



Gene Advocate

Issue 60

2010

Preimplantation Genetic Diagnosis

While there are a number of different prenatal tests and procedures available to diagnose the development of a baby, another option where there is a risk that the baby will have a genetic condition is preimplantation genetic diagnosis (PGD).

Some Important points

- PGD involves testing for certain genetic conditions in an embryo created using assisted reproductive technologies (ART) such as in vitro fertilisation (IVF), prior to transferring it to the uterus and allowing it to develop normally
- After hormonal stimulation of the woman's ovaries, some eggs are removed and then fertilised in the laboratory with sperm
- One to two cells are removed from the embryo at the eight cell stage (after 3 days) or at blastocyst stage (after 5 days), for testing
- Only those embryos that do not have the specific genetic condition that was tested for will be transplanted into the woman's uterus
- Usually, no more than one or two embryos will be transferred to the uterus at any one time to avoid the possibility of multiple births (more than one baby in a pregnancy)
- Success rates for having a child from an IVF cycle followed by PGD varies from IVF centre to centre but tend to follow standard IVF success rates
- Like any IVF procedure, stress and often disappointment can accompany PGD. Couples will need to balance the financial and emotional burden of the IVF procedure

followed by PGD with that of termination of an affected child conceived naturally

- In Australia, PGD is currently only offered in the private setting

Every woman hopes for a healthy baby. In some cases, the baby may have either a serious physical or intellectual problem.

There are a number of different prenatal (meaning before birth) tests and procedures available to diagnose the development of the baby. Each has advantages, disadvantages and limitations.

Prenatal diagnostic tests include:

1. Ultrasound
2. Chorionic villus sampling (usually simply called CVS)
3. Amniocentesis
4. Cordocentesis

This article discusses a diagnostic pre-pregnancy option called preimplantation genetic diagnosis (PGD) that, with the use of in vitro fertilisation (IVF) therapy, is a diagnostic test performed on an embryo prior to implantation in the uterus.

What is Preimplantation Genetic Diagnosis (PGD)

Preimplantation genetic diagnosis (PGD) was first reported in 1989. It is a very specialised technique that can help couples who are at risk of having a child with a genetic condition avoid doing so without the need for decisions regarding termination of an affected pregnancy.

PGD involves testing an embryo that has been created using assisted reproductive technology

(ART) such as in vitro fertilisation (IVF), prior to transferring it to the uterus and allowing it to develop normally.

How is PGD performed?

Hormones are given to the woman to stimulate her ovaries and enable the collection of a number of eggs or oocytes.

- After the eggs are removed, the eggs are fertilised in the laboratory with sperm
- Those eggs that are successfully fertilised divide and multiply to form a developing embryo called a blastomere
- After three to five days, the developing embryo contains either about eight cells (after three days) or is a blastocyst (after five days)

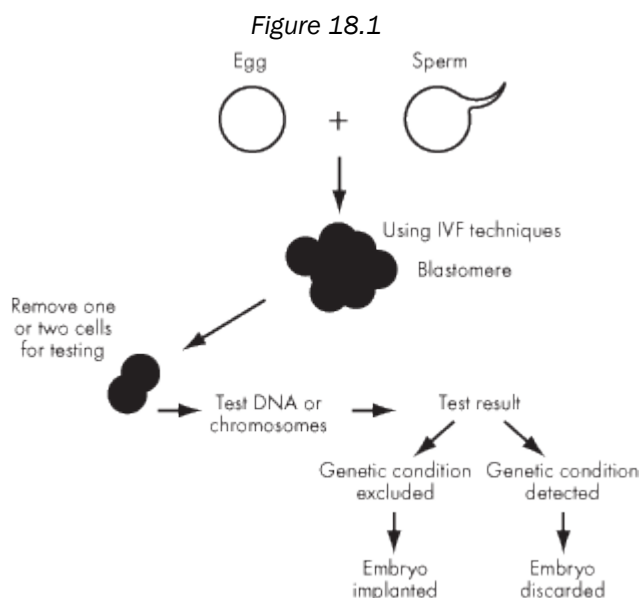


Figure 18.1. The PGD process. In some laboratories, the embryo is allowed to grow for up to five days so that cells are removed from the blastocyst, rather than the blastomere.

