THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

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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

The launch of Genetic Disorders Awareness Week on 20th June 2001 was a memorable night with an excellent turnout.

We are indebted to our speakers, Professor Bruce Dowton, Dr Michael Buckley, Dr Andrew Wilson, Lauren Gatt, Daren Fisher and Kelly Fisher and our Chairperson, Professor John Dwyer, the combined talents of whom made it a very special evening. Certainly the major outcome of the launch meeting was that with any genetic condition the person with the condition becomes the primary focus of attention. However the theme *Genetics and the Family* showed how differently the family is viewed if looked at holistically.

I would like to thank you for attending such a special evening. Without the sponsorship of the Powerhouse Museum, Transkaryotic Therapies and Sporte Leisure the evening would not have been possible. Our thanks also must go to Maggie Lanham of Lanham Public Relations for generating publicity during the week.

AGSA is presently looking at running a sibling workshop with a prominent overseas speaker, a Breast Cancer seminar, Triple X support group meeting and a rare chromosome meeting.

I have been invited to be one of the consumer representatives for the National Approach to Genetic Services in Cairns prior to the Human Genetics Scientific Meeting. If you have any views you would like me to express on your behalf please let me know as soon as possible.

AGSA's AGM will be upon us soon so if you are interested in becoming a committee member please give me a ring to discuss what is involved and the projects AGSA is working on.

I look forward to hearing from you.

DIANNE PETRIE

Support Group News as at 11th June 2001

Cumberland Newspapers has 17 newspaper titles from Campbelltown to Lake Macquarie. If you want to publicise a meeting, seminar or raise awareness Call Bev Jordan, Group Reporter, on Ph: (02) 9894 4797 or Fax (02) 9689 5353.

South Easterly – The Newsletter for South East Health, April 2001

On 14th March 2001, The Hon. Bob Carr MP, Premier NSW, officially opened the Sydney Children's Hospital Research Complex. The Research Laboratory comprises the Westfield Research Laboratory and the Strasser Research Laboratory. The newly refurbished laboratories provide the most modern facilities, where research can be conducted to enhance treatments for many childhood diseases and enable training of surgeons in the Surgical Skills Laboratory. The new facilities provide vital locations for clinicians and research staff to conduct cutting-edge research into paediatric specialties including immunology, genetics, respiratory disorders and neurological diseases.

Tourette syndrome – May 2001 newsletter

Tourette syndrome have a number of publications for sale. These include topics like Parenting a child with TS, Adult Life, Learning problems with TS Child. For a full list, contact the Association on (02) 9382 3726.

Centre for Developmental Disability Studies – April 2001 newsletter

CDDS website is: www.cdds.med.usyd.edu.au/ The Centre is collaborating with colleagues from the Universities of NSW, Macquarie and Sydney and College of Health Sciences in developing a course proposal in the Graduate Program for Developmental Disability. CDDS and Children's Hospital, Westmead, are doing a study on preventative health care in children with cerebral palsy and children with Down syndrome, aged 1-4 years who live in Western Sydney.

CanDO - Community Services Commission newsletter – Autumn 2001

The Community Services Commission and the Intellectual Disability Council of NSW have released a discussion paper: *Crime prevention in residential services for people with disabilities*. There are other reports available from their website - www.csc.nsw.gov.au

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.

BEHCET'S SYNDROME

Contact and information is sought for a 23 year old male with this condition. Please contact AGSA for details.

EMERY-DREIFUS (DREYFUS) MUSCULAR DYSTROPHY Autosomal Dominant

A 36 year old woman living in NSW would like contact with anyone who has had experience with this condition. Please contact AGSA for details.

PALLISTER-HALL SYNDROME

AGSA now knows of two adult males with this condition seeking contact with others – if you know of others please let AGSA know.

WOLFF-PARKINSON WHITE SYNDROME

A newly diagnosed 36 year old man would like contact and information from others – please contact AGSA for details.

