



**THE ASSOCIATION
OF GENETIC SUPPORT OF
AUSTRALASIA INC.**

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NEWSLETTER

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MISSION STATEMENT

To facilitate support for those affected directly or indirectly by genetic conditions throughout Australasia.

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EDITORIAL

This is the April newsletter running very late so we have combined April and June. We apologize for keeping everyone waiting.

It has been an exciting time for AGSA. I was invited to attend the World Life Sciences Forum-BioVision in Lyon, France in 8 - 11 April 2003 along with sixteen representatives of genetic patient organizations from 6 continents. We met numerous times during the conference and founded the International Genetic Alliance (IGA). Over one thousand people attended the conference including 129 NGO's from around the world. Among the guest speakers was James Watson along with 8 Nobel Laureates plus other dignitaries from around the world. (*To learn more read my report inside.*)

As I was out of the office for 3 weeks incorporating the Easter period and there is no funding for full time staff, the work piled up. Anne Mount, Administration Officer works Wednesday 10 - 4 -6 hours and Sue Hawkins, Project Assistant, Thursday and Friday 9.30 - 3.30 - 12 hours. I work

28 hours per week. Forty-six hours per week equates to one full time person plus a very part time person per week. A lot of time is given supporting callers ringing after the diagnosis of a genetic condition. This may take up to one hour to deal with their support needs and to provide practical information follow-up by organising fact sheets and a family match.

AGSA's database with over 550 rare genetic conditions constantly needs updating and the general administrative work is continuous.

This year we have applied for funding to run sibshops and telegroup counselling.

Our office is small and is situated in The Old Children's Court in Surry Hills which is an old building owned by DOCS and managed by New South Wales Council Of Social Services (NCOSS). The building is shared with other community groups Network, Local Community Services, The Australian Association of Social Workers, Women's Electoral Lobby and of course NCOSS. There are two meeting rooms, a shared photocopier and other equipment available for hire at a reasonable rate from NCOSS. The rent is reasonable and we are close to Central Station.

AGSA has been funded by the NSW Health Department since 1994 and we currently receive \$63,100. This is why your membership subscription is so important to us because it provides funding for genetic seminars, Genetic Disorders Awareness Week and research into information on genetic conditions.

AGSA is particularly concerned about the plight of support groups whose leaders (often the founder) are deciding it is time (after 20 years or so), to step down only to find there is no one to take their place. The information and technology available today makes running a support group hard work and

very time consuming. Often at the expense of the family.

A few years ago AGSA tried to find suitable premises which could be a joint head office/resource/contact centre for support groups. These groups would have a permanent mailing address, access to computers, photocopiers, laptops etc. AGSA applied for an office at Prince Henry Hospital. As Premier Carr now has major plans to develop this site we were unsuccessful. It is AGSA's goal to have a formal meeting with both the State and Federal minister for health outlining these problems. Without support groups (750 Australia wide) services would be swamped. It is important something is put in place and the government made aware of the valuable work volunteers do. Where would the Olympic Games have been without the volunteers?

Australasian Genetic Alliance (AGA)

On 9 May 2003 AGSA organised a meeting with Trish Fallon Self Help Qld, Amanda Foudoulis SHOUT Act, Eilis Hughes Genetic Support Network Victoria, Terry Keating Genetic Support Council of WA and John Forman of NZORD to discuss the feasibility of setting up a national genetic support alliance. The meeting was held at the Novotel Hotel Darling Harbour and was very kindly sponsored by Genzyme Australia Limited. It was especially great to put faces to names. The day was very successful and a lot was achieved. A Vision, mission statement, aims and objectives were agreed upon. It is now up to the respective committees to submit their reports. The alliance will be a united national voice for genetic support groups in the Australasian region.

This month I was also asked to present at the Congenital Malformation and

Birth Defect National Data Review Workshop on 12-13 May 2003. After many interesting presentations and discussions it was agreed there should be a National Birth Defect Register. AGSA believes it is very important to have a national register, which will then report to an international register. The advantages are enormous; not only will it indicate environmental trends, provide valuable information for requirements and planning of health services, assist in research and more importantly, improve the health of the children in the future.

Until next time, Best wishes from
Dianne Petrie



“Making the right connections since 1988”

THE WORLD LIFE SCIENCE FORUM-BIOVISION and the Third Meeting of the **International Genetic Alliance** Lyon, France 7-11 April 2003

The International Genetic Alliance is a global joint venture of regional and continental alliances of parent and patient organizations with an interest/involvement in the opportunities and implications of genetics and biotechnology. Founded Sixteen representatives of genetic patient organizations from 6 continents met in Lyon, France from April 7-11 for the third World Life Sciences Forum - BioVision and founded **the International Genetic Alliance (IGA)**.

Pat Terry, Co-founder and President of PXE International and member of Genetic Alliance (USA), was elected the first president of IGA. Other founding members include Ysbrand Poortman VSOP of the Netherlands, Alastair Kent UK Genetic Interest Group. Other countries signing on as founding members and in attendance at the Lyon

meeting were, Australia, Brazil, Germany, India, Iran, New Zealand South Africa, and Ukraine.

International Genetic Alliance

Vision

The International Genetic Alliance seeks a world where genetic conditions are understood, prevented, treated, ameliorated, and cured.

Mission: To promote medical genetic services, research, technologies, and access to information, in order to alleviate the burden of genetic conditions for individuals, families and communities.

Aims and Objectives

Support and accelerate research to prevent, treat, ameliorate and cure genetic conditions, through the translation of biotechnologies to accessible services.

Develop policy in partnership with policy makers.

Engage in partnerships with clinicians, researchers, industry, and NGOs with a shared mission.

Establish best practice in medical genetic services that respect the autonomy of the individual, family, and community within a cultural context.

