition was diagnosed nine months after his first visit to the dermatologist. During that time we protected Alex from the sun as best as we could.

On diagnosis we really did not know what we could do. No known cure existed and there was no literature. We were told that unless Alex was kept indoors he was unlikely to reach the age of 30. We mourned the loss of our son's future and then said,

"That's it. We are going to be more positive and find ways to allow Alex to live as near a normal life as possible."

Our search to help Alex

Our journey first led us to the internet, where we learnt more about the condition and found an online forum where we could ask as many questions as we liked. Some questions were answered by other people with the condition, but some, as in our case, were answered by professionals. Our question was, "We have done all the reading and seen the photos, but our Alex does not look like a typical case, so could the diagnosis be wrong?" Dr Ken Kraemer, a research clinician at the National Institutes of Health in the United States, asked who had diagnosed the condition. When we told him it had been Professor Alan Lehmann at Sussex University, he replied, "Your son has XP [xeroderma pigmentosum]."

Our next stop was social services, where we were allocated a sympathetic social worker who was keen to learn more about the condition and to work with us to achieve something for Alex. First of all, she arranged for our home and car to be fitted with special window films that block ultraviolet light; this was financed by a special needs budget in the children's services of our local social services department. As Alex was almost of school age, she then arranged a meeting for us at Amersham Hospital with the local education authority, an educational psychologist, and the head teachers of the two schools that Alex would attend up to the age of 11. The meeting went well, and our request for Alex to be included in mainstream school was accepted.

The next steps were to get Alex's new schools protected from ultraviolet light and to arrange for a welfare assistant to be on hand to help with his application of sun cream, which is repeated several times throughout the school day. Although Alex was unable to start in the term of his fifth birthday, which was the norm, he was able to start the following term.

Living with xeroderma pigmentosum

At this stage our journey was becoming smoother. My husband and I started a support group for families with xeroderma pigmentosum which helped us to put living with the condition into perspective. We met families who were coping very well and others who weren't, and by pooling our information and resources we soon became a small active group.

This is one of a series of occasional articles by patients about their experience of traumatic medical events that offer lessons to doctors. The *BMJ* welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance.

In forming the group, we were particularly helped by our dermatologist, Dr Sheru George at Amersham Hospital, Professor Lehmann, and the support team from the UK charity Contact a Family (which helps the parents of disabled children, www.cafamily.org.uk). Dr George believed that our group would help not only patients with xeroderma pigmentosum but the many more patients with other ultraviolet and light sensitive conditions. We have since developed relationships with many organisations, such as the Skin Care Campaign and the Genetics Interest Group. We will shortly be embarking on a training session with the government's Department for Work and Pensions to help our members

complete the claim form for the disability living allowance.

One of our more unusual collaborations has been with the European Space Agency, which through its technology transfer programme has been working on producing protective clothing for children with xeroderma pigmentosum. The progress has been very slow, but we hope that the first prototypes will appear soon.

Now, nine years after Alex's diagnosis, we feel in control because we have access to the information we need. Alex's routine includes the application of sun cream every two hours during daylight hours; he still has a welfare assistant at school who ensures that he

A DOCTOR'S PERSPECTIVE

Xeroderma pigmentosum is a rare autosomal recessive genodermatosis with a worldwide incidence of 1:250 000 live births. Patients have a genetic inability to repair DNA damage that has been induced by ultraviolet light. This manifests clinically as photosensitivity and an incidence of skin cancer that is 1000 times higher than the average. Xeroderma pigmentosum usually presents at age 1-2 years with photosensitivity and burning after minimal sun exposure. Later cutaneous manifestations include increasing dryness of skin, freckling, and telangiectasia. There is an increased incidence of skin cancer on sun exposed sites. The median age of onset of skin cancer is 8 years. Ocular abnormalities include photophobia, ectropion, conjunctival injection, keratitis, and tumours (which usually occur on the areas of the eye that are exposed to ultraviolet light, such as the conjunctiva, cornea, and eyelids). A fifth of patients have associated neurological abnormalities such as gait disturbance, areflexia, difficulty swallowing, deafness, growth delay, and low intelligence.

Genetically, xeroderma pigmentosum can be subdivided into seven complementation groups (XPA-XPG) and XP Variant (XPV). The diagnosis is established by skin biopsy for fibroblast culture to measure unscheduled DNA synthesis. Prenatal diagnosis is available.

Treatment includes protection from ultraviolet light (avoidance, protective clothing, protective glasses, face shield, and sunscreens with a high SPF (sun protection factor)). Due to the high incidence of skin cancer linked with xeroderma pigmentosum, patients need skin cancer surveillance every three months by a consultant dermatologist, an annual ophthalmological review, and, if they have an associated neurological disorder, an assessment every six months by a consultant neurologist.