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.

Reprinted with kind permission from ***Children Living with Inherited Metabolic Diseases (CLIMB) Newsletter Volume l, No. 5 March 2001.***

Professional News

DIAGNOSIS OF RARE DISORDERS:

Professor Bryan Winchester

Many parents express frustration at the length of time that can be spent on reaching a diagnosis in their child. In his talk on the diagnosis of rare disorders, Professor Winchester explained the processes involved in obtaining a diagnosis and the additional complexities when it is realized that doctors are dealing with a rare disease. Bryan describes the biochemists as the "backroom boys" (or increasingly girls!). His presentation in fact clearly demonstrates how they are at the forefront of the whole process.

Rare diseases, according to the European Union definition are those with a prevalence (that is, the total number affected by the disorder in a given population at any one time) of less than 5 per 10,000. While individually rare, collectively these conditions are numerous (some 5000 in total) and account for a significant public health problem.

Doctors recognize that rapid and correct diagnosis is important because 90% of rare diseases are genetically determined, often several organs are involved, and management can be complex. However, the low prevalence of these disorders does make rapid diagnosis difficult, as, in the first instance, common causes of the perceived symptoms must be ruled out and a number of steps made to progress towards diagnosis.

Family

The first point of reference is the family. Is the child showing strong physical characteristics suggestive of a genetic disorder rather than a family trait? Is the child failing to thrive? Is there recurrent infection? Is there delayed development? Is this the first affected child? Is there a high ethnic prevalence?

Positive answers to theses questions indicate a visit to the GP.

GP

The GP will treat fever and infection, conduct simple urine tests, and, as indicated, request preliminary screens from the local chemical pathology laboratory. Dependent on the results of these actions, the child will be referred to the paediatrician at the local District General Hospital.

Local Paediatrician

The local paediatrician will carry out a clinical (physical) examination and will order certain tests to be under taken by the local hospital laboratory, including blood electrolytes, sugar, liver function, thyroid, chromosome analysis and scans. If necessary, the paediatrician will also request tests from specialist laboratories. In perhaps 30-40% of cases. A diagnosis will be made at this stage and management started. Where this is not possible, the child will be referred to a tertiary center- that is, a hospital with access to specialists and researchers and

attached to a university with specialist facilities and experts.

Tertiary Children's Hospital

At the tertiary hospital, the child would normally be seen by a paediatrician who has specialized in a particular area of medicine such as nephrology (kidneys) hepatology (liver) or metabolism (body chemistry) depending on the suspected problem.

Another physical examination will be carried out and management discussed. Further laboratory tests will be ordered based on the clinical indication and results of the first tests. Sometimes a biopsy is needed (a tiny sliver of skin which is grown in the lab and then examined for significant changes that indicate a particular condition). These, together with blood samples will be sent to a specialist laboratory, sometimes overseas.

Specialist Laboratories

These laboratories deal with complex screening involving biochemistry and molecular genetics. They are attached to a medical school (university) and specialize in a particular group of disorders, there being a small number in each field in the UK. For this reason, it is essential to accumulate experience in rare disorders, both of staff and samples. If, for example, the laboratory is attempting to diagnose a disease with a prevalence of l in 100,000 like Pompe's disease (GDS2) there will only be 7/8 cases a year in the UK and on average, the local district general hospital will never see a case!

The Role and Importance of DNA analysis

Many diseases are due to a fault in a single gene (about 400 per 100,000 live births). Many of these genes have now been cloned (copies) and the mutations (the changes in the gene that cause the problem) can be found in both the patient and the parents. This means that DNA analysis can provide a complete diagnosis for the patient and a family specific test.

It may also be possible to predict the course of the disease which will be relevant for planning patient management and the needs of the whole family. Sometimes the genetic disease and the symptoms it causes have a predictable pattern (genotype and phenotype correlation). DNA analysis can help in the selection of appropriate patients for new forms of treatment such as enzyme replacement or gene therapy and it is relevant for accurate carrier testing, genetic counseling and increasingly for prenatal diagnosis.