Ensure access to relevant information, services, technologies, and medical treatments

Members of the IGA plus members of the World Alliance of Prevention of Birth Defects are: Southern Africa Inherited Disorders Association (SAIDA), Ukrainian Alliance for the Prevention of Birth Defects, March of Dimes from USA, Women's Health, Germany, European Platform for Patients' Organisations, Science and Industries, Indian Genetic Alliance of Patients Organisations

Other IGA members:

The Association of Genetic Support of Australasia (AGSA) Inc.
Dutch Genetic Alliance of Parent/Patient Organisations (VSOP)
HEART EU, Patient Network, Spain
National Organisation of Rare Diseases New Zealand (NZORD) S
South American Genetic Alliance of Patient Organisation
For more information:
<http://www.geneticalliance.org/partnerships/IGA.html> for more information and to review IGA's Guiding Principles, Goals and Objectives and Founding Members

What is BioVision?

As the analysis of recent decades clearly illustrates, Science and Technology are making exponential progress in all fields. This is particularly true of two areas:

Information and Life Sciences

(Industry, Environment and Society). recognised as the keys to economic development and substantial changes in our lifestyle throughout the XXI Century. Health-related and food-processing industries, like all sectors with an environmental component, are finding themselves increasingly influenced, and indeed transformed, by the progress made in Life Sciences. There is a need for objective reflections on progress and clear prioritisation wherever technical advances impinge on people's lives. Nowhere is this need more urgent than in the Life Sciences where the blistering pace of technical change and industrial development are outrunning Society's ability to embrace them

This is why a work group, set up on the initiative of former Prime Minister Raymond Barre (France) and the Academy of Sciences, had decided to create this essential World Forum designed to chart - and regularly discuss - the progress of Life Sciences and its techniques faced with the challenges of the future, without evading the associated risks.

The extensive participation of experts and leaders from Science, Industry, International Institutions, Policy and Regulatory Bodies, together with representatives of Non-Governmental Organisations, the Media, and other gatekeepers of Public Opinion maintain the reputation of BioVision for detachment and balance, and facilitate constructive exchanges between Opinion-Leaders and Decision-Makers from all backgrounds.

BIOVISION: A PLATFORM FOR CONSTRUCTIVE EXCHANGE MEETING

The World Life Sciences Forum is an unprecedented event that enables all stakeholders to address a wide range of challenging issues surrounding Life Sciences (Industry, Environment, Society).. It offers a unique and ideal opportunity for all to exchange views, therefore contributing to the understanding, acceptance and development of Life Sciences.

BioVision establishes itself as a platform for dialogue.

Key Orientations

Facilitate debate on major ethical problems set by the development of Life Sciences at an international level.

Coordinate campaigns of communication for the general public, open up debate, particularly with NGOs.

Make the younger generation aware of Life Sciences potential and train tomorrow's experts within a strict ethical framework.

Encourage the alternative choices that new technologies and their by-products represent by integrating their economic, social, ethical and ecological dimensions.

Encourage technology transfer (health, agronomy, food, environment) to developing countries, along with training programmes.

Encourage the development of technologies resulting from academic research by creating and developing start-ups.

REPORT FROM WORLD LIFE SCIENCE FORUM BIOVISION INTERACTIVE SESSION

LifeSciences:

BioVision calls for more solidarity from G8 countries

Lyon, April 11, 2003 - At the close of its third edition, the World Life Sciences Forum, BioVision, drew up a series of recommendations destined for international organisations and governments, especially G8 countries. The recommendations stem from the discussions conducted during the forum on four topics:

- Improving the Beginning of Human Life
- Life Expectancy and Longevity
- Securing Enough Safe Food
- Life Sciences, Environment and Industrial Revolution

Amongst the key points of its recommendations, BioVision calls for international mobilisation in favour of a health policy more radically oriented to prevention

BioVision urges the developed world, in particular G8 members, to immediately take steps to reverse the decline in contributions to international aid.

BioVision draws the attention of international institutions, public organisations, and the private sector, on the urgent need to combat maternal and foetal malnutrition, leading to low-birth-weight children in developing countries and fighting cancer and obesity in the Western World.

BioVision asks World Health Organisation to launch a priority programme on questions related to ageing.

In addition, BioVision recommends discussions on key ethical questions raised by Life Sciences and their applications be widened. The discussions must be supported by reliable scientific data and continuous comparative analyses of public opinion.

The conclusions of the experts are available, in their entirety, on

BioVision's website: www.biovision.org



Support Group News

PLEASE NOTIFY AGSA IF YOU WOULD LIKE YOUR SUPPORT GROUP WEBSITE LINKED TO AGSA'S WEBSITE.

"The Book of John"

The story of a remarkable young man who lived life with joy!_

This book is written in memory of Coral and Guido Rizzalli's son, John. John was affected by a rare genetic condition called **Ring 18 Syndrome**. He was born in 1971 when there were no services available in his local area for disabled children and adults. His tenacity and love of what he called "Real Life" was inspirational. A reminder of the value of life, the joy, the hope and the power to change. A book of love.

All proceeds from "The Book of John" will go to Ingham Disability Services, Queensland. This book is available for \$22.00 each plus \$3.00 postage and handling within Australia. To purchase a copy, please contact:

Coral Rizzalli, P O Box 810, Ingham QLD 4850 or ph: (07) 4776 1583

email: coralrizz@ozemail.com.au

PRESS RELEASE · URGENT · FOR IMMEDIATE DISSEMINATION

Washington DC, May 9, 2003 – On April 21, 2003 the Genetic Alliance launched the first stage of Disease InfoSearch - an innovative public information search tool for genetic disease information that can be viewed at

<http://www.geneticalliance.org/DIS/>.

The Genetic Alliance developed this Internet-based search tool to facilitate public access to quality lay-oriented information about genetic and rare diseases in language that is understandable and useful.