One or two cells are removed in order to test for the specific genetic condition in question. The removal of these cells does not appear to harm the developing embryo.

Only those embryos that do not have the specific genetic condition tested for will be transplanted into the woman's uterus on the same day, or, in some IVF units, on day five of development.

Usually, no more than one or two embryos will be transferred to the uterus at any one time to avoid the possibility of multiple births.

In some IVF units, unaffected embryos that are not used can be frozen for transfer in another cycle.

What are the advantages and disadvantages of PGD?

Success rates for having a child from an IVF cycle followed by PGD vary from IVF centre to centre but tend to follow standard IVF success rates.

Therefore a pregnancy and an unaffected child cannot be guaranteed using this technique.

It is important for an IVF unit to have a high pregnancy rate to be able to offer reliable preimplantation genetic diagnosis (PGD) with a real chance of achieving pregnancy.

Like any IVF procedure, stress and often disappointment can accompany PGD. Couples will need to balance the financial and emotional cost of the IVF procedure followed by PGD with that of termination of an affected child conceived naturally.

For couples with a moral or religious objection to pregnancy termination and who also have a risk of having a child with a genetic condition, this technique may provide the opportunity to have an unaffected child. In others, PGD may be a preferred option over prenatal testing in a naturally conceived pregnancy. It can also eliminate the possibility of repeated miscarriages for couples where one partner carries a chromosomal translocation.

In Australia, PGD is currently only offered in the private setting. Genetic counselling is important before considering PGD.

Author/s: A/Prof Kristine Barlow-Stewart and Mona Saleh
Fact Sheet 18: Preimplantation genetic diagnosis (PGD)

The Centre for Genetics Education (online).
www.genetics.edu.au

Accessed 12/05/2010

FTO Gene and Obesity

Many genes have been linked with obesity but how they contribute to disease is unclear. Now for the first time, researchers have been able to directly link one such gene (FTO) and the control of energy expenditure to explain how the gene contributes to obesity development.

Common human variants of the FTO (fat mass and obesity) gene predispose to obesity and having the 'fat' version can pile on as much as 3 kg. Researchers used mice without the FTO gene and showed that they do not become obese — in fact they do not grow properly after birth and have less fat tissue overall. The team show that it's because they expend more energy — even though they move less and eat lots.

A/Prof David Cameron-Smith is an expert in obesity at the Centre for Physical Activity and Nutrition Research at Deakin University. He is also Vice-President of the Australasian Society for the Study of Obesity.

"Why is it that obesity in some children starts early? These kids need to eat, really need to eat all the time. They also grow, obese children are often taller and bigger in all respects. The uncontrollable urge to eat (and grow) must start somewhere. The genetic difference can either be in the brain (appetite control) or in the metabolic pathways controlling energy use".

Nature reports the impact in mice of knocking out the actions of a novel gene linked to human obesity. Ticking more than one box, Nature carries a report on a novel gene, FTO.

FTO is expressed in the appetite centre of the brain and appears to have a major impact on food intake and energy

regulation. Using mice to knock FTO out has a dramatic effect to lower body weight in these genetically modified mice, mainly by simulating metabolic rate. The human FTO gene has previously been shown to be linked to human obesity, however this research helps unlock the complex interplay between factors expressed in the brain that control both appetite and metabolism.

A cure, genetic or pharmaceutical for human obesity is many years away, although any new knowledge on how the brain controls hunger and growth will help solve the complex disease blighting current and future generations of Australians."

Professor Bob Williamson is Professor of Medical Genetics at the University of Melbourne and Chair, National Committee for Medicine, Australian Academy of Science

"The human genome is a tool that lets scientists identify genes that may be involved in complex health-related conditions, such as obesity, height, or even behaviour."

One gene that has been shown to be involved in some cases of obesity is 'FTO', which seems to control the expression of genes for hormones that regulate how efficiently the body can use energy-rich foods. Fischer and her colleagues have removed the corresponding gene from a mouse, and have found that mice with no FTO are indeed thin, and their data suggests that the FTO protein controls energy expenditure.

Alas, lab mice are not men (they are inbred, which is why they are so good for gene experiments, but not as good a model for

human disease as sometimes claimed). While obesity has a genetic component, many genes are involved, not always the same ones for different individuals. A lab mouse also lives in a controlled environment, while people are influenced by the many things around them, about which we have the power to make choices.