Carrier Testing

This is particularly important for X-linked (mother to son) disorders such as ALD and muscular dystrophy, and for populations already identified as being at high risk of expressing a particular disease such as Cystic Fibrosis, Tay Sachs and Sickle Cell.

Prenatal Diagnosis

Specialist laboratories play a vital role in analyzing the results of chorionic villus samples taken at around 11 weeks gestation or following amniocentesis at 12/15 weeks. It is essential to have experienced specialists to carry out analysis of chromosomes, DNA, morphology of cells and enzyme and protein determination.

Newborn Screening

Newborn screening is important because early diagnosis of a disease can lead to effective treatment of the condition to prevent the onset of symptoms. Currently, there are only two conditions that are screened for nationally from a bloodspot taken from the heel of the newborn baby. In certain areas, the haemoglobinopathies (sickle cell and thalassaemia) are also screened. There is currently much debate about extending newborn screening to other conditions, because new diagnostic technology called tandem mass spectrometry would make this possible. The UK National Screening Committee (1998) has outlined 17 criteria that should ideally be met before each new condition is added to the national screening programme. There are four categories to consider:

- The condition – what is known about it and its natural history?
- The test – is it reliable? Are there false positives/negatives possible?

- The treatment – is there a proven treatment if early diagnosis is achieved?
- The screening programme – can this be effectively administered? How are parents informed? Who owns the data? Where is it stored?

For many rare genetic diseases it will not be possible to meet these criteria and an alternative, more workable approach will be needed.

Technical Advances

As already mentioned, there have been tremendous technical advances in diagnostics and screening within the last decade. However, these also throw up ethical dilemmas, such as the age at the which potential carriers should be tested; the use of mass spectrometry to screen for late-onset and incurable diseases and the use of the results of genetic screening to decide the level of insurance premiums.

The newest advance will be the DNA chip which can carry all the known mutations for a disease gene such as cystic fibrosis and BRAC1breast cancer on a thumb-nail size chip.

While all such advances are to be welcomed if they speed up potentially difficult diagnoses, there is concern that the technology, used without careful consideration of the ethical issues involved, will encourage a 'genetic underclass" and a "designer babies".

The Future: New Approaches for the Diagnosis of Rare Diseases

There still needs to be greater awareness amongst GPs and paediatricians of the potential for a rare disease within their patient population. NHS Direct Online, the CaF Directory and numerous patients' group websites will help here.

The population needs to be more educated about the effects of rare genetic diseases – after all, we are all potential victims of a genetic disorder. If the general public understands the issues, it will be encouraged, through charitable giving, to fund research.

Specialist laboratories need adequate funding and staff who are motivated by the value placed in their expertise and the training they are given, so that each centre of excellence can grow and develop to meet the needs of a populations with ever-increasing expectations of their capabilities.

The ethical issues around consent in order to take and retain samples for analysis, research and future diagnosis need to be addressed to the satisfaction of parent and patients and doctors and researchers. Both parties need clear guidelines so that neither harm is done nor research restricted because samples have been inappropriately taken, stored or used.

By the conclusion of Bryan's talk it was clear to us all that diagnosis of any condition requires skill and those involved in rare diseases display particular expertise and dedication.

Furthermore, the consensus at the meeting was that, given a clear and honest explanation, consent would not be withheld by parents if samples would benefit research into their child's condition.

Rare Diseases in Europe:

Anders Olauson, President Eurordis

Report by Pam Davis, from a transcript of a Conference 2000 address.

Anders opened his talk by saying the audience are his teachers

Argenska, in Sweden, began in 1989 in response to and on behalf of the families of children with rare disorders. The aim was to put down a holistic perspective, to look at any aspects of life. Many important people were invited from different perspectives to do with having a disabled child: chairmen of medical services, hospital, school, social services and Argenska. The Chairmen of The Organisation for Disabled Children stated that to understand, one has to be a parent of a disabled child.

