THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

FUNDED BY THE NSW HEALTH DEPARTMENT

N E W S L E T T E R

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MISSION STATEMENT

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

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Contents

EDITORIAL

Welcome to the first issue for 2002. We hope you had a wonderful Christmas / New Year break.
Dianne Petrie has asked me to introduce myself by throwing me into the deep end to write the editorial for this issue. My name is Halimah Simpson and I am the new Administrative Officer at AGSA. This is a new, part time position so hopefully the role will evolve as I do!

Hopefully, my being here will mean that Dianne has more time to work with families and organize seminars etc. I will generally be working on Thursdays and Fridays. Feel free to say hello!

AGSA had a discussion forum on the 11th February 2002, with the Australian Law Reform Commission regarding the Protection of Human Genetic Information Issues Paper. The deadline for submissions is now the 18th March 2002. If you have any privacy, employment or insurance issues or examples, please contact us so that we can include your experiences in the AGSA submission. It would be useful to get examples of "Good Practice" as well as "Bad Practice", so if you have either, please let us know.

Best wishes,

Halimah Simpson and Dianne Petrie

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.

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POLYCYSTIC KIDNEY DISEASE
Contact is wanted with anyone who has had experience with this condition. Contact AGSA for details.

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FAMILIAL SPASTIC PARAPARESIS
Contact with others who have this condition is wanted. Contact AGSA for details.

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CLEIDOCRANIAL DYSOSTOSIS
A mother of a two and a half year old girl would like contact with other parents or people with experience of this condition. Contact AGSA for details.

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X-LINKED AGAMMAGLOBULINAEMIA
The mother of a 6 month old baby boy would like contact with other parents or people with experience of this condition. Contact AGSA for details.

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MIXED GONADAL DYSGENESIS
The mother of a 4 ½ year old boy would like contact with other parents and contact with other children for her son. Contact AGSA for details.

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RUBINSTEIN TABYI
The mother of an 11 month old baby girl would like contact with other parents or people with experience of this condition. Contact AGSA for details.

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DUBOWITZ SYNDROME
The mother of a boy would like to make contact with others who have experience of this condition. Contact AGSA for details.

SUPPORT GROUP NEWS as at 27th February 2002

OVERSEAS NEWS

(From Donor Conception Support Group Newsletter January 2002)
The United Kingdom has just released a consultation paper on Donor Information. It is asking for people's opinions on the type of information that should be recorded on donors and who should have access to that information. You can access the paper at the health department's web site: www.doh.gov.uk/gametedonors

The Canadian government has just released a standing committee report looking at legislation in the area of reproductive technology and it includes donor conception and access information. The report can be accessed at: www.parl.gc.ca

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CARERS NSW Inc
UPDATED CARER INFORMATION

PACK The updated Carer Information Pack now includes the "Carer Relaxation" recording on CD and cassette. If you would like to receive a free Information Pack, contact the Carer Resource Centre on (02) 9280 4744.

Coeliac Awareness Week March 13th to 20th

For every known person with Coeliac Disease there are probably five to ten others who have not been diagnosed

Fatigue, weakness and lethargy?

Flatulence, bloating, abdominal pain?

Unexplained anaemia?

Vitamin deficiencies?

Diarrhoea or constipation?

Unexplained early onset of Osteoporosis?

Coeliac Disease is a genetically based permanent intolerance to dietary gluten (found in wheat, rye, barley and oats). This leads to damage to the lining of the small bowel. Coeliac Disease can occur at any age.

While there have not been any recent studies undertaken in Australia to determine how common Coeliac Disease is, a recent study has been undertaken in New Zealand. Dr Bramwell Cook, a New Zealand Gastroenterologist, reports that the true prevalence probably lies somewhere between 1 in 100 and 1 in 200. What can be said is that for every known person with Coeliac Disease there are probably five to ten others who have not been recognized.

The symptoms can vary from one or more of the following: Anaemia-fatigue, weakness and lethargy, vitamin deficiency, diarrhoea, sometimes constipation, flatulence, bloating, abdominal pain, nausea and vomiting, mouth ulcers, bone and joint pain, miscarriages and infertility, delayed puberty, skin rashes, dental abnormalities, retarded growth in children, mood changes and irritability.