The significance of this work is NOT that this is 'the gene for obesity' (a silly concept in any case), but that knowledge of how this pathway works (in mice and men) could lead to tests that identify those at high risk of obesity, and suggest strategies for pharmaceuticals that might alter the expression of this gene in the right direction."

Source:

*Australian Science Media Centre
(25/02/2009).*

Available: http://www.aussmc.org/genetics_of_obesity.php.

Accessed 23/04/2010

GPs & prenatal screening

General Practitioners need better support with informing women of prenatal screening tests for foetal abnormality, according to new research at Murdoch Children's Research Institute.

Recent developments have made screening tests for foetal abnormalities available earlier in pregnancy and women have a range of testing options accessible to them.

It is now recommended that all women, regardless of their age, are provided with information on prenatal screening tests.

"GPs are often the first health professionals a woman consults in pregnancy. As such, they are well positioned to inform women of the increasing range of prenatal screening tests available," Professor Jane Halliday said.

The research found providing this complex information warrants longer consultations and that GPs need more targeted educational resources.

"In order to improve women's capacity to make an informed decision about screening, attention needs to be directed to better support the GPs role."

Source:

Murdoch Childrens Research Institute.

Available: <http://www.mcri.edu.au/pages/research/news/2009/4/GPs-prenatal-screening.asp?TID=4>.

Accessed 23/04/2010

A Decision Aid - Testing in Pregnancy for Fetal Abnormalities

Available: <http://www.mitec.com.au/catalogue/category391/c860/p12640>

Inquiry into a National Disability Long-term Care and Support Scheme

As part of the National Disability Strategy, the Australian Government has commissioned an Inquiry into a long-term care and support scheme for people with disability in Australia.

The disability support system faces pressures on several fronts. There is a growing number of people with disability - the Australian Institute of Health and Welfare estimates that around 2.3 million Australians will have a high level of disability by 2030 - and an expected decline in the availability of informal care.

Calls for a feasibility study on a long-term care and support scheme have come from a cross-section of society, from unions to disability advocacy groups, from medical associations to carers' representatives, as well as from individuals living with disability or in a caring role. These groups have argued that a new approach is needed to tackle the disability challenges of the future.

Advocates have argued that a long-term care and support scheme would deliver incentives for early intervention, provide certainty for people with disability and their families and encourage efficiency in the disability services system.

The Productivity Commission will undertake the Inquiry, which will examine a range of approaches for providing long-term care and support.

An Associate Commissioner with particular expertise and knowledge in this area will be appointed to the Productivity Commission. The Productivity Commission's Inquiry began in April 2010 and report by July 2011.

The Government has also established an independent panel of people with expertise and knowledge of disability issues to advise Government and the Productivity Commission during the course of the Inquiry.

Why an Inquiry?

The appointment of the Productivity Commission to undertake the Inquiry recognises that this is a complex area of public policy which requires detailed consideration.

A range of important issues need to be examined, including: the design and parameters of any long-term care and support scheme; financing issues; service delivery and workforce issues; the interface with existing major areas of service delivery; the potential impact on Commonwealth and state/territory responsibilities for the provision of services and support; the impact on carers; the interface with existing workers' compensation, medical indemnity insurance and third party motor vehicle insurance arrangements; necessary changes to legislation and options for governance.

A serious examination is required including extensive modelling and analysis of interactions with other existing service systems such as health, aged care and income support. The Inquiry will assess whether a long-term care and support scheme would be appropriate, practical and economically responsible in the Australian context.

What will the Inquiry do?

The Government is committed to finding the best solutions to improve care and support services for people with disability.

The Productivity Commission will examine alternative approaches to funding and delivering disability services with a focus on early intervention and long-term care by undertaking an Inquiry into long-term care and support.