Anders recognizes that professionals used different language but were fighting for the same thing. He also recognizes that although he cannot understand what it is like to be a parent of a disabled child, he up-dates his knowledge to understand how it is and to understand and believe what parents are saying. This is what Argenska is about.

The most important activity looked at was family activity. Family activity brings parents and families who have children with the same diagnosis, throughout Sweden, together to Argenska over one week, to meet all the expertise from their particular diagnosis and to bring parents together. There is a respite service and this is very important to bring relief for the parents but this also allows the youngster to train and to take responsibility for their lives, drugs etc…There is a project which has a structure and strategy for issuing information about education for every child with a disability which recognizes each child's possibilities. This means that teachers can use this information. There are also on-going research activities.

Parents say that they feel normal for the first time. Anders realizes the impact of families meeting their situation and swoop information. Children have a chance to meet other children with the same disorders. He has seen children who are infants who recognize others with the same sort of problems and relate to each other. Siblings will do anything to protect their family but there is also a need for them to be siblings as well. They need to express their feelings, even if that involves saying "I do not like my brother or sister" – It is a normal part of life to be able to do this. It does not mean that they do not love each other, they do. It is a normal process. It is very important that they share their experiences with others. Hospitals or other professionals must recognize that this is a normal process.

Anders discussed were or at what point parents should think about the future. This should be in the first second of realization. The attitude needs to be right from the start, to recognize that life can be good and to have a positive vision for the future. This will help parents a lot. Contact with other parents will help them with technical aids, financial support, by creating and spreading information and by starting family organizations. That is what Argenska is doing in Sweden.

Eurordis was created in France and Lesley Greene was involved with this from the beginning. It was created to implement the Orphan Drugs Regulation, to put the pressure on the pharmaceutical industry to have proper drugs, even if they were for very small numbers. It was also to create a link between all the associations that worked for rare disorders in different countries. Even if one person in any country may be suffering from a particular disorder, there should be treatment. Consequently, Eurordis is an umbrella organization for all associations that work with rare disorders. There are nearly two hundred member associations from fourteen countries, covering nearly tow thousand different rare disorders. Members are increasing and the organization is well known among Members of Parliament and patient groups, lobbying the cause.

Two seats have been confirmed in the Committee of Orphan Medicinal Products for Eurordis. This is the organization which makes decisions about the drugs that should be funded by the commission and adopted. We also make sure that there is a budget for this, David Bow is responsible for this.

The Frame for the Programme of Action on rare diseases creates a network of patients regarding medical products for rare disorders. There will

be financial alliances, development of a new communication strategy and the creation of an awareness day in the European Parliament in Brussels. This has been modeled on awareness days in England. American has had an orphan drugs regulation 15 years. Europe needs to fight for Eurordis to get over the barriers of so many different countries and borders.

In the future, Eurordis is planning to implement the first project accepted by the E.U. This is to set up the national alliances of rare disorders. The idea is to bring the national alliances together and to create national alliances within each country of Europe and to make them work together. The reason for this is because Eurordis believes that within each country, a link, an umbrella organisation has to be made because the natural culture and legislation is shared. From this point, the national alliances must be connected to help them to empower each other and to empower within each part of the country. Eurordis is proud to announce that the budget is ready for this and the initial workshop will soon commence.

Also in the future, Eurordis is planning to develop a European portal site for Rare Disorders part 11. It has taken a lot of discussion to create one European database for rare disorders. Difficulties have included cultural and language/translation problems. Adding one more language will cost $15 million and will take three years. Eurordis believes that there should be not one big mother database that there should different databases that are connected, enabling each patient to go to one site and then on to find the information that you require in your own situation.