The diagnosis relies upon proving that the small bowel lining shows the typical (villous atrophy) of Coeliac Disease. This is done by endoscopy. A specific panel of blood tests that measure antibodies to gluten is available as a screening aide in the diagnosis.

Although Coeliac Disease cannot be cured it can easily be controlled by a gluten fee diet for life. A gluten free diet should never be started before an endoscopy or blood tests, as it will interfere with establishing the correct diagnosis or may delay the diagnosis of another condition with similar symptoms.

GRIEF AND LOSS – SUPPORT FOR PARENTS AND CHILDREN

24 hour telephone services are:
Kids Help Line: 1800 55 1800
Lifeline: 131 114
Bereavement C.A.R.E. Centre: (02) 9869 3330
A free service is available for people experiencing financial difficulties. Other specialists who work with children and adolescents such as paediatricians and child psychologists may also be able to provide help.
Youthline (youth counselling):
(02) 9951 5522 (Sydney)
(02) 9633 3666 (Parramatta)
Compassionate Friends for parents whose children have died (02) 9290 2355 or 1800 671621 (Freecall outside Sydney)

NEWS FLASH – Mucoppolysaccharide and Related Diseases Society (MPS)

have been offered the opportunity to raise funds for the society through the sale of tickets to a new show coming to Sydney in March 2002. The show is called "**A Handful of Keys**" and is a 2 man comedy cabaret, and comes to Sydney from a long season in South Africa. The opportunity to benefit has arisen from the writer and one of the show stars, Mr Ian Von Mementy, who is an MPS parent. Regular show ticket prices are $37.50, and for each ticket sold through the Society, they will donate $15. Two show sessions have been identified: **Friday 8th March, 8:15pm** **Sunday 10th**

March, 3pm The show is being held at the Paddington RSL auditorium. Please contact David Oliver on (02) -9476 8411 or Isabel Glasson on (02) 9907 9660.

***DISABILITY, NSW, 2001* (ABS)**

1 in 5 people in NSW have a disability

1 in 8 were carers

47% own their home

45% of working age were employed

42% held a post school qualification

29% participated in sport/physical recreation

25% needed transport assistance

ARE YOU LOOKING FOR DISABILITY INFORMATION IN QUEENSLAND?
The Disability Information and Awareness Line can help you:
(07) 3224 8444
1800 177 120 Toll Free
1800 010 222 TTY Toll Free

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.

Pallister-Hall Syndrome (PHS)

By George William Helon

Introduction

These days it is commonplace for genetic disorders and syndromes to be diagnosed within the first few weeks of an infant's life.

However some 20 to 30 years ago reaching a diagnosis was a difficult task.

Patients could present with, or suffer from the effects of a single, or a multitude of complex and varied symptoms, abnormalities and malformations that were in some cases quite evident, or in others cloaked for a number of days, weeks, months, or, as in my case – years.

In June 2001 I was diagnosed with Pallister-Hall Syndrome (PHS); I was 35 years of age.

What is PHS?

Pallister-Hall Syndrome (PHS) is an extremely rare genetic disorder that may be apparent at birth; it is a multiple congenital anomaly syndrome, not a disease.

PHS follows a pattern of genetic inheritance in families called autosomal dominant inheritance.

The disorder can be both sporadic or inherited.

At this time (February 2002) there are approximately only 100 known diagnosed cases of PHS worldwide.

I am only the second individual diagnosed with PHS in Australia.

What causes PHS?

Pallister-Hall Syndrome (PHS) is a genetically transferable (inheritable) disorder that is caused by an alteration (or mutation) to the gene GL13 (glee three) which is located on the short arm of the seventh (7^{th}) chromosome in the region known as 7p13.

Symptoms and Features

Those symptoms, findings, abnormalities, malformations and features associated with, and

peculiar to Pallister-Hall Syndrome (PHS) can vary greatly in both range and severity from patient to patient and can affect individuals to different and varying degrees.

The central and most significant feature of any diagnosis of PHS is the presence of a Hypothalamic Hamartoblastoma (HH) which may, or may not cause any health problems.

A HH is a benign, non-cancerous malformation, or brain tumor of the, or in the Hypothalamic region of the brain that can cause deficiencies in one or more hormones, seizures and precocious puberty.