Disease InfoSearch (DIS) is a robust catalogue of genetic conditions - rare to common - that is searchable according to Clinical Description (signs & symptoms), Treatment Research, Genetic Information (molecular), Support Groups & References (newsletters, listservs, self-help books), Insurance Issues, Arts & Literature and Other Resources. At this time, Disease InfoSearch has been populated by more than 60 genetic conditions, including Tuberous Sclerosis, Tay-Sachs disease, Lowe syndrome, Breast Cancer, Gaucher disease, Fabry disease, Mannosidosis, Progeria and others. It is now ready for expanded population by the 600 member organizations within the Genetic Alliance coalition. This dynamic and up-to-date public information system is driven and maintained through the expertise and vigilance of disease-specific lay advocacy organizations and their professional advisory boards. Editorial review and quality control are provided by genetic counsellors that staff the Genetics Education and Resource Centre.

With increasing frequencies, the patient advocacy community is the place where the public goes to find useful and credible information about specific genetic diseases in lay language. Capitalizing on the enhanced expertise

of disease specific lay organizations, sixteen years experience responding individually to Helpline caller inquiries and new technologies that allow a high tech/ high touch approach, Disease InfoSearch tool.

The NSW Department of Health has recently released Guidelines for predictive and diagnostic DNA testing for serious adult onset neurogenetic disorders, with predictive implications for other family members and which are likely to reduce normal life expectancy They can be found on the Health Department's website:

<http://www.health.nsw.gov.au/fcsd/rmc/cib/circulars/2003/cir2003-25.pdf>

The guidelines state that DNA predictive testing for serious adult onset neurogenetic disorders carried out by NSW Health public hospital laboratories shall only be undertaken when requested by certified clinical geneticists.

The rationale behind this policy is to ensure that patients receive care according to best practice guidelines. Taking a predictive DNA test is a major life decision and the results are irreversible. Predictive testing raises a number of complex issues and it is essential that prior to undertaking testing, the patient is fully informed about the implications of testing and is prepared for the results.

Disability Services Abuse and Neglect Hotline

1800 880 052

TTY 1800 301 130

NRS 1800 555 677

TIS 131 450

The Hotline is a single contact point for anyone to report claims of abuse and neglect against people with disabilities in any commonwealth, state or territory government run or funded disability service.

Toll Free Numbers

Carers Respite Centre 1800 059 059
Disability Council of NSW 1800 044 848
People with Disabilities NSW Inc 1800 020 613
Telstra Assistance for People with a Disability 1800 068 424

Technical Aid to the Disabled

Admin.Website:
www.technicalaidnsw.org.au
Custom designed aids service
email: cda@technicalaidnsw.org.au

QANTAS CARER CONCESSION CARD

The QANTAS Carer Concession Card is issued to people with a disability and high-level support needs who require the full time assistance of a carer whilst they are on the plane.

The persons are eligible if they need to have one to one support when seated on the plane for tasks such as feeding, transferring to the bathroom, communicating with the flight staff etc.

Persons would not be eligible if they only need assistance boarding the plane, or when they arrive at their destination.

Cardholders receive 50% discount on the standard full price domestic air travel, in addition to 50% off their carers fare. Please note that the Carer Concession Card does not apply to already discounted fares or 21 days in advance fares. The card will not reduce a child's fare any further but will reduce their adult carers fare by 50%.

This card is a photo ID card which is valid for three years and has an administration fee of \$27.50 including GST.

For further information and an application form contact NICAN on:
Phone: 1800 806 769 Fax: (02) 6285 3714



CONTACT CORNER

AGSA will publish requests for contacts and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The AGSA office may be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-judgmental approach is recommended in establishing contact.

Aarskog Syndrome

A family with 2 generations affected by this syndrome would like contact with other families.

Congenital Protein C Deficiency

A young woman with this condition is looking for individuals or any support groups in Australia that cater for this or similar conditions.

Hypoglossia-Hypodactylia syndrome

The parents of a young baby boy are seeking contact with other families that may have a child with this condition.

Scheuermann's Disease

A family whose husband has recently been diagnosed with this disease would like to make contact with other families.

Multiple Epiphyseal Dysplasia

A mother of an 11-year-old boy would like to talk with other parents of children with similar disorders.

Tuberous Sclerosis

The mother of an 11-year-old girl would like contact with other parents of children with this condition.

PROFILE**A - Z GENETIC
CONDITIONS**

It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness within our membership of the range of genetic conditions. Also it hopes that where overlaps occur in conditions, support Groups may liaise with each other and thus gain a broader understanding of facilities, aids, etc. that may be of value to your individual membership.

Please ensure that all support group information is recent and reliable. It is of paramount importance that you let us know your group is "Alive and Well" and happy to take referrals.

XERODERMA PIGMENTOSUM**Also known as:**

XP

Kaposi disease

Xeroderma Pigmentosum, Variant type XP-V

Disorder Subdivisions:

Xeroderma Pigmentosum, Type A, I, XPA;

Classical form

Xeroderma Pigmentosum, Type B, II, XPB;

Xeroderma Pigmentosum, Type C, III, XPC;

Xeroderma Pigmentosum, Type D, IV, XPD;

Xeroderma Pigmentosum, Type E, V, XPE;

Xeroderma Pigmentosum, Type F, VI, XPF;

Xeroderma Pigmentosum, Type G, VII,

XPG;

Xeroderma Pigmentosum, Dominant Type

General information about Xeroderma**Pigmentosum:**

Xeroderma pigmentosum (XP) is a group of rare inherited skin disorders in which the features are:

- a heightened reaction to sunlight (*photosensitivity*) with skin blistering occurring after exposure to the sun;
- pain and blistering *immediately* after contact with sunlight in some cases;
- acute sunburn and persistent redness or inflammation of the skin (*erythema*).

In most cases, these symptoms may be apparent immediately after birth or occur within the next three years. In other cases, symptoms may not develop until later in childhood or, more rarely, may not be recognised until adulthood. Other symptoms of XP may include discolourations, weakness and fragility, and/or scarring of the skin. Xeroderma pigmentosum affects the eyes as well as the skin, has been associated with several forms of skin cancer, and, in some cases, may occur along with short stature, mental retardation, and/or delayed development.