The Inquiry will assess the costs, including

cost effectiveness, benefits and feasibility of an approach which:

- ☞ provides long-term essential care and support for eligible people with a severe or profound disability, on an entitlement basis;
- ☞ is intended to cover people with disability acquired early in life rather than as the natural process of ageing;
- ☞ calculates and manages the costs of long-term care and support for people with severe and profound disability;
- ☞ replaces existing funding for the eligible population;
- ☞ ensures a range of support options are available, including individualised approaches;
- ☞ provides care and support for each person taking into account their desired outcomes over their lifetime;
- ☞ includes a coordinated package of care services which covers accommodation support, aids and equipment, respite, transport and a range of community participation and day programs available for a person's lifetime;
- ☞ assists the person with disability to make decisions about their support; and
- ☞ provides supports for people to undertake employment where possible.

The Inquiry will consider costs, implementation and design issues, governance arrangements and administrative issues, including for a social insurance model that reflects a shared risk of disability across the population.

How much will a long-term disability care and support scheme cost?

The Productivity Commission Inquiry will examine a range of options and approaches, including international examples, for the provision of long-term care and support for people with severe or profound disability.

What is the Independent Panel?

The Independent Panel has been established by the Government to act in an advisory capacity to the Productivity Commission and the Government. It includes individuals with a lived experience of disability and caring for people with disability, and with relevant professional expertise.

How can we get involved?

The Productivity Commission's consultations will be the means by which you can have input.

To receive updates about the consultations and other inquiry-related issues, you may wish to submit your details to the Productivity Commission's Interested Party List online at <http://www.pc.gov.au/projects/inquiry/disability-support>.

Alternatively:

email: disability-support@pc.gov.au,
or phone Roberta Bausch on (02) 6240 3221.

Freecall for regional areas is 1800 020 083.

If you are deaf, or have a hearing impairment or speech impairment, contact the Productivity Commission through the National Relay Service.

TTY users phone 133 677 then ask for (02) 6240 3221.

Speak and Listen users phone 1300 555 727 then ask for (02) 6240 3221.

Internet relay users connect to National RELAY Service website ask for (02) 6240 3221

Huntington's disease breakthrough

Melbourne researchers have made a breakthrough in the research into Huntington's disease.

The genetic condition inevitably leads to dementia and a shorter life. But in a world-first study scientists have found that leading a more active lifestyle can delay the onset of symptoms.

About one in every 10,000 Australians has Huntington's disease. It usually starts in people aged between 30 and 50.

Involuntary movements are an early symptom and as brain cells are gradually destroyed sufferers experience a change in personality and eventually dementia.

Its progress can't be stopped and the sufferer will die early.

The genetic disease will have been passed on to them by their parents and they then face the agonising decision of whether or not to have children themselves knowing that there's a 50 per cent chance that they too will pass it on.

Professor Martin Delatycki at Melbourne's Murdoch Childrens Research Institute says new research may help delay the onset of the disease's symptoms.

"We assessed activity in individuals in the early stages of Huntington's disease and assessed their activity before they developed symptoms.

And what we found is that those who led a passive lifestyle - passivity is activities such as watching television, certain social activities, certain professions - had on average a four year earlier onset than those who led the least passive lifestyle."

Professor Delatycki says staying physically and intellectually active has also been found to play a role in delaying other neurodegenerative disorders like Alzheimer's and Parkinson's diseases and dementia.

He says those at risk of getting Huntington disease will now be given some clear advice.

"People at risk of Huntington's disease avoid spending long periods of time in passive activities. All of us need down time and watching television is part of that. But what we're saying is don't spend hours and hours a day in passive activities.

Find activities in your life that have physical elements, intellectual elements. And we believe that this can delay the onset of the condition.

So we are making sure that the Huntington's community both within Australian but also internationally are aware of this research and we're encouraging people that we see to take on this information in leading their lifestyle. And we think it's very important that this begins from childhood."

How significant is the research internationally?

Professor Delatycki says "Well we think it's significant that for the first time something that someone at risk of this condition - currently there's no treatments proven to delay onset or slow progression - so we think this is significant that for the first time there is something that people at risk of Huntington's disease can do to delay the onset and hopefully slow progression of the condition."

Huntington's groups says they will now be able to advise potential sufferers of the incurable disease that there appears to at least be something they can do to delay its onset.

Source:

Eleanor Hall

The World Today—ABC News

<http://www.abc.net.au/worldtoday/content/2010/s2898496.htm>

Accessed 14/05/2010

Coordinated Care for patients with diabetes

The prevalence of diabetes is rising with 818,200 Australians with diagnosed diabetes in 2007-08 (double the number in 1995). By 2020 it is estimated that more than 2.2 million Australians will have type 2 diabetes.