Some of the other prominent features and conditions peculiar to PHS include: the presence of a "Y"-shaped metacarpal or metatarsal bone and extra fingers and/or toes; syndactyly; polydactyly (both central and postaxial); bifid or cleft epiglottis; imperforate anus; micropenis; pituitary dysfunction and craniofacial malformations.

Secondary features are both numerous, complex and varied.

Who Can Get PHS?

Although statistically males seem to be more affected than females, Pallister-Hall Syndrome (PHS) is not specific to either gender, nor panethic, because the gene responsible for the disorder is found on chromosome 7.

PHS is found in persons of all nations.

Treatment for PHS

Generally, there is no specific treatment for Pallister-Hall Syndrome (PHS) in its entirety because it is an extremely rare multiple congenital anomaly syndrome.

However, some of those characteristic symptoms, conditions, abnormalities or malformations peculiar to PHS can be treated individually by hormone supplements and/or replacements, by surgery, or by other means.

In all cases though, those diagnosed with PHS should have periodical examinations in order to monitor the state, that is the size, condition and/or composition of the Hypothalamic Hamartoblastoma.

Periodical monitoring should be by way of Magnetic Resonance Imaging (MRI).

Children and PHS

Those couples contemplating having children obviously would be concerned about passing the disorder onto their off-spring.

Where one parent is affected by Pallister-Hall Syndrome (PHS) there is a fifty-per-cent (50%), or 50/50 chance that the child could inherit the chromosome that does not contain an altered GL13 gene.

There is no evidence to suggest that the severity of inheritance is lessened when a parent is "mildly" affected.

Further Information

To find out more about Pallister-Hall Syndrome (PHS): its causes; symptoms; abnormalities; malformations; findings; diagnosis; treatment and for a full explanation of difficult words, terms and abbreviations specific to PHS, go to my website at:http://www.geocities.com/ghelon/pallister-hall.html

Footnote

Between December 3 to 7 of last year I attended at the National Institutes of Health (NIH) National Human Genome Research Institute (NHGRI), Bethesda, Maryland in the United States of America.

The NIH is one of the world's foremost facilities for biomedical research.

After detailed work-ups, evaluations and testing my diagnosis was confirmed by Dr Leslie G. Biesecker, M.D. who is deemed the world's leading authority on PHS.

Dr Biesecker is a Certified Clinical Geneticist and Pediatrician.

Subcutaneous Immunoglobulin Therapy in Primary Antibody Deficiencies

The following article was written by:

Dr Melanie Wong MBBS FRACP
FRCPA, Staff Specialist,
Department of Immunology and Infectious Diseases
The Children's Hospital
Hawkesbury Road, Westmead, NSW 2145

Subcutaneous immunoglobulin is a well recognised and for the most part, well tolerated, alternative to receiving immunoglobulin infusions intravenously. It has distinct advantages but also disadvantages and decisions to undertake therapy must be individualised since it is more suitable to some patients and their families than others.

We established a subcutaneous immunoglobulin infusion programme at The Royal Alexandra Hospital for Children (now known as The Children's Hospital at Westmead) in early 1994 after an inspirational visit by Swedish immunologist, Dr Lennart Hammarström, an early proponent of this therapy. However, it would not have occurred had it not been for the enthusiastic encouragement by one of our families who had 2 sons affected by X-linked agammaglobulinaemia. They were tired and frustrated by the monthly trips to hospital, the time consequently lost from school and work, and the lifestyle limitations imposed by the need to plan around these monthly appointments, particularly with respect to camping excursions, holidays and overseas trips. They wanted a safe treatment that could be administered at home.

At that time, subcutaneous immunoglobulin therapy was already the treatment of choice for outpatient antibody replacement in many European, especially Scandinavian, countries. There were publications dating back to the early 1980's of successful treatment of individuals, but in the mid 1990's onwards there have been a number of larger studies directly comparing this to traditional intravenous therapy with excellent results with respect to efficacy, safety and patient/family satisfaction.

The process

With donated funds, we purchased simple syringe drivers (Graseby MS16A) that were then fitted with syringes to a maximum of 20mls and a very fine 25-gauge butterfly needle. The intramuscular preparation of immunoglobulin was used (16% vs 6% solution) and the dose over a month was equivalent to the monthly IVIG dose. Infusions were given 2 to 3 times a week to minimise the volumes delivered (usually 10 to 15mls) on any infusion.