Several subtypes of XP have been identified, based upon different defects in the body's ability to repair DNA damaged by ultraviolet light (UV). According to the medical literature, the symptoms and findings associated with the classic form of xeroderma pigmentosum, known as XP, type A (XPA), may also occur in association with the other XP subtypes. These include: XP, type B (XPB); XP, type C (XPC); XP, type D (XPD); XP, type E (XPE); XP, type F (XPF); XP, type G (XPG).

These XP subtypes all follow a pattern of genetic inheritance in families called autosomal recessive inheritance. In addition, another subtype of the disorder, known as XP, dominant type

follows a different inheritance pattern called autosomal dominant inheritance.

Symptoms of Xeroderma Pigmentosum in more detail:

The earliest signs of xeroderma pigmentosum (XP) include excessive freckling and an extremely heightened reaction to sunlight (*photosensitivity*), with skin blistering occurring after exposure to the sun. In some cases, pain and blistering may occur immediately after contact with sunlight. Acute sunburn and persistent redness or inflammation of the skin (*erythema*) are also early symptoms of XP. In most cases, these symptoms may be apparent immediately after birth or by the age of three years. However, in some cases, symptoms may not be apparent until later in childhood, or more rarely, during adulthood.

Additional skin abnormalities often develop, including abnormal darkening of the skin (*hyperpigmentation*), diminished pigmentation of the skin (*hypopigmentation*), and/or excessive scarring. Small red skin lesions (*telangiectasias*) may develop due to abnormal widening of blood vessels. In addition, the skin may become weakened and excessively fragile, degenerative (*atrophic*) changes may occur, and the skin may appear unusually dry and smooth.

Most affected individuals also have abnormalities of the eyes, most notably:

- an extreme intolerance to light (*photophobia*);
- inflammation of the corneas (*keratitis*);
- inflammation of the transparent membrane lining the insides of the eyelids and covering the whites of the eyes (*conjunctivitis*);
- excessive tears (*lacrimation*);

- clouding of the lens of the eyes (*opacities*).

In addition, in many individuals with XP, the eyelids may abnormally turn outward (*ectropion*) or inward (*entropion*).

Several types of benign (*non-malignant*) and malignant tumours may be associated with XP, with onset possible before the age of five in some cases. Sunlight-induced skin cancers (e.g., malignant melanoma, basal cell carcinoma, and squamous cell carcinoma) commonly develop in people with XP, most often affecting the head, neck, and face. Individuals with XP may also be prone to developing certain pre-malignant or benign (*non-cancerous*) skin tumours, such as tumours consisting of blood vessels (*angiomas*) or certain cells that cover particular body surfaces, as well as tumours of the eyelids, corneas, and/or tip of the tongue. The severity of associated skin and eye abnormalities may depend on the amount of exposure to ultraviolet light.

Unusually slow development, dwarfism, mental retardation, and neurological impairments may also be associated with some cases of XP. Such neurological impairments may include:

- absent or weakened reflexes (*areflexia or hyporeflexia*);
- an abnormally small head (*microcephaly*);
- hearing impairment (*sensorineural deafness*);
- increased rigidity in certain muscles, causing stiffness and limitation of movement (*spasticity*);
- loss of voluntary coordination (*ataxia*).

Research has shown that there is a correlation between the degree of sensitivity that cells have to UV light and the severity of neurological symptoms. It is concluded that the repair of DNA,

thus preventing the premature death of neurons or nerve cells, is required to maintain a healthy nervous system.

DNA repair rates have been calculated for some forms of XP. In Group A, it is less than 2% of normal; in Group B, 3-7%; in Group C, 10-25% and in Group D, 25-55% of normal.

When XP occurs in association with mental retardation, short stature, abnormal reproductive function (*hypogonadism*), and, in some cases, neurological abnormalities, the disorder is referred to as De Sanctis-Cacchione syndrome.

The range and severity of associated abnormalities may vary dramatically among affected individuals. The previously mentioned symptoms and findings may occur in association with any of the subdivisions of XP.

XP, type B (XPB) is a rare form of XP and is sometimes found in association with Cockayne syndrome. Cockayne syndrome is a rare inherited disorder characterised by growth retardation, abnormal sensitivity to light (*photosensitivity*), an abnormally small head (*microcephaly*), and a prematurely aged (*progeroid*) appearance. Children with this disorder may scar easily and have an increased amount of colour (*pigmentation*) in the skin.

In some cases, **XP, type C (XPC)** may be associated with an inflammatory autoimmune disorder of the connective tissues (*lupus*). In addition, individuals with this type of XP are particularly prone to developing a certain type of skin cancer (*malignant melanoma*). After XPA, this is the most common form of XP.

XP, type D (XPD) is a rare form of XP which is characterised by sparse and brittle hair (*trichothiodystrophy*), short stature, unusual facial features,

abnormal sexual development (*hypogonadism*), and scaling of the skin (*ichthyosis*). Some individuals with XPD may also experience a late onset of neurological symptoms, such as mental deterioration and/or loss of voluntary coordination (*ataxia*).

XP, type E (XPE) is very rare and has been associated with minimal or no neurological symptoms, and most people with this XP subtype have a less severe form of skin disease. However, individuals affected by XPE may be susceptible to early onset of some types of skin cancer (e.g., basal cell carcinoma, squamous cell carcinoma, malignant melanoma).

XP, type F (XPF) has been associated with minimal or no occurrence of skin cancer, neurological abnormalities, or disorders of the eyes in most cases. This is a rare form of XP and most cases have been found in Japan.

XP, type G (XPG) has been associated with normal physical and neurological development and mild skin changes.

Little is known about XP, dominant type other than that it is transmitted as an autosomal dominant trait and is a milder form of the classic type of XP.