Diabetes can and should be managed out of hospital, yet many patients with type 2 diabetes experience poorly managed care, contributing to avoidable hospital admissions, poor health outcomes and a reduced quality of life.

The Australian Government is taking action under its National Health and Hospitals Network. The Government is committing \$449.2 million over four years to fund the flexible delivery of primary health care services through general practice for treatment and ongoing management of people with diabetes who voluntarily enrol with their general practice. The initiative will start from July 2012.

How does this initiative work?

Patients with diabetes will have the option of enrolling with the general practice of their choice. Current funding through the Medicare Benefits Schedule (MBS) for the patient's GP primary health care will be redirected to a flexible funding package to the general practice with which the patient is enrolled, with funding for allied health services to be managed by the local primary health care organisation (Medicare Local). This will involve a new way of funding primary care services for people with diabetes.

Instead of paying for each individual service delivered (through a number of different programs), the Government will provide funding to general practices for their enrolled patients. The patient's general practice will

be able to manage funding linked to its enrolled patients flexibly – in order to provide patients with the GP primary health care services that best meet their needs, including for the management of their diabetes. Their general practice will arrange for the allied health services they require through the local primary health care organisation.

The Government's new funding will cover services for patients as required, including the cost of:

- ⊙ routine GP visits for ongoing primary health care
- ⊙ chronic disease care by the patient's GP and allied health professionals.

The flexible funding provided to practices under this program should minimise the likelihood of charges for patients.

Patients will maintain the right to use other medical services at other practices when required.

How will these new arrangements be developed?

The Government will work with patient and health consumer representatives and key primary health care groups, including GPs and allied health providers, on detailed implementation arrangements for this new diabetes initiative.

The costs of the initiative reflect the total new program costs to the Government.

Further information on the National Health and Hospitals Network is at www.yourhealth.gov.au

Psychological impact of PGD - literature review

This review begins with a summary of the known psychological aspects of IVF, followed by a review of the limited available literature exploring the psychological and broader psychosocial impact of PGD.

Through a selection and exclusion process, 19 studies were deemed as relevant for the literature review. The studies included attitudinal surveys of non-PGD users, and retrospective and cross-sectional studies with PGD users.

Psychological adjustment and well-being were a small component of two studies exploring the development of children born after PGD at two years postpartum.

Overall findings suggested that those with a significant history of traumatic reproductive experiences were more likely to perceive PGD as a possible reproductive option. Additionally, PGD was reported to be 'extremely stressful' and 'emotionally draining'.

The findings confirmed that there was a need for additional research in this developing area, with a focus on prospective studies using validated psychometric scales, that explores anxiety and depression at various time points through the PGD process, as well as the impact of PGD beyond the establishment of a pregnancy, parent infant attachment, and long-term follow-up of women for whom PGD was unsuccessful.

To obtain the full paper please email jkaratas@med.usyd.edu.au.

Psychological impact of preimplantation genetic diagnosis: a review of the literature.

JC Karatas, KA Strong, K Barlow-Stewart, C McMahon, B Meiser, C Roberts

Source:

Centre for Genetics Education's newsletter, Snippets, March 2010.

Available: www.genetics.com.au/pdf/marchsnippets.pdf. Accessed 16/04/2010.

Name Game

In clinical genetics, syndromes are named in a number of different ways.

In some cases, syndromes are named after those who first described them in honour of their scientific contribution.

Sometimes syndromes are described symptomatically, by the use of several key findings which appear frequently in affected patients. Some syndromes have been named using acronyms, using the first letters of clinical features associated with the syndrome.

In syndromes which are caused by chromosome rearrangements, syndromes may be named by the type of rearrangement and the chromosome involved.

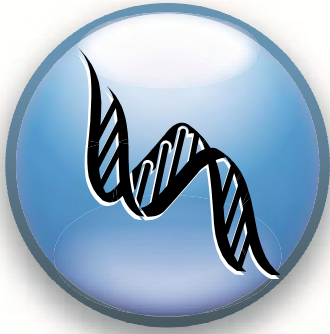
There have been a small number of syndromes which have been named in other ways, including using the name or initial of the first known patient, after the place or institution where it was first described, or by a specific metabolic fault associated with the syndrome.