The preparation of the third part of the project – a joint initiative of a European Conference on Rare Disorder and Disability in Copenhagen May 18th and 19th 2001 is underway and will be joined by The National Alliance in Denmark and also The General Assembly for Eurordis. It will feature talks on rare disorders encouraging the pharmaceutical industry and biotechnology to develop treatments for rare disorders.

We have to establish more links between patients, the pharmaceutical industry and biotechnology because they, like all of us are still learning. A key role must be developed within the COMP in the development of a policy for Orphan medical products in the E.U. a network of expertise in Europe must exist to link them together. There must be an active search for new partners and sponsors for funding. The funds from members alone will never be enough.

There is caution when approach and involving a pharmaceutical company because a certain sense of freedom is needed. One way to do this may be to create an awareness event, each year in Europe. An event will be held in each country, possibly at the same time to arise awareness and to collect money. For example, in France Telethon organized a twenty-four hour event which raised half a billion French francs.

Research needs to be promoted in all aspects of everyday life, which are related to diagnosis. Medication and drugs are only part of people's lives. Pressure needs to be placed on the Government and its legislation to improve all aspects of life in schools, social support and accessibility in the cities.

Eurordis, for the next four or five years will involve all the countries, share the best practice and learn from each other to create knowledge that can be shared by all. A common knowledge and common picture of every level of life, a common goal to fight for. Barriers and borders must be torn down so that any child whether from Greece, Iran, Spain or any country is recognized a being valuable and important, the children are the ones that we are fighting for.

Anders Olauson, President Eurordis

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BREAST CANCER

Breast cancer is one of approximately 200 different types of cancer. Cancer is a disease in which abnormal cell development occurs causing destruction of healthy cells. Breast cancer is the commonest form of cancer occurring in women

in Australia affecting about 1 in 12 before the age of 79. Men can also be affected but this is rare.

When detected early, breast cancer is treatable. In about 5% of the cases of breast cancer in women, the development of the cancer has been contributed to by the inheritance of a fault (mutated) gene.

When an individual inherits the mutated gene from a parent, she/he has a higher chance of developing breast cancer than average, i.e. is predisposed to developing breast cancer at some time in his/her lifetime. Mutations in several genes (called BRCA1 and BRCA2) have been identified as causative in the development of some cases of breast cancer. Mutations in these genes also predispose a woman to developing ovarian cancer. However it is clear that there are a number of other genes in which mutations will predispose to breast or ovarian cancer and research is continuing.

Familial cancer clinics have been established to provide information, counseling and some families, predictive genetic testing.

Reprinted with permission from the NSW Genetics Education Directory 2000/01.

A PERSONAL STORY

Genetic Testing: A Patient's Perspective.

I am immensely grateful for this opportunity to share the emotional impact that genetic testing has on the patient.

For me, genetic testing has been like a roller coaster of emotions. From the pre-testing counseling to the actual test, getting the results and the aftermath.

Due to my mum's death from ovarian cancer I had always expected to "just have my ovaries out" when I got older. It wasn't until I arrived at the Familial Cancer Clinic at Westmead Hospital that I discovered we also had a history of breast cancer and how extensive the cancer was.

When my family tree was laid out in front of me, it broke my heart. It was like looking at a cemetery with rows and rows of crosses signifying my deceased relatives. Ironically, that day was the first time I felt some hope that I may escape the same fate.

The test for my family's mutation was explained to me. Perhaps I would test negative. The fact that I have children made me desperate for a negative result, because I want to be here for them always but also that would mean I couldn't pass the gene onto them.

The pre testing counseling is quite involved. There are numerous questionnaires to fill out which require you to think deeply about your overall satisfaction with your life. All of this took place in the incredibly supportive and compassionate environment provided by the Familial Cancer Clinic at Westmead.