As with the other subtypes of xeroderma pigmentosum, XP, variant type (XP-V) may be characterised by symptoms and findings seen in those with the classic form of XP. However, XP-V has been associated with no neurological symptoms. In addition, affected individuals may be prone to the early onset of certain sunlight-induced skin cancers (e.g., basal cell carcinoma, squamous cell carcinoma, malignant melanoma).

What causes Xeroderma Pigmentosum?:

Xeroderma pigmentosum (XP) types A through to G as well as XP, variant type

are thought to follow an autosomal recessive inheritance pattern. In this form of inheritance, both parents carry one faulty copy of the gene for XP which they pass on to their children. Whilst carrier parents do not have symptoms of XP, it is thought that being a carrier may predispose an individual to developing skin cancer, particularly if that individual has had a lot of exposure to the sun.

In some cases, the parents of individuals with XP have been closely related by blood (*consanguineous*). In these cases, if both parents carry the same disease gene, there is a higher-than-normal risk that their children may inherit the two faulty genes necessary for the development of the disease.

One rare form of xeroderma pigmentosum, known as XP, dominant type, is transmitted as an autosomal dominant trait.

In most subtypes of XP, associated symptoms and findings result due to the body's inability to repair deoxyribonucleic acid (DNA) damaged by exposure to ultraviolet light (UV). DNA is the carrier of the genetic code within cells. The coded DNA instructions within genes consist of different arrangements of four basic chemicals (nucleotide bases) known as adenine (A), cytosine (C), guanine (G), and thymine (T). In many individuals with XP, cells are unable or have a reduced capacity to carry out "nucleotide excision repair" (NER), a complex process during which nucleotides with UV-induced damage are removed. As a result, as damaged DNA replicates itself during the division and reproduction of cells, errors or "mutations" within the coded DNA instructions continue to accumulate. Researchers have classified the different XP groups (e.g., XPA, XPB, etc.) based upon specific defects in the body's ability to

repair UV-damaged DNA. These groups are known as complementation groups.

In addition, researchers have classified XP, variant type (XP-V) based upon the fact that XP-V cells have a normal or near normal ability to repair UV-induced DNA damage. However, such cells are defective in replicating UV-damaged DNA; in other words, genetic material within the XP-V cells is significantly more likely to mutate due to exposure to UV radiation than otherwise expected (*hypermutability*).

Disease genes that may be responsible for causing XP subtypes A through to G have been mapped to particular chromosomes. The affected genes are all thought to play some role in the repair of UV-damaged DNA (i.e., nucleotide excision repair). Chromosomes are found in the nucleus of all body cells. They carry the genetic characteristics of each individual. Pairs of human chromosomes are numbered from 1 through to 22, with an unequal 23rd pair of X and Y chromosomes for males and two X chromosomes for females. Each chromosome has a short arm designated as "p" and a long arm identified by the letter "q." Chromosomes are further subdivided into bands that are numbered.

A disease gene has been located on:

- the long arm (q) of chromosome 9 (9q22.3) in the **classic form of XP, type A**;
- on the long arm (q) of chromosome 2 (2q21) in **XP, type B**;
- on the short arm (p) of chromosome 3 (3p25) in **XP, type C**;
- abnormal changes (mutations) of a DNA repair gene (known as ERCC2), mapped to the long arm of chromosome 19 (19q13.2-q13.3), are thought to play a role in causing **XP, type D**;

- **XP, type E** is thought to be caused by mutations of a gene linked to the short arm of chromosome 11 (11p12-p11);
- mutations of the "ERCC4" gene, which has been mapped to the short arm of chromosome 16 (16p13.2-p13.1), have been identified in individuals with **XP, type F**;
- abnormal changes of a DNA repair gene on the long arm of chromosome 13 (13q33) have been implicated in cases of **XP, type G**.

In individuals with **XP-V**, researchers have identified mutations in a gene known as POLH (also known as the DNA polymerase eta gene) on **chromosome 6**, which is the human counterpart (*homologue*) of a similar gene in yeast (RAD30). According to researchers, the POLH gene (like its yeast counterpart) appears to encode an enzyme (i.e., DNA polymerase) that enables error-free copying of cells by "bypassing" UV-induced damage. As a result, proper functioning of this so-called "damage-bypass replication protein" plays an important role in helping to minimise the occurrence of certain skin cancers that may be induced by exposure to sunlight. Accordingly, in individuals with XP-V, mutations of the POLH gene appears to result in impaired replication of UV-damaged DNA and an increased susceptibility to UV-induced skin cancers.

Who is affected by Xeroderma Pigmentosum?:

Xeroderma pigmentosum (XP) appears to affect males and females in equal numbers. The onset of symptoms usually occurs immediately after birth or within the next three years of life. However, in some cases, symptoms may not develop until later childhood. In addition, rare cases of adult onset have been reported in the medical literature.

XP affects approximately one in 100,000 individuals worldwide and appears to occur more frequently in Japan.

Is there any treatment for Xeroderma Pigmentosum?:

Diagnosis

In some cases, a diagnosis of xeroderma pigmentosum (XP) may be suspected before birth (*prenatally*) based upon the results of specialised tests, such as amniocentesis. During amniocentesis, a sample of fluid that surrounds the developing fetus is removed and studied. Such prenatal screening procedures may be conducted for families with a history of XP.

In many cases, XP is diagnosed or confirmed during infancy or childhood based upon a thorough clinical evaluation, characteristic physical findings, a detailed patient and family history, and certain specialised laboratory tests. For example, in some cases, samples of certain cells, such as white blood cells (*lymphocytes*) and skin cells (*fibroblasts*), may be exposed to UV radiation and studied microscopically to detect impaired DNA repair or defective replication of UV-damaged DNA.

Affected individuals should be regularly monitored by physicians to ensure prompt detection and appropriate treatment of skin lesions, eye abnormalities, and/or other symptoms and findings potentially associated with the disorder.

