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# XP SUPPORT GROUP NEWSLETTER

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Issue 21

October 2006

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## XP Support Group in Washington DC

The XP Support Group was represented at the recent Workshop on XP and other diseases of Human premature aging and DNA repair. Sandra Webb gave a presentation to clinicians and researchers on "A Day in the Life of Alex". She was also part of a panel along with Michele Milota of the XP Family Support Group, Laura Niederhofer of the XP Society and Jackie Clark from CS Network e that answered questions on living and coping with XP. A report of the conference is given on page 3. Sandra and Michele also posed the questions that XP patients and families had supplied in advance of the Workshop. The answers are being collated and will be in the next issue of the Newsletter.

## Annual Bonfire & BBQ

Our annual social event will take place in Alex's Night-time Garden, 2 Strawberry Close, Prestwood on Saturday 4th November from 16.30. This event is for all members and volunteers.

This will be our 7th event which will be a barbecue followed by a short walk to a local firework display. There will be plenty of opportunity to meet and talk to others. To help us with our catering arrangements, please return the form on page 4 or let us know if you plan to attend by **Monday 30 October**.

For those travelling to Prestwood, please note that we can make a contribution to your travel and accommodation expenses. If you need overnight accommodation, please contact Sandra Webb, at XP UK Headquarters.

## Owl Patrol 2007

The next Owl Patrol will take place from **Friday 9 to Monday 12 February 2006**.

Preparations are already on the way and volunteers will be going through training in November and January to make this camp extra special for you all.

The application form for camp is on page 4 of this Newsletter. Camp is free to XP patients and one accompanying adult if under 16. For other family members we ask for a donation of £10 per person. Places are limited so we ask you to return your forms as soon as possible. For those of you planning to travel from abroad, please contact Sandra Webb at XP UK Headquarters as soon as possible.

## Jeans for Genes

Friday 6<sup>th</sup> October was Jeans for Genes day, a national campaign that raises money for several charities supporting those with genetic conditions. This year our Group is one of the benefiting charities. The XP Support group managed to enlist the support of Ian Wright, recently voted "Rear of the Year". Ian is the relative of a child with XP.

One of the projects that will be supported by Jeans for Genes is the training of two peer counsellors and the preparation and production of a Patient Support and Schools Information Pack. If you are interested in becoming a peer counsellor or feel you would be able to contribute to the production of the two packs, please contact Sandra Webb at XP Headquarters as soon as possible.



Ian Wright sporting his Jeans for Genes T-Shirt

# Fundraising

## Donations

We would like to express our thanks to the following for their recent donations:

We reported in the last Newsletter about **The Princes Risborough School XP Youth Enterprise Group** who chose the XP Support Group to be the beneficiary of any profits received from the company they set as part of the Youth Enterprise Scheme. In addition to the **£1055** already donated they have raised a further **£50**.

Congratulations and special thanks to **Kate and John Middleton** who asked their guests at their recent wedding to donate to the Support Group rather than gifts. We were delighted to receive **£247**

**Prestwood Women's Fellowship** for their donation of **£25**.

**Moog Controls** for their donation of **£200**

**The Thatched Cottage Pub** of Maidenhead for their donation of **£500**.

## Hydro Active Run – 3 September

This year saw a record entry of 32 ladies representing the XP Support group at the Hydro Active run. So far **£490** has been collected with still more to come.



A few of the XP Support Team at Hyde Park

## 100 Club

Please find below the draws for April to September 2006. Our numbers have dropped dramatically so we are only able to give a prize of £35. We shall continue the draws until March 2007. If our numbers have not increased significantly we will have to review the feasibility of running the club

April:	Dirk Prenting, Germany
May:	Val Corteen, Prestwood
June:	Steve Webb, Prestwood
July:	Sue Sexton, Maidenhead
August:	Diana Brown, Prestwood
September:	Susan Lue, London

Please help us get back to 100. If you would like be part of the 100 Club, please contact Sandra Webb at XP UK Headquarters

## Yellow Moon Catalogues

Copies of the Christmas Yellow Moon catalogue are now available from XP headquarters, please contact Sandra Webb if you would like one. You can also order direct on line at [www.yellowmoon.org.uk](http://www.yellowmoon.org.uk) quoting ref SXP10001. Up to 25% is donated back to the Support group on all orders placed.

## Football Prediction Competition

The pool for this years competition was **£94**. The winners are:

1<sup>st</sup> Prize £28- Adam Markham

2<sup>nd</sup> Prize £14 – Nick Spray

3<sup>rd</sup> Prize £5 – John Larkin

£47 to the XP Support Group

## Upcoming events

**BBQ and Fireworks – 4 November 2006**

Venue : Alex's Night-time Garden, Prestwood, Bucks

**XP Friends Weekend – 5/6 November**

Venue : Hotel Jagd Glashutte, Volkhholz, Germany

**Family Retreat Weekend – 10-13 November 2006**

Venue : Disneyland, Los Angeles, USA.