Patients and care-givers were trained to set up and administer treatment and close contact maintained until they were comfortable with the process. Depending on proximity and other factors, this was either done as an outpatient over several visits or during a short admission. The immunoglobulin is infused into the subcutaneous tissue (layer between skin and muscle which is mostly fat) of the abdominal wall. Infusion sites are rotated around the abdomen. Some centres also use the thigh. Local anaesthetic patches (Emla) may be applied 30 minutes prior but are not essential and often abandoned once the child became familiar with the process because discomfort on insertion of the butterfly needle is minimal. Infusions were commenced at 5ml per hour but rates were gradually increased as tolerated until 10, 15 or 20ml infusions were given in less than an hour. There was flexibility in the timing and amounts of infusions as long as a set weekly dose was maintained. The pump can be strapped unobtrusively to the body in a pouch that allows total mobility during the infusion.

Safety

The most difficult part of initiating therapy was local irritation as the body became accustomed to this new route of administration. Swelling, redness, and pain or discomfort was not uncommon in the first few months but invariably resolved with perseverance, reducing in severity with ongoing infusions. We found that using the immunoglobulin cold from the fridge and local application of ice-packs was very effective in reducing discomfort. Occasionally paracetamol was also necessary. We also found that the children (and probably adults) who experienced least problems were those with more subcutaneous tissue, although several of our children were slim young boys who did very well after the initial settling in period.

The intramuscular immunoglobulin preparation has been shown over many decades to be safe. In contrast to some intravenous preparations, there has never been a report of associated viral transmission. This is probably because of differences in the manufacturing process.

We have not seen any significant acute side effects such as 'anaphylactoid' reactions that may present as fever, rash, headache or even collapse. Nor are they reported in any of the published papers. Such reactions were well recognised with intravenous immunoglobulin, especially older preparations. The difference may be due to the tissues acting as a filtering mechanism for immune complexes which would otherwise have gone straight into the circulation and/or the absence of a huge systemic load of antibodies, but rather a constantly replenished therapeutic level. The latter is supported by the fact that we see more adverse reactions with higher doses of intravenous immunoglobulin and that slowing the rate often prevents or reduces such reactions.

We and others have not experienced problems with infections at the sites of infusion.

Efficacy

We have found that there is a noticeably improvement in overall well-being, and the rate and severity of infections, with particular relevance to the 'trough' often experienced in the week prior to monthly doses of intravenous immunoglobulin. As in recent reports, we have found the maintenance of much higher 'trough' levels of antibody that supports our clinical observations.

Until recently, we have had few problems with the supply of the intramuscular immunoglobulin preparation, unlike the more frequent problems with the intravenous preparation. Supplies have been collected on a monthly basis and stored until use in the refrigerator. There have however been occasional shortages recently which may become an issue if subcutaneous infusions become more prevalent or if there is a change in policy dictating the production of this preparation by CSL. However, if the intramuscular preparation is not available, the intravenous preparation could be given either intravenously for the month or subcutaneously at volumes delivering the equivalent amount of antibody.

Patient and parental perception

As for any therapy, we have seen both successes and failures, thankfully more of the former. Significant commitment is required to overcome the initial settling in period and so not surprisingly the more motived families are more likely to succeed. They have reported dramatic changes to lifestyle and freedom from the rigidity of hospital tied therapy. Some have taken their infusions away on camp and others have taken their immunoglobulin, pump, syringes and needle (with adequate letters and documentation!) overseas for months at a time. Some children prefer to have their infusion quietly whilst doing homework or watch television, others go riding their bike or other more boisterous activities. Adolescents have found that taking control and ownership of their infusions has significantly increased their self esteem and complemented their need to achieve independence and the appearance of normality. It has reduced time taken off school and work (for parents) since infusions are performed at times that suit them.

Reasons to consider subcutaneous immunoglobulin infusions

- Psychosocial – as mentioned above, geographic separation from treatment centres
- Poor venous access
- Serious reactions to intravenous immunoglobulin therapy
- Where poor therapeutic antibody levels are achieved despite high dose and frequent intravenous infusions, leading to poor infection control
- IgA antibodies leading to reactions to intravenous immunoglobulin (subcutaneous infusions have been shown to be well tolerated in these people, often with proven reduction in the level of these anti-IgA antibodies with ongoing subcutaneous therapy)

It is important to discuss the pros and cons of this treatment with respect to the individual family's

needs and resources. What is an easy process for some, may be difficult for others. If the child is young and insertion of the butterfly is a stressful exercise, not improving over a period of time, there is little benefit in persisting. For some, the controlled safe environment of the hospital is a much more reassuring place to have complicated treatment. Again, subcutaneous therapy may not suit these families.