The night before the test was agonizing. I felt I was about to embark upon something that would change the course of my life regardless of the result. I didn't feel fear so much as a sense of foreboding – a sense that my life had been a series of journeys leading me toward this momentous event. Now that it was here, all I wanted to do was envelop myself in a comforting cloud of denial. I hated the thought I may have this horrible mutating gene, which could cause my body to declare mutiny and leave my husband and children alone. I was terrible afraid of the legacy I may have passed onto my children. My greatest fear was of history repeating itself – my children watching me suffer as my brother and I watched our mum. I found it impossible for me to separate my fate from my mum's. I felt that all the anguish she had suffered was being revisited on me. The day of the test itself was fraught with anxiety. Now that I was on the path to knowing my genetic status I just wanted the result. The idea of waiting six to eight weeks seemed excruciatingly long.

After the test, the long wait. At first, time went by quite quickly, then the closer I came to knowing, the slower time dragged on. I oscillated between feeling sure I would be spared then I would pitch into depths of negativity – believing there was no way I would escape my family's faulty gene. Then there was the constant anxiety. If I was given a negative result, would I believe them? Would it not be possible that I had another faulty gene that hadn't been found yet? In that sense, a positive result would almost be a relief.

The day of the results I felt an overwhelming desire to attend the clinic looking as feminine as possible. In my heart, I felt sure I would get a positive result and had already decided on preventative surgery. That day I wanted to received my results looking like a woman.

Thankfully, both the doctor and the genetic counsellor had told me there would be no small talk, they would just tell me my results. Once my husband and I had sat down, we were told it was bad news.

How do I begin to describe how I felt? I felt as though history was repeating itself. My first thought was the devastating possibility that I may have passed the gene onto my children. There is a tremendous feeling of guilt, almost shame associated with being the carrier of a faulty gene. I felt that my body was somehow "defective".

The positive result for me means I am a carrier of BRCA1. This means I have a significantly increased risk of breast and ovarian cancer. The options facing me were explained briefly at that visit, as both the doctor and the genetic counsellor were aware I was incapable of absorbing any information.

Later, they were clearer. Aggressive screening, participating in drug trials or preventative surgery. I don't want a lifetime of testing and I'm not interested in feeling like a guinea pig. I want to reduce my risk as much as possible. For me, surgery is the only option.

One of the things I have found most difficult is the lack of support for women like me. I don't have cancer, yet I do have a faulty gene predisposing me to cancer. There is no neat category for me.

For some time I felt quite isolated, alone and almost a novelty – despite the ongoing support and invaluable information from Westmead Hospital. Finally I found a website called F.O.R.C.E. (facing our risk of cancer empowered). Here I found immense love, understanding, compassion and support from women who I have never met and am unlikely to do so. I no longer feel so alone.

For me, knowing my genetic status is something to be grateful for. I have information and choices therefore I have power. I have some control over a situation that has, at other times, overwhelmed me and made me feel quite helpless.

Surgery may seem an extreme measure, especially as my risk will not then be at zero. However, I will do everything in my power to avoid the same fate as the women I have loved and lost – anything I can do to avoid my children watching me suffer. I am a very positive person and am grateful for the chance to make an informed choice.

The bottom line is that I want to live and I want to live without fear.

My sincere thanks and love to my family and friends, Westmead Hospital and the beautiful women at FORCE for your continued love and support.

CONFERENCES

Australian Leukodystrophy Support Group – International conference will be held at the Royal Children's Hospital, Victoria, 17th and 18th August.

Costello Syndrome Conference – Toronto Canada, August 2001. Contact Cath & Colin Stone, 90 Parkfield Rd, New Mosten. Manchester. M40.3RQ, England, email: C.A.Stone@mmu.ac.uk

The Chromosome 18 Registry & Research Society (Australia) 1st National Conference – 22nd-23rd September 2001 at Cronulla Sharks Leagues Club, Cronulla, NSW.
Two speakers from San Antonio, Texas, USA, have been invited to educate families and up-date them on Chromosome 18 research. They are Jannine Cody, who is the Founder and President of the Society & Dr Dan Hale,

Endocrinologist. They will also be speaking at Sydney Children's Hospital, Randwick and The Children's Hospital, Westmead on Chromosome 18 disorders and growth.
For more information regarding the conference or speakers, please ring (02) 9580-5707.