Even though Alex's parents had known since he was 7 weeks old that his photosensitivity was abnormal, their son's sensitivity had been dismissed as normal by various clinicians over several years. Delay in diagnosis in xeroderma pigmentosum is not uncommon. Although the condition is rare, an accurate early diagnosis based on clinical symptoms, diagnostic tests, and DNA analysis is essential. Early diagnosis may prevent complications associated with unprotected exposure to sunlight and enables genetic counselling and prenatal diagnosis to be

provided to families at risk. Babies and children with photosensitivity should always be referred to a consultant dermatologist for assessment. Childhood skin cancer is rare, may be a presenting feature of xeroderma pigmentosum, and should always be investigated. Patients with xeroderma pigmentosum who have associated neurological abnormalities may initially present to neurologists.

Alex has normal skin, with no lentigines or skin cancers, which is a tribute to the excellent care and protection from ultraviolet light that he has received. He is neurologically normal. Even though Alex has expert parents, he must continue to have regular dermatological and ophthalmological follow-up.

I originally set up a specialist clinic for patients with xeroderma pigmentosum at Worthing Hospital. The clinic was subsequently moved to the St John's Institute of Dermatology at St Thomas's Hospital in London with the help of Dr Robert Sarkany. It operates with a multidisciplinary team. Patients have the opportunity to be seen by consultant dermatologists, ophthalmologists, and neurologists with a special interest in the disorder in a single appointment. Sandra Webb is an integral part of our team, providing invaluable support and information to the patients. She also provides the team with a useful insight into the disorder and how it affects day to day life from a patient's and parent's perspective. Via the XP Support Group, Sandra provides us with regular feedback from patients about the clinic and how it can be improved for patients.

As clinicians, we must not feel undermined by patients or parents who have built up a knowledge base for their condition. We can learn from them as they can from us. As this case illustrates, we can work together to build on our expertise and improve the quality of care and awareness of rare diseases.

Useful resource: Kraemer KH, Lee MM, Scotto J. Xeroderma pigmentosum cutaneous, ocular and neurologic abnormalities in 830 published cases. *Arch Dermatol* 1987;123:241-50.

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RESOURCES

XP Support Group

The XP Support Group (www.xpsupportgroup.org.uk) is a British charitable trust, founded in 1999. Its objectives are to relieve the needs of people with xeroderma pigmentosum (and other related conditions) and their families. The group raises funds for research; provides grants for ultraviolet protective equipment and products; and helps families to attend the night-time conference Camp Sundown (run by the Xeroderma Pigmentosum Society) or take respite in a protective environment.

XP Family Support Group

The XP Family Support Group (www.xpfamilysupport.org), based in Sacramento, California, raises money to promote awareness and educate the public about the disorder, to fund research, to give grants for ultraviolet protective equipment and products, and to help families attend annual xeroderma pigmentosum conventions.

Xeroderma Pigmentosum Society

The Xeroderma Pigmentosum Society (www.xps.org) is based in Craryville, New York. Its objectives are to increase public awareness about xeroderma pigmentosum and related conditions; to provide protection, through support and information exchange, to victims of this disease and their families; and to promote medical research to achieve a cure for the disorder.

Skin Care Campaign

The Skin Care Campaign (www.skincarecampaign.org) is an umbrella organisation representing the interests of all people with skin diseases in the UK. It works for the improvement of health care for people with skin disease; educates and informs the public and others about skin diseases and their treatment; and supports other organisations to pursue these objectives.

Genetic Interest Group

The Genetic Interest Group (www.gig.org.uk) is a UK based national alliance of patient organisations with a membership of over 130 charities that support children, families, and individuals affected by genetic disorders.

puts it on. He wears long sleeved clothes and long trousers all year round. The only restriction on his going out is that he needs to wear a face mask and gloves. He chooses when and if he wants to go out. Before Alex started his secondary school, I was asked

by his headmaster to speak to all the year groups and teaching staff about xeroderma pigmentosum. This has ensured that the children have a good understanding of why Alex has the routine he has and means that the children and staff look out for him. He is now in year 9 and very happy. After a recent visit to the National Institutes of Health in Bethesda, we have made minor changes to his care: he now takes a regular vitamin D supplement and we have also managed to persuade him to apply moisturiser to his arms and legs regularly.

We still feel, as we did nine years ago, that early diagnosis is the key to managing this condition. The children we know who have been wearing the protective clothing and taking the measures that we advocate have not had any skin cancers. Our dermatologist now expects Alex to have a normal life expectancy as long as the protection continues. Awareness of the condition has improved, and children with xeroderma pigmentosum are not stared at as much as they used to be. But the most important part of our journey has been to get the professionals to accept that parents know more about living with xeroderma pigmentosum than they do, and that we can help them to help other families when they are diagnosed. I was recently greatly honoured to be invited to address a conference of 120 researchers and clinicians from around the world.

That the medical profession is taking more interest in xeroderma pigmentosum is evidenced by the fact that a multidisciplinary clinic for people with the condition, led by consultant dermatologist Dr Robert Sarkany, has recently been established at the St John's Institute at St Thomas's Hospital in London.

Competing interests: None declared.

Provenance and peer review: Not commissioned; externally peer reviewed.

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