There is no general consensus on the best naming system, and as a result, some syndromes are known by more than one name or system.

Robert J. Shprintzen,
Ph.D. Professor and Director
Center for the Diagnosis, Treatment, and
Study of Velo-Cardio-Facial Syndrome
State University of New York Health Science
Center
at Syracuse

Full Article Available:

<http://www.vcfsef.org/pdf/NAMEGAME.PDF>



Member Profile

The Australian Cystinosis Support Group

What is Cystinosis

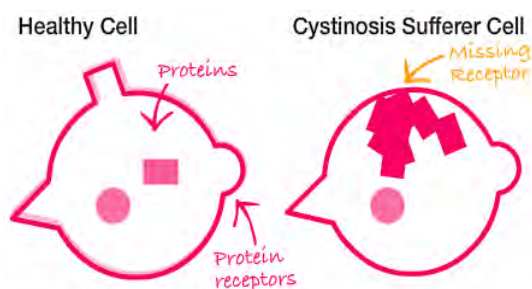
Cystinosis is a Metabolic disease characterised by an abnormal accumulation of the amino acid cystine in various organs of the body such as the kidney, eye, muscles, pancreas and brain. Different organs are affected at different ages.

Is It Inherited?

The disease is inherited in an autosomal recessive fashion, meaning that each parent of a child with Cystinosis carries one defective gene and one normal gene. The parents never have any signs of the disease.

What causes Cystinosis?

The Cystine content of Cystinotic cells averages 50-100 times the normal value. The cause is a defect in the transport of cystine out of a cell compartment called the lysosome, in which Cystine accumulates.



Because of cystine's low solubility, this amino acid forms crystals within the lysosomes of cells, and this is probably what destroys the cells.

What are the Symptoms?

There are three clinical forms of Cystinosis.

- Infantile (or nephropathic) Cystinosis
- late-onset Cystinosis; and
- benign Cystinosis

The latter form does not produce kidney damage. Infantile and late-onset Cystinosis differ in the age of appearance of the first symptoms and in the rapidity of the clinical course.

Infantile Cystinosis is usually diagnosed between 6 and 18 months of age with symptoms of excessive thirst and urination, failure to thrive, rickets and episodes of dehydration.

These findings are caused by a disorder called Renal Tubular Fanconi Syndrome, or a failure of the kidney to reabsorb nutrients and minerals. As a consequence, these important molecules are lost in the urine.

Children with Cystinosis also have crystals in their eyes {after one year of age} and an increased level of cystine in their white blood cells.

Without specific treatment, children with cystinosis develop end-stage renal failure i.e, lose their kidney function, at approximately 9 years of age.

If Cystinosis patients receive a kidney transplant and reach adult hood, then their new kidney will not be effected by the disease.

However, without Cysteamine treatment they can develop complications in other organs due to the continued accumulation of Cystine throughout the body.

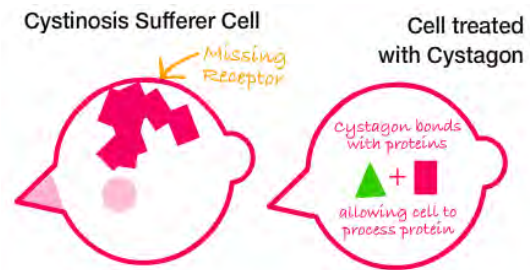
These complications can include muscle wasting, difficulty swallowing, diabetes, hypothyroidism, and blindness. Not all older patients develop these problems, however.

Can Cystinosis be treated?

The symptomatic treatment of the Fanconi syndrome is essential. The urinary losses of water, salts, bicarbonate, and minerals must be replaced. Most children receive a solution of sodium and potassium citrate, as well as phosphate. Some also receive extra vitamin D.

The aim of specific treatment for Cystinosis is to reduce Cystine accumulation within the cells. This goal is achieved by cysteamine treatment, which has proven effective in delaying or preventing renal failure.

Cysteamine also improves the growth of Cystinosis children. The food and drug administration has approved a capsule form of Cysteamine called CYSTAGON.