TURNER SYNDROME ASSOCIATION OF AUSTRALIA LTD
NATIONAL CONFERENCE - PERTH
6th – 7th October, 2001
CWA, 1174 Hay Street, West Perth

Speakers -
Dr Geoff Byrne & Dr Fiona Frazer - Endocrinologists
Dr Andrew Bulloch - Cardiologist
Mr Max Keyt - In Vitro Fertilization
Ms Katrina Berkov - Motivational Speaker
Plus Perspectives Panel, Discussion Groups and Workshops

Approximate Costs -
Accommodation - Twin or Double with en suite - $75.00 per night, including breakfast
Single Standard Room (share bathroom) - $42.00 per night, including breakfast
Plus Registration Fee (to be advised)

For more information contact:
Mrs Glenn Fisher - National President
Email: <turnersyn@netpro.net.au>
Telephone: (02) 9452 4196 Fax: (02) 9975 4037
PO Box 112, Frenchs Forest 1640

The 3rd International Respite Care Conference for People with Disabilities. "Global challenge…Local Solutions" –
11th-14th September 2001
Masonic Centre, Sydney.
For information contact
Interchange NSW,
The Carrington Centre, Carrington Square, Campsie NSW 2194.
Ph: (02) 9789-1348 or
email: interchangensw@SO54.aone.net.au

Australasian Tuberous Sclerosis Society Annual Family Conference on 11th-12th August 2001, at Deaf and Blind Children Institute, North Rocks, Sydney. Contact (02) 9630-3147

Diabetes Australia-NSW is holding a 2day event, 'Diabetes Down Under 2001' on 18th & 19th August, at the Sydney Super Dome, Homebush Bay. For further enquiries contact Anne Maree Stratford, Health and Education Coordinator on (02) 9552-9965 or email: annemars@diabetesnsw.com.au. It aim to provide up-to-date information to people with diabetes and health professionals.

The 1st National Scientific Conference on Osteogenesis Imperfecta

Friday 27th July 2001
Centenary Lecture Theatre,Royal North Shore Hospital, St Leonards, Sydney.

Celebrating World Health Organisation Bone and Joint Decade 2000-2010
Proudly sponsored by: Novartis, Aventis, NOCA, The Australian and New Zealand Bone and Mineral Society of Australia and the Osteogenesis Imperfecta Society of Australia
Further enquiries: Mrs. Lyn Foxall,
OI Australia, PO Box 401,
Epping NSW 2121
Phone No. (02) 98691486
E-mail: petlyn@hotket.net.au

Parental Quality of Life Study

Between December 2000 and March 2001, parents of children suffering from a lysosomal storage disorder, took part in focus group discussions in Adelaide and Sydney as part of a study commissioned by Lysosomal Diseases Australia, and being conducted by the Research and Evaluation Unit at the Women's and Children's Hospital in Adelaide. Since the focus of research is usually on the affected child, little is known about the broader day-to-day quality of life of parents. Issues discussed included the emotional, financial and time impact of caring for an affected child.

The concerns raised during these discussions, and in interviews with medical professionals, will be formulated into a questionnaire that will be used to assess the health-related quality of life of parents caring for a child with a lysosomal storage disorder in Australia and the United Kingdom. The results of this questionnaire will then be analysed and communicated to government bodies and policy makers to help improve the quality of life of parents. We would like to take this opportunity to thank all of the parents who have participated thus far, for sharing your breadth of experiences with us.