Commencing a trial of subcutaneous immunoglobulin treatment does not commit the child to continuing if this form of therapy proves unsuitable for the child or family. It also does not negate a subsequent trial if circumstances change, especially as the child gets older. Some children and their families benefit from periods of 'rest' from the pressure of frequent administration at home and return periodically to monthly intravenous infusions in hospital. Flexibility remains a key component of this programme and it's availability advances our care of adults and children with antibody deficiencies.

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(This article was first published in the Australian Primary Immunodeficiency Association (APIA) Newsletter, Issue 3, December 2001.)

FLAWED GENE FOUND FOR RARE TYPE OF SMA

Scientists have identified the defective gene that causes a rare form of spinal muscular atrophy (SMA) – a discovery that's expected to improve diagnosis and yield further insight into the disease.

SMA refers to a group of diseases that attack the muscle-controlling nerve cells (motor neurons) in the spinal cord, leading to muscle wasting. The most common form of the disease is linked to defects in the SMN1 gene, which encodes the survival motor neuron (SMN) protein. SMN deficiency tends to cause weakness that begins in the trunk muscles and progresses to include the respiratory (breathing) muscles.

A rare form of the disease called SMA with respiratory distress type 1 (SMARD1) causes pronounced weakness in the extremities and early respiratory problems.

In the August issue of Nature Genetics, a research team based at Humboldt University in Berlin reported linking SMARD1 to the gene encoding immunoglobulin mu-binding protein2 (IGHMBP2). Interestingly, IGHMBP2 appears to have a similar function to that of SMN, which regulates the processing of RNA (the chemical step between genes and proteins). Thus, the researchers suggest that further studies of IGHMBP2 will help sort out the disease process in SMARDI and the more common form of SMA.

Reference: *In Touch*, the magazine of the Muscular Dystrophy Association of New Zealand, Volume 36, December 2001. Originally published in *Quest*, the magazine of the Muscular

Dystrophy Association of the United States, Volume 8, No 5, October 2001.

KURRAJONG EARLY INTERVENTION SERVICE (KEIS)
NEW BOOK - "ONE DAY AT A TIME"
All the families who tell their story are local families who have a child with a disability or developmental delay and all have attended KEIS. The book is a wonderful collection of stories as told by families. The children have a range of disabilities ranging from Autism, Down Syndrome, Cerebral Palsy and developmental delay. Some early stories provide an ongoing story as the children progressed from birth to transition to school. The book is priced at $22.00. Please contact KEIS on: Tel (02) 6926 2466 or Fax 902) 6926 2820

CONFERENCES

ADULTS WITH ARTHROGRYPOSIS

Saturday 2nd March 2002

This informal lunch is an opportunity to network and to see how many people with Arthrogryposis there are living in Melbourne and even Australia. 12.00 Midday at Zagame's Caulfield Club Hotel, Cnr Dandenong Road and Derby Road, Caulfield East 3145. RSVP by 22nd February 2002. John McKenna: 03 9963 3989 (bh)Mob: 0419 877 712 Email: john.mckenna@team.telstra.com Lina Pane-Hawkins: 03 9418 0455 (bh) Mob: 0408 398 750 Email: lpane@paraquad.asn.au

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"MAKING IT POSSIBLE"

National Info Expo

Wednesday, 20th March, 2002
Presented by the Association for Children with a Disability.
Melbourne Park Function Centre.
Information, Speakers, Workshops, Displays. Entry is FREE for families. $70 for professionals & service providers.
Info: (03) 9500 1232 or 1800 654 013

One Day Symposium:

"NEW GENES ON THE X CHROMOSOME AFFECTING MENTAL DEVELOPMENT"
Saturday 23rd March, 2002
Speakers: Professor Hilger Ropers, Dr Josef Getz

Lecture Theatre, John Hunter Hospital, Newcastle.Info: Professor Gillian Turner or Margaret Jobber (Tel): 02/4985 3131 (Fax) 02/4985 3133. Registration Closes: 1st March, 2002