Kidney transplantation has proven very helpful in patients with Cystinosis, and Cysteamine therapy should be considered to try to prevent the late complications of the disease. For both young children with Cystinosis and older patients with kidney transplant, cysteamine eye drops may be available to remove the corneal cystine crystals.

For further information:

Australian Cystinosis Support Group
1 Clifton Crescent,
PINJARRA WA 6208

Phone: (08) 95312135

Email:

cystinosis.australia@bigpond.com

Web: <http://www.cystinosis.com.au>

*[Article reviewed by the Australian
Cystinosis Support Group]*



Coming Events

OFFICE SPACE AVAILABLE IN NEDLANDS

The Management Committee of Oasis Lotteries House, situated at 37 Hampden Road Nedlands, invites expressions of interest from not-for-profit charitable organisations seeking office accommodation.

Tenancies up to 53sqm are now available.

Oasis Lotteries House has undergone extensive refurbishment and offers tenants a number of facilities including the use of fully equipped meeting and training rooms, kitchens, onsite photocopying and allocated car parking.

Further information may be obtained by contacting Sharon Van der Laan on 0423 937 073

Men's Health Week Men's Health Information and Resource Centre

Date: 14-20 June 2010

Men and boys face different health concerns than women and girls, and IMHW is an opportunity to both acknowledge these differences and look for ways to improve the health and wellbeing of men and boys.

The week provides and an opportunity to acknowledge the diversity of men and boys and celebrate the positive contributions of men and boys to their communities

For further information please go to:
www.menshealthaustralia.net

11th International Symposium on Mucopolysaccharide and Related Diseases



Date: 23rd - 26th June 2010

Location: Adelaide Convention Centre

The theme for this conference is "Translating Research into Clinical Reality".

Our scientific and family programmes will be exciting, and relevant with a focus on the areas of newborn screening, prognostics, understanding pathology and therapeutic options.

Genuine opportunities for thorough discussion and debate will be a feature of the program - not only for the academics but also the families.

Registration Discounts

For Families

One registration form can be used to register and pay for up to two adults, four accompanying affected adults and four children.

Carer's and Volunteers

Carer's and volunteers who are attending the Symposium to provide care and support to children and affected adults participating in

the Children's Program, are requested to complete a Carer's Registration Form.

No registration fee is payable. You will be requested to provide your personal details and will be able to book accommodation if appropriate.

For further information please go to:

www.mps2010.com.au

Advance Health Directive and Enduring Power of Guardianship

Date: Friday 25 June 2010

Time: 9.30 am to 1 pm

Venue: St Catherine's College
(UWA)

2 Park Road, Nedlands



If you become seriously ill or unconscious you may be unable to communicate your healthcare decisions. An advance health directive is a good way to plan what medical treatment you would like in the event that you cannot make decisions for yourself.

The Health Consumers' Council WA (HCC) is hosting this free public forum.

Phone 9221 3422 or email info@hconc.org.au to register for this workshop

Registrations close on Friday 11 June 2010

Huntington's Disease National Conference

Date: 9-10 September 2010

The Conference will provide an opportunity to meet and share knowledge and experiences with family members, researchers, allied health professionals, care workers and members and supporters of all Huntington's Disease

Associations across Australia.

The program will focus on world wide research into Huntington's Disease; best practice in care; and support needs of families and community services caring for those affected by Huntington's Disease.

For further information please contact:

Huntington's Queensland
(07) 3391 8833

or

email: admin@huntingtonsqld.com



22nd Annual Batten Disease Support and Research Assn International Family Conference

"Field of Dreams and Hope"

Date: 8th-10 October 2010

Location: Sea World Resort & Water Park,
Gold Coast, Queensland

The conference aims to provide families and their friends, teachers and medical staff, affected or involved by this Disease with information, education and social interaction in various aspects of Batten Disease.

** The conference duration is Saturday and Sunday

For further Information visit:



www.battens.org.au/family_conferences.html



Promote your event here and on our website at no cost! Contact us on

(08) 9389 6722 or e-mail
info@geneticsupportcouncil.org.au



Resources

BioMedSearch

BioMedSearch is a biomedical search engine that contains NIH/PubMed documents, plus a large collection of theses, dissertations, and other publications not found anywhere else for free, making it the most comprehensive free search