**Walk by the Light of the Moon – 11 November 2006**

Venue : Southern California

**Volunteer Training – 25/26 November 2006**

Venue : Green Park Conference Centre, Aston Clinton

**Volunteer Training – 13 January 2007**

Venue : St Katharine's, Parmoor

**Owl Patrol – 9-12 February 2007**

Venue : St Katherine's, Parmoor

**Volunteer De-Brief – 4 March 2007**

Venue: Prestwood, Bucks

**Owl Patrol for Teenagers – 20-22 July 2007**

Venue : St Katharine's, Parmoor

**Auction of Promises – 15 September 2007**

Venue : St Katherine's, Parmoor

For further information about any of these events. Please contact Sandra Webb at XP Headquarters

## XP New Information Leaflet

With this Newsletter, you will have received a copy of the new XP information leaflet which was sponsored by Awards for All. We think you will all agree that it gives our Support Group a more professional image. Further copies can be obtained from XP Headquarters

# XP Workshop Report

By Mimi Sander, Page One Editorial Services,  
Boulder, Colorado

## Clinicians, Researchers and Patient Support Groups Meet to Discuss Xeroderma Pigmentosum and Other Diseases of Human Premature Aging and DNA Repair

Representatives from XP Support Groups in the US and the UK (Sandra Webb, Michele Milota and Tammy Zinkann) and the CS Network (Jackie Clark) attended a recent workshop on XP, CS and related diseases at the National Conference Centre in Lansdowne, Virginia (September 5-8, 2006). Approximately 100 leading researchers and clinicians also attended this workshop, which was the second in a series of National Institutes of Health workshops organized by Dr. Vilhelm Bohr (National Institute of Aging), and Dr. Kenneth Kraemer (National Cancer Institute). The discussions at the workshop revealed areas in which great progress has been made, areas ripe for future study, and bottlenecks that are inhibiting progress in either basic understanding of the diseases or their clinical management. In the spirit of a true workshop, the participants engaged in extensive discussion and interaction during question/answer periods after each seminar as well as during five panel discussion sessions. Four of the workshop sessions provided the opportunity for patients, family members and/or family support group representatives to describe their personal experiences with XP or CS. Throughout the workshop, researchers and clinicians were eager to acknowledge their gratitude towards patients and patient family members, who play a key role in allowing the research on these diseases to move forward.

*Some of the key findings presented at the workshop were: 1) There is great need for improved understanding and better management of the neurological aspects of XP, CS and TTD. 2) For some XP patient subgroups, the severity of the disease correlates well with the severity of molecular defects in XP-related proteins or mRNA transcripts. 3) Eye defects in XP patients are related to sun exposure and premature aging, while eye defects in TTD, XP/TTD and CS patients reflect developmental defects. 4) XP patients exhibit cerebral and cerebella atrophy; in contrast, TTD patients have a defect in myelination of the white matter of the cerebrum.*

The patient support group representatives who attended the workshop made numerous contributions to the formal and informal workshop sessions. During the opening workshop session, Sandra Webb, Jackie Clark, Dr. Laura Niedernhofer (on behalf of the XP Society) and Dr. John DiGiovanna (on behalf of the Foundation for Ichthyosis and Related Skin Types; FIRST) presented basic information about each of their respective organizations. During a later workshop session, Sandra Webb gave a slide presentation describing one day in the life of her son, Alex, who has XP. Dr. and Mrs. Loren and Betty Smith also presented a slide show of their

daughter, Jill, a 41 year old XPA patient with progressive neurological disease, and discussed their experience raising her. Jill accompanied the Smiths to this session and interacted with workshop participants. Jackie Clark also showed a DVD presentation of the life of her daughter, who died of CS at the age of 5. All the patient support group representatives participated in a panel discussion session on the last day of the workshop, during which many of their concerns for the future were discussed and addressed. They indicated their continued desire to raise money to support research, interest in helping to establish patient registries, the need for more comfortable protective wear for XP patients, and interest in better understanding and management of the neurological aspects of XP, TTD and CS.

Other discussion points of particular interest to family support groups included the following: while both cover-up and sunscreen use is usually recommended for patients with sun sensitivity syndromes, the appropriate level of protection must be decided on an individual basis. Recent studies by Dr. Thomas Ruenger (Boston University) also suggest that there may be some risk of skin damage from exposure to UVA light, which passes through glass. Thus, the value of shielding from UVA light should also be reconsidered. Dr. Dan Yarosh (AGI Dermatics-New York) described promising clinical studies of a topical agent called Dimericine, whose active ingredient is a bacterial DNA repair enzyme that stimulates repair of UV-induced skin damage. Dimericine reduced the incidence of actinic keratoses on the skin of treated XP patients significantly more than a placebo control. However, because Dimericine is not yet FDA-approved, it is not currently possible to make it available to XP and other sun-sensitive patients, despite requests from patients who used the drug during its clinical trial.

Diagnostic testing for XP, CS and TTD was discussed during one of the workshop panel discussion sessions. Diagnostic testing based on functional assays but not on DNA genotyping is currently available in the Netherlands, Germany, Italy, Japan, Israel, and the UK. These tests are used to confirm clinical findings and for prenatal diagnosis of at-risk pregnancies. No CLIA-certified laboratory currently performs these tests in the US. However, there is strong consensus among researchers and clinicians that CLIA-certified testing for these diseases is urgently needed in the US. One laboratory in the US currently offers DNA diagnostic testing for known mutations in CS families.

The workshop entitled *Xeroderma Pigmentosum and other diseases of human premature aging and DNA Repair: Molecules to Patients* was co-sponsored by the National Institutes of Health Office of Rare Diseases, National Institute of Aging, National Institute of Environmental Health Sciences and the National Cancer Institute.

**BBQ & Fireworks – 5 November 2005**

We will/will not\* be attending the BBQ

We will be \_\_\_\_\_Adults \_\_\_\_\_ Children

Name\_\_\_\_\_ Tel No:\_\_\_\_\_

Transport required Yes/No\* If yes name of nearest train station\_\_\_\_\_

Accommodation required Yes/No\*

*\*Please delete as appropriate*

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**Owl Patrol 9-12 February 2007**

Name\_\_\_\_\_

Address\_\_\_\_\_

\_\_\_\_\_ Tel No \_\_\_\_\_

**Other family members who will attend** (please provide names and ages)

While there is no charge for patient campers and accompanying adult for patients under 16, we do ask for a contribution of £10 for each additional person.

Cheques should be made payable to the XP Support Group

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