Treatment

There is no cure for XP. However, for individuals with XP, it is essential to minimise sun exposure as much as possible to protect against ultraviolet radiation. Appropriate measures include

limiting time outdoors during daylight hours, using special (e.g., broad-spectrum) topical sunscreens, wearing sunglasses and double layers of clothing, and shielding glass in windows to prevent the passage of sunlight. In addition, affected individuals should take appropriate precautions to avoid exposure to certain chemical carcinogens, such as those in cigarette smoke.

Early surgical removal of and/or other appropriate treatment measures for skin tumours (*neoplasms*) are essential for individuals with XP.

A plastic surgery procedure (dermabrasion), chemical peels, or grafting of skin from areas that have not been exposed to sunlight have been used in some individuals with xeroderma pigmentosum (XP). In addition, premalignant or early lesions may be effectively treated in some cases. In some children with XP, application of specific cream medication may help to prevent tumour development.

Other experimental procedures are being investigated to help with eye symptoms.

Resources

The following overseas groups may be able to provide additional information and support:

Skin Cancer Foundation

245 Fifth Avenue

Suite 1403

New York, NY 10016 USA

Tel: (212) 725-5176

email: info@skincancer.org

XP (Xeroderma Pigmentosum) Society, Inc.

Box 4759

Poughkeepsie, NY 12602 USA

Tel: (518) 851-2612

e-mail: xps@xps.org

Home Page: <http://www.xps.org>

A FAMILY STORY

Imagine a bunch of 20 campers and their families enjoying morning activities including craft, games, computers etc. During this session parents have a chance to learn more about their child's illness and adult sufferers gain confidence to live as normal a life as possible. After a meal the campers and their families hop on a coach to destinations unknown, one outing is a country fair with food, rides, animals etc, another is adventure climbing and abseiling or maybe hiking and canoeing. After a final meal the coach returns the exhausted campers to a well deserved sleep.

But this is no ordinary camp. Morning activities commence inside at 1.00pm and the coach load of campers leaves the campsite at 8.30pm as the sun goes down. As the sun's first light appears on the horizon weary campers drag themselves off to bed.

Campers share the experience of living with Xeroderma Pigmentosum, a rare genetic disease that prevents the patient's cells from repairing DNA after exposure to ultraviolet radiation (UVR). Eventually the build up of damage leads to the development of all types of skin cancers. As sunlight is the main source of UVR, to prevent or minimize the risk of getting skin and other cancers, sufferers go to extensive lengths to protect themselves from sun damage.

Our daughter, Mary, was diagnosed with Xeroderma Pigmentosum in 1997 at the age of four and a half. By the time Mary was three years old she had suffered 4 or 5 peeling sunburns. We tried to be diligent in sun protection but became alarmed as we realized her skin seemed to be showing signs of extensive damage. What appeared to be little

white scars covered her arms and legs and her face was marked with freckles and moles despite her skin not being very fair. Her face and hands always seemed to be red and sore and in the summer months she would shield her eyes whilst outside.

At this point our family doctor referred Mary to a dermatologist who, to my dismay, raised the possibility that Mary may have a condition that sees children presenting at an early age with skin cancer. (The average age for development of skin cancer is about 58 years of age.) I had expected to be reassured that Mary's symptoms were common and that I needn't worry. Over the next 8 months Mary was seen by a paediatrician, and geneticist with the diagnosis confirmed by her skin cells being sent to England and tested to determine their ability to repair after being damaged by UVR.

Our lives changed forever from this moment. Mary's wardrobe completely changed to include long pants and long sleeve cotton shirts. Wrap around hats and gloves have become a normal part of Mary's attire. A strict routine of sunscreen applied liberally every two hours throughout the day, every day of the year has been implemented. We had tinting applied to the windows of our home and car to increase the protection from UVR that comes through glass. We also had an airconditioner fitted to the family van so that we could keep the car windows closed to maximize the protection.

Family picnics and BBQ's on Sunday afternoons are a distant memory and our family routine is very different to how we expected to live our lives. Either my husband or I accompany our four boys to sporting events and sometimes it is impossible for us to participate in extended family celebrations. We make the most of evenings and indoor pools

and playgrounds. We turn up to events and venues as everybody else is going home.

Mary's school has implemented the same precautions that we have put in place at home

Mary is also one of the 25% of sufferers of XP who also have neurological symptoms including difficulties in balance, delayed speech, difficulties in gross motor and fine motor skills. Mary receives some special help with her schoolwork.

When asked what she would wish for if she had just one wish, Mary often replies that she wishes she could be like other kids and play outside. In some ways we were able to grant her this wish when we took her to Camp Sundown in America. She was just like the other kids and she did get to play outside without fear of being sun burnt. We got to meet other parents who had their world turned upside down and literally outside in. We keep in contact with the Xeroderma Pigmentosum Society, which is based in America, through the internet.

Despite her restricted lifestyle I would describe Mary as an optimistic little girl, always making the most of every opportunity that comes her way. Her teachers say that you just can't help feeling happy around her.

It is sometimes hard to make decisions for the well being of a child that then isolates us from family, friends and the rest of the community. What we need to ask of schools and community groups is unusual. Diagnosis is not always speedy and testing is costly. Our family has valued the support we have gained from being in contact with other families overseas via the internet. I am very interested in starting a support group here in Australia so that we can support and encourage one another, also so that there is someone for families to turn to

when a diagnosis of Xeroderma Pigmentosum is suspected or newly confirmed. To this end I would welcome hearing from other families living with XP.

Lynda Begg
9 Southern Terrace,
Holden Hill, S.A.5088
Ph: (08) 8369 1086 Email: lynda@badit.com

Presentation of stem cell research – Muscular Dystrophy Association

I attended the afternoon symposium on stem cell research carried out in Sydney, which was held on Saturday 15th March 2003. The afternoon of research and update talks was presented by the NSW Muscular Dystrophy Association, and held at the Burwood RSL Club. It was well attended and well received by members of the public, families and professionals alike.