Once the preparation of this questionnaire is complete, we will be contacting parents to request their participation in the next stage of the study. We envisage implementing the questionnaire in late-2001. If you are a parent of a child with a lysosomal storage disorder and would like to take part or would like more information, please contact the study coordinator, Ms.

Leanne Whaites (tel: (08) 8161.6913, or email whaitesl@wch.sa.gov.au). At the completion of the parental component of the study, we intend to develop a series of modules that focus on other family members such as the siblings of an affected child.

Lauren's Talk for Genetic Disorders Awareness Week Launch at the Powerhouse House Museum on 20th June 2001

Hi. My name is Lauren and I am talking to you this evening because I am a bit different. Most girls my age think being different is having their navel pierced or getting a tattoo somewhere that their mother wouldn't approve of. I have Turner Syndrome.

Turner Syndrome is a rare genetic variation that affects about 1 in 2500 girls. It means that some or all of the cells in the body, are missing some or all of the information on the second X chromosome. In my case all of the second X chromosome is missing in each of my cells. I only have 45 chromosomes instead of the regular 46.

So what does all this mean? You probably think I look and sound pretty much like every other little girl on the planet. If I tell you that I am going into high school next year, you might realise that I am pretty small for an eleven-year-old. That is because of the Turners.

Most, but not all Turner women are a bit on the short side with an average height of around 145-cm, (that's about four foot nine inches.) I have been on the growth program with the Children's Hospital since I was four. I have a needle every night containing a synthetic form of human growth hormone. Usually my mother does it, although sometimes I do it myself. My endocrinologist predicts that I will be about 5 foot when I finish growing, which is not as tall as the rest of my family, but at least I can look forward to being a bit taller than my Nanna.

Most Turner women have problems with having children. Fortunately these days, IVF has made it possible for women with Turners, to have children. For many Turner women, in the past, infertility was the hardest part about having Turner Syndrome.

It is not something that I worry too much about right now, not just because I am still young, but because I know that when I grow up it is possible that I will be able to get this help when I decide that I might want to have children. First all I have to do is stop thinking that all boys are revolting.

At my age, the worst thing about being a bit different is the teasing I get. Kids always come up to me in the playground and stand over me to show how much taller they are, and of course they call me "shorty" and "puny" and other things that must have really stretched their imaginations. Once a boy even bent my fingers back. To teach him a few manners I hung one on him. He still hates me, but at least he has a reason now.
Because Turner Syndrome is relatively rare, some doctors and even some text books still give inaccurate descriptions of what the Syndrome involves.

When I was born I had puffy hands and feet but I was an otherwise healthy baby. The paediatrician was smart enough to know that the fluid in my hands and feet meant I had Turners, but not smart enough to avoid telling my Mum and Dad that I would probably be retarded.

When I started talking at nine months and did all the things that other babies were doing, my Mum finally contacted the Turner Syndrome Association and Glenn Fisher gave her some correct information in books and on videos. Although she felt happy that I was going to be all right, she was pretty angry that she had wasted all that time being so upset. Mum has been in the Turner Syndrome Association of Australia for 11 years now, and for me it means that I know some other girls with Turners too. We write and email each other and sometimes visit for sleepovers. We always end up talking all night. It is

really good to know someone who is having the same problems that you are.
Today, I am in extension groups for Maths and I do well in the rest of my studies. Even though I am one of the youngest in my year I always come in the top ten in my class. I play Piano and Flute and I am in the school concert band and the school choir. I really like sports and I play softball for my school and was recently a district representative for cross country. I am a peer support leader in my school and I really like being with the younger children at school.

As far as I am concerned, I may have Turner Syndrome, but I am not all that different. I still do all the things that everyone else does, in fact most of the time I try just a bit harder.
These days most of my friends know about my Turner Syndrome and they don't care either.

Most of the problems associated with Turner Syndrome can be managed with a little bit of help from doctors, a bit of love from our families and support from groups like the Turner Syndrome Association of Australia. Thank you for listening to my story.