ANNUAL REPORT 1999-2000

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WHAT IS XERODERMA PIGMENTOSUM

Xeroderma Pigmentosum (XP) is a rare genetic disorder that causes extreme sensitivity to the sun's ultraviolet rays. Unless patients with XP are protected from sunlight, their skin and eyes may be severely damaged. This damage may lead to cancers of the skin and eye. XP has been identified in people of every genetic group all over the world. There are about 100 cases in the UK, many of which are undiagnosed.

Causes

Ultraviolet light damages the DNA in cells and disrupts normal cell functioning. DNA (deoxyribonucleic acid) within in our genes contains all coded information needed to direct cell functions.

Two factors combine to cause the abnormalities in XP. First, a person inherits traits from each parent which, when combined, lead to an unusual sensitivity to damaging effects of ultraviolet light. Second, exposure to the sun, which contains ultraviolet light, leads to changes in the skin and eyes.

Damaged DNA is mended by the DNA repair system. But the DNA repair systems of people with XP do not function properly. As a result, un-repaired DNA damage builds and causes cancerous cell changes or cell death.

Symptoms

Many persons with XP get an unusually severe sunburn after a short sun exposure. The sunburn will last much longer than expected, perhaps for several weeks. This type of sunburn will usually occur during a child's first exposure, and it may be a clue to the diagnosis of XP. However, some people with XP will not get a sunburn more easily than others, and the disease will be undetected until unusual skin changes appear over time.

Most patients with XP develop freckles at an early age. Continued sun exposure will lead to further changes in the skin, including irregular dark spots, thin skin, excessive dryness, rough-surfaced growths and skin cancers. These skin changes will resemble those of elderly people who have spent many years in the sun. In people with XP, these changes caused by sun damage often begin in infancy and almost always before the age of 20.

The eyes of a person with XP are often painfully sensitive to the sun and may easily become irritated, bloodshot and clouded. Non cancerous and cancerous growths on the eyes may occur.

Treatment

There is no cure for XP, but much can be done to prevent and treat some of the problems it causes:

- Protection from ultraviolet light, by a combination of physical and chemical means. These include sun avoidance, shade, clothing, hats, optical filtration and sunscreens.
- Frequent skin and eye examinations.
- Prompt removal of cancerous tissue.
- Neurological examination in some cases

Text extracted from "Understanding Xeroderma Pigmentosum" published by US Department of Health & Human Services

AIMS OF THE SUPPORT GROUP

- To relieve the need of persons with Xeroderma Pigmentosum (XP) and UV-related conditions and their families;

- To advance the education of the public in Xeroderma Pigmentosum;

- To promote research into Xeroderma Pigmentosum.

OBJECTIVES OF THE GROUP

- To raise funds for the XP Research fund set up by the XP Society;

- To assist families to attend Camp Sundown (a night-time camp held once a year in New York State, USA) or respite in a protected environment;

- To give grants for UV protective products;

- To raise public awareness by means of an educational campaign.

ACHIEVEMENTS DURING THE YEAR

In May 1999 we were awarded charitable status. Registered number 1075302.

We are now affiliated to Contact-a-Family, the Genetics Interest Group, The Skin Care Campaign and Climb (Children living with inherited metabolic diseases).

We have seen membership of the Group grow from 36 to 220. Members consist of patients and their families, the medical professions, other professions who work with XP patients such as social services and education, volunteer helpers and those who have given donations.

We acted as advisors on two BBC programmes – Casualty (shown 26 February) and Horizon –Moon Children (shown 4 April). We were also due to be the subject of an ITV documentary for Real Lives, sadly filming was stopped and the programme will not be shown. Channel 4 News in January also did a report about the clinical trials of the enzyme cream TN 4-5 which finished this year, results are expected in 2001. Sandra Webb also appeared on BBC Breakfast News.

We have received a lot of Press coverage, which has helped to raise profile of this condition which affects so few people.

We now have a poster ready for printing which will be distributed to Dermatology units throughout the country.

Sufficient funds have been raised for us to book 3 families to attend Camp Sundown in July 2000.

We held 2 meetings a social evening in November (Fireworks and Barbecue) and a meeting where our speaker discussed patient’s right. We plan to hold the fireworks and barbecue as our annual social evening.

A visit was organised to the Cell Mutation Unit at Sussex University, by Professor Alan Lehmann. This gave families the opportunity to see the methods that are used to diagnose XP.

For the first time ever a family was informed when their child was diagnosed that a Support Group exists, this we feel is our biggest achievement.

We take this opportunity to thank Dr Celia Moss, Dr Sheru George, Professor Brian Diffey and Professor Alan Lehmann for the help they have given us to date.

AIMS FOR 2000 -2001

To complete A Patients Handbook for distribution to Dermatological Units and patients.

To send at least one family to Camp Sundown 2001

To start fundraising for Camp Sundown UK to be run in February 2002

To appoint an official medical advisor – we currently have an informal arrangement.

To increase the number of people actively involved in running the Support Group.

To open our web site.

Finally we hope to secure a patron, we have approached four people so far without success. Any ideas for a suitable person should be passed the Secretary at the XP Support group Office.

HOW YOU CAN HELP

DONATIONS	All Donations gratefully received
FUNDRAISING	Organise an event, such as a sponsored event, coffee morning
VOLUNTEERS	We currently need an Editor for the Newsletter and a fundraising Co-ordinator. We also need volunteers to help with meeting especially The Creche. We are also looking for Volunteers for Camp Sundown UK 2002.
SPREAD THE WORD	Show this report to friends and colleagues

TRUSTEES

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