Dr Thomas Yeoh from the Victor Chang Institute, began proceedings by discussing some of his work on muscle stem cells, and also summarized some of the research being carried out worldwide. Dr Yeoh's work is proudly supported by the New South Wales Muscular Dystrophy Association.

Adult muscle stem cells are important for the continual renewal of injured or damaged muscle tissues. A **stem cell** is a cell whose daughter cells may give rise to other cell types. Dr Yeoh discussed different groups of **pluripotent** stem cells (potential to differentiate or develop into a number of mature cell types with different functions) in the muscle. The identification of at least two different stem cell populations in muscle tissue – satellite stem cells, and muscle derived stem cells (MDSC), has led to an interest in the potential use of these cells in the treatment of muscle disorders. Both of these cell types

display different capacities to activate regeneration of muscle and some other cell types, such as blood cells.

Dr Yeoh's specific interests are to identify and better understand the naturally occurring chemical triggers in the body that activate differentiation or specialization of stem cells into mature muscle cells. Delineating these specific enzyme pathways that occur in the human body may be useful in treatment applications. For instance, it is known that by activating stem cells to differentiate into mature muscle cells in a culture dish in the laboratory, and then transplanting these cells back into animals with damaged heart muscle, results in better heart muscle function. By mimicking natural growth conditions of stem cells as best as possible in the test tube, it is presumed that there may be greater success in such transplant models.

Success of such treatments is also dependent on the ability of transplanted stem cells to repopulate and integrate into the tissue in a 'natural, orderly way', so that the regenerated muscle functions normally. For instance, healthy muscle with normal function requires a network of blood vessels, which need to feed the system. These vessels would also need to be regenerated, along with muscle fibres. There are mouse models that show that it is possible to regenerate the bone marrow (following depletion) by injecting MDSCs. This is an example of transplanted stem cells successfully repopulating depleted or damaged tissue.

It is thought that were there is an inherent dysfunction of muscle, such as the muscular dystrophies, the pool of stem cells and system of regenerations is 'exhausted'. Perhaps it is possible to override this problem by introducing cultured stem cells, which have been activated to form muscle tissue.

Professor Anne Cunningham from The Children Medical Research Institute Westmead presented a thorough overview of adult stem cell biology and her exciting work in the area of adult neural stem cells.

Although adult stem cells have been identified in a number of human tissues such as the brain, liver, bone marrow and skeletal muscle, their normal functions have not been completely elucidated. An implied function in tissue repair makes stem cells an important medium for research into treatments of human disease. It was originally thought that stem cells from a particular organ have only the capacity to differentiate into cells of that organ. It has now been found that adult stem cells are perhaps more 'plastic' than first thought, and may have the ability to differentiate into multiple cell types, perhaps not restricted to a particular organ or tissue type. It is therefore thought to be the specific environment or stimuli, which enable specific differentiation of a stem cell into a particular mature cell type.

Therapeutic potential is recognized in stimulating natural occurring stem cells in a given organ, to form mature, functional tissue of a specified type. This may be an alternative to transplantation of mature cells or stem cells into a damaged organ.

Professor Cunningham's group is working on neurons or nervous tissue of the olfactory system (important for the sense of smell). They are in the process of perfecting the techniques for growing neurons from putative stem cells in a culture dish, in lab-controlled conditions. Professor Cunningham presented exciting video footage of the growth of these cells in microscopy real time. Neurospheres, or balls of neural cells as seen in the human brain, were observed in culture. These balls of cells

had the ability to move and attach to the culture dish they were growing in. They were then able to differentiate into daughter cells that formed neuronal projections, much like those seen in human nerve growth.

Professor Cunningham's neurospheres demonstrated fast proliferation or growth, and the ability to move. These properties are believed to be important in the success rate of stem cell therapy. A potential application may be the biopsy of a patient's tissue to be cultured in the laboratory, in order to test agents/drugs for possible problems or benefits, specific to that patient. This may assist in the tailored management of any given individual.

Professor Cunningham has been involved in creating a Stem Cell Interest Group, which enables scientists to pool and compare research in stem cells, to assist progress in this area. This is the first such interest group in Australia, in the area of stem cell research.

Gayathri Parasivam, Associate Genetic Counsellor, AGSA Committee Member

Stem Cells – the legislation

The Prohibition of Human Cloning Bill 2002 and the Research Involving Embryo Bill 2002 appear to be approaching the last stages of their passage through the Federal Parliament.

If the Bills pass unscathed into legislation, we can expect that the State and Territory Parliaments will follow with corresponding legislation as agreed by the Council of Australian Governments on 5 April 2002 and that the Embryo Research Licensing Committee will be established.

This Paper will cover:

- the recent history of Government activity in relation to the debate

over human cloning, unacceptable practices and research involving embryos and the 5 April 2002 decision by COAG;

- the passage of the Bills through Federal Parliament to date;
- the regulatory scheme established by the Bills; and
- likely developments and issues to consider once the legislation is passed.

CONFERENCES

A National Conference on the Physical Mental and Social Health of People with Developmental Disabilities

The Masonic Centre,
279 Castlereagh Street Sydney
Presented by Centre for Developmental Disability Studies in association with Association of Doctors in Developmental Disability (ADIDD)

Keynote speakers

Gillian Calvert is Commissioner for children and Young People in New SW Australia.

Eric Emerson is Professor of Clinical Psychology at the Institute for Developmental Disabilities Studies.

Steve Moss is a Consultant Research Psychologist at the Estia Centre.

Guy's Hospital, London.

Patricia Noonan Walsh is Ireland's first Professor of Disability Studies and Vice-President of the European Network on Intellectual Disability and Ageing.