GENETIC SUPPORT NETWORK OF VICTORIA CONFERENCE for Support Groups

Friday 3rd and Saturday 4th May, 2002

Info: Eilis or Caroline (03) 8341 6315

9TH NATIONAL MPS CONFERENCE "SHARING OUR STORY"

Friday 12th April to Sunday 14th April, 2002
Rydges Resort, Eagle Hawk Hill, Canberra
The conference theme "Sharing Our Story" encourages us to share our own unique story of the effect of MPS or related disorder on our life. If you are a newly diagnosed family, if you have recently lost your child with MPS or you are a grandparent, aunt or uncle of a child with MPS; if you yourself have MPS or you work as a professional with families with MPS – there is something in the conference program for you. The conference program includes updates on research, news of current studies and information on the management of these disorders. The keynote speaker will again be Dr Ed Wraith from Manchester, UK.
For registration forms contact the MPS National Office: Tel (02) 9476 8411 Fax (02) 9476 8422

8TH AUSTRALASIAN PRADER-WILLI SYNDROME CONFERENCE
"Possibilities Not Disabilities"

Saturday 27th – Sunday 28th April, 2002
Carlton Crest Hotel, 169 – 179 Thomas St, Sydney. Info: PWS Conference Secretary
Tel/Fax: **9624 13 49**.

The theme of this conference is PWS – Possibilities not Disabilities. A program has been prepared that reflects the various possibilities in the support and management of PWS that are now available in the 21st century.
The conference is not just for health professionals or other service providers, but is most definitely focused on families and carers who provide the primary lifestyle support for all those with PWS. The conference includes lectures, workshops (e.g "Workshop for Fathers – how to be a 'real Aussie bloke' when a child doesn't fit the mould"), panels and stalls.
The keynote speaker will be Dr Tony Holland, M.D., who is a senior academic in the Department of Psychiatry at the University of Cambridge, U.K. He is particularly interested in behavioural aspects of PWS, and has given presentations at International Prader-Willi Conferences since their inception.

Professional Development Course

"CARING FOR PEOPLE WHO HAVE DEMENTIA & ALZHEIMER'S DISEASE"

Thursday, 4th April, 2002

The aim of this session is to promote understanding and knowledge associated with caring for a person with Dementia and/or Alzheimer's Disease and the implications of the disease.
Parramatta Leagues Club, 15 O'Connell St, Parramatta. Time: 9.30am – 3.30pm. Cost: $132
Info: Educational & Behavioural Consultancy
Tel: (08) 8288 7511 Fax: (08) 8288 7522
Email: ebc@primus.com.au

AUSTRALIAN HUNTINGTON DISEASE ASSOCIATION (INC) WA

FIRST NATIONAL CONGRESS
April 18, 2002

A unique opportunity for members of the West Australian Huntington's community to hear international, interstate and local keynote speakers talk on different aspects of Huntington's including: behaviour, care, communication, and Pre-implantation genetic diagnosis.
Info: Tel: (08) 388 3200
Email: huntingt@cygnus.uwa.edu.au
Website: www.ahda.asn.au

IDEAS INC

EXPO IN ALBURY

Friday 24th and Saturday 25th May, 2002

IDEAS Inc is holding EXPO 2002 to showcase disability products and information to the southern rural community of NSW. There will be 50 exhibitors that will have products/services on display including the NSW Anti Discrimination Board, Community Services Commission and the Down Syndrome and Brain Injury Associations.
Albury Convention and Performing Arts Centre
Info: 1800 029 904 Email: ideas@ideas.org.au

AUSTRALIAN LEUKODYSTROPHY SUPPORT GROUP

"IMPACT OF LONG TERM ILLNESS ON FAMILY DYNAMICS"

Lecture presented by Nadia Miocevic.
Sydney: 7th March, 2002, 7 – 9pm.

Info: Nada Miocevic Tel/Fax (03) 9352 6301.
Email: miocevic@alphalink.com.au.
Anna Capra: (02) 9744 8409.

Brisbane: 12th March, 2002, 7 – 9pm.

Info: Anne Keenan: (07) 3395 4398 Mob: 0417 788 775. Vanessa Eilert: (07) 3279 6250. Email: vanessaeil@hotmail.com. There are a myriad of issues that long term illness brings to the individual and family. Nada Miocevic, a social worker with more than 30 years of experience, will address the issues around the impact of long term illness on the individual, family and wider systems in her lecture. Open to anyone affected by long term illness.

HIPPOCRATES & SOCRATES V11

"DISABILITY AND DIVERSITY: SUCCESSFUL LIVING"

15th and 16th March, 2002
Sponsored by CHERI and CDDS
Children's Hospital Westmead
Information: Leslee Edwards: Tel (02) 9845 3017 or FAX (02) 9845 3082
Email: LesleeE@chw.edu.au

The Children's Hospital at Westmead Education Research Institute (Cheri) will be holding their Hippocrates & Socrates V11 conference - Disability & Diversity - Successful Living. The Keynote Speaker to the conference is Professor Barry Carpenter, OBE of the Sunfield School, UK.

Barry was until late 1997 the Chief Executive and Principal of Sunfield, a residential school for children between the ages of 8 and 19 years with severe and complex learning needs. Many of the children have challenging behaviours and a high percentage demonstrate severe autistic behaviour.

In the next two years Barry will be chairing a National Inquiry into the Mental Health needs of young people with learning difficulties. In this role he will represent the UK and the European Union Working Group on Early Intervention and and represent Europe on the World Coucil of Early Intervention. Most importantly Barry is the father of a child with severe learning difficulties and he has extensively written on families who support a member with a life long disability.

Conference themes will cover the following

1 Health and disability
2 Quality of life
3 Self and disabilities
4 Families and disabilities
5 Issues for Agencies and Practices
6 Philosophy
7 Good practice models /interventions that work
8 Living with disability
9 Communication and language

This will be a very exciting conference and it will bring together a diverse group of people.

GENETIC ALLIANCE EVENTS CALENDAR (USA)

March 7-8: Texas Birth Defects Conference 2002
Radisson Plaza Hotel, Fort Worth, TX. For information, call Amy Case at 512-458-7232, e-mail amy.case@tdh.state.tx.us or visit the website.

March 14-17: The American College of Medical Genetics Annual Clinical Genetics Meeting
Hyatt Regency, New Orleans, LA. For information, call 856-423-7222, ext. 360.

April 5-7: International Parent-To-Parent Conference 2002
Philadelphia Marriott, Philadelphia, PA. For information, call 717-731-6770.

CLIMB CONFERENCES 2002 (UK)

Children Living with Inherited Metabolic Diseases (CLIMB).
Help us celebrate and commemorate successful diagnosis and management of Metabolic Diseases, in this our 21st Anniversary year. This year we have a dynamic programme of exciting and interesting topics backed by some excellent speakers from medicine, industry and care services as well as from first-hand experience as patients or parents. The first conference is in the

Hilton Hotel, Newbury on **Saturday 11th May 2002**. The second conference will be in the Britannia Hotel, Newcastle upon Tyne (by the airport) on **Saturday 7th September 2002**. As usual, prices have been kept as low as possible. Please put the dates in your diary now and contact us (0870 7700 325) for a registration form as soon as possible. We look forward to meeting you in May and/or September.

LAST CHANCE TO HAVE YOUR SAY!!!

INQUIRY INTO PROTECTION OF HUMAN GENETIC INFORMATION

What is the enquiry?
The Federal Government has asked the Australian Law Reform Commission and the Australian Health Ethics Committee to conduct an inquiry into the protection of human genetic information. It is considering whether any changes to the law are needed to:

- Protect the privacy of individuals
- Prevent discrimination
- Ensure high ethical standards of conduct

Have your say!
AGSA is currently writing a submission on behalf of members and we ask that you send in your concerns and experiences for inclusion. Issues to consider include:

- Privacy
- Insurance
- Employment
- Education
- Medical Research

If possible, please send your responses via fax or e-mail, as the deadline is rapidly approaching! Please send in your comments by **Wednesday 13th March 2002**
Fax: (02) 9211 8077
Email: agsa@ozemail.com.au

You can also make your own submissions to the Australian Law Reform Commission.
The Australian Law Reform Commission
Level 10, 131 York Street
SYDNEY NSW 2000
Tel (02) 9284 6333 Fax (02) 9284 6363
For a free copy of the ALRC/AHEC issues paper, complete and return the "**Fax Back Form**" included in our newsletter.

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