For further information Contact Conference CDDS on 02 8878 0500

6th International Turner Syndrome Conference 10th to 13th July 2003

**Masonic Centre
279 Castlereagh Street Sydney**

Overseas Guest Speakers

Dr Gerard Conway (UK) - TS in AdultLife
Dr Rodney Baber (Aust)- Current Issues with HRT

Dr Morgan Forbes (Canada) Celebrating Turner Culture

Laetitia Greyling (Sth Africa) - Research and Recommendations regarding support and Guidance to Turner Syndrome

Dr Charmian Quigley (USA) Results of TS Toddler Study

For further details:-

Contact Glenn Fisher on

02 9452 4196 or

email: turnersyn@netpro.net.au

website: www.turnersyndrome.org.au

NFAA Family Camp - 19 - 21 September 2003

Contact the Neurofibromatosis

Association of Australia on:

Ph: (02) 9874 0844

Fax: (02) 9874 0144

Perspectives on Health in Developmental Disability.

A National conference on the Physical, mental and social health of children & adults with developmental disability will be presented by the Centre for

Developmental Disability Studies, The University of Sydney in association with

the Association of Doctors in Developmental Disability (ADIDD) on

June 26 - 28 2003 at the Sydney Masonic Centre. For further enquiries:

Ph (02) 8878 0500 Fax (02) 9807 7053

or E-mail Helen Moore-

hmoore@med.usyd.edu.au

Cornelia de Lange Syndrome

Association of Australia is hosting its first Australian based international conference at Penrith Panthers Sydney 3 - 6 July 2003.

July 3 is the professional day for genetic counsellors and geneticists and the 4-6 July will be a conference for families living with the syndrome.

For more details and programme information email Jenny Rollo at email: jennyrollo@bigpond.com or website: www.cdlsau.org.au

**International Congress of Genetics
"Genomics The Linkage to Life: 6-11
July 2003 Melbourne Australia**

Within this conference the Human Genetics Society of Australasia and the Australian Society of Genetic Counsellors conferences will be incorporated. Many excellent speakers have been invited, eight of whom are Nobel Laureates.

For more information:

www.geneticscongress2003.com

Attention Deficit and Hyperactivity Disorder Information and Support Services (ADDISS) is hosting a conference on "Ways and Means to Reach to Teach" on Friday 22 August 2003 at the Carlton Crest Hotel Brisbane. This conference is aimed at schoolteachers, and will feature speakers who will present a collection of practical strategies to assist in catering for the needs of children with ADHD. For registration enquiries contact Jenny on: ph: 07 3855 3711 Fax: 07 3855 2811 or email: jenny@eventsolutions.com.au

**5-6 September 2003
"Shaping Our Future"**

Partnership for Positive Outcomes National MDDA Conference held in conjunction with the ASIEM's International Congress on Inborn Errors of Metabolism Brisbane Convention Centre, Queensland. Cost to be advised Contact MDDA on 1800 288 460 or email. mdda@vtown.com.au

Life Activities International Conference on Disability 21-24 September 2003 Newcastle NSW Australia

Keynote speakers will include Dr Patricia A Morrissey, Commissioner for the US Dept. Of Health & Human Sciences Dr Colleen Wieck Executive, Director for the Governor Council on Developmental Disabilities More Info: Web: www.lifeactivities.org.au/conference

Huntington's Disease National Conference 2003 "Spring in Tasmania"

The Tasmanian HD Association is hosting the 2003 National Conference on 6th & 7th November at the Chancellor Inn, Burnie. Contact your state office for more details or e-mail Joyceabblitt@bigpond.com

**Perspectives on Health
In Developmental Disability**

National conference on the physical, mental and social health of children & adults with developmental disability.

Presented by:

Centre for Developmental Disability Studies, The University of Sydney in association with Association of Doctors in Developmental Disability (ADIDD)

26th 27th 28th June 2003

The Sydney Masonic Centre, 279 Castlereagh Street, Sydney NSW Australia

National & International Keynote Speakers include:

Professor Eric Emerson, Lancaster University, UK

Dr Steven Moss, Estia Centre, Guy's Hospital, London, UK

Professor Patricia Noonan-Walsh, University College Dublin, Ireland

Professor Bob Williamson, Murdoch Children's Research Inst. Melbourne

Enquiries

02 8878 0500 Fax: 02 9807 7503

email: Helen Moore - hmoore@med.usyd.edu.au or Leela Pacheco - lpacheco@med.usyd.edu.au

Workshops

Monday 30 June 2003

Mental Health: An introduction for direct support staff

Tuesday 1 July 2003

Developing health indicators for people with intellectual disability in Australia

Wednesday 2 July 2003

Clinical assessment of psychiatric disorders in people with intellectual disability

Thursday 3 July 2003

Accredited Training in the Mini PAS-ADD psychiatric interview schedule

**GENETIC DISORDERS AWARENESS WEEK
LAUNCH 9th September 2003**

AGSA NEWSLETTER

BECAUSE OF THE LATEST OF THIS NEWSLETTER WE HAVE COMBINED THE APRIL AND JUNE ISSUES. THANK YOU.

OVERSEAS NEWS



Sturge-Weber Foundation (US) Annual International conference Denver, Colorado

11 - 13 July 2003

For more information

See: www.sturge-weber.com

2003 Genetic Alliance Annual Conference

“Securing Our Futures Conference”

will be held on August 1 - 3, 2003 at the Key Bridge Marriott Hotel, Arlington, VA. USA. This will be an international gathering of lay advocacy organisations, community representatives and professionals. For more information go to

<http://www.geneticalliance.org>.

VCFS Research Study The Children's Hospital at Westmead is now recruiting patients with VCFS for a study. Adults are invited to participate, whilst children must be over 5 years of age. For further details contact

Dr Julie Curtin,

Department of Haematology,

The Children's Hospital at Westmead,

on 02 9845 3082 or

Dr Meredith Wilson,

Clinical Geneticist, Head of Department

on 02 9845 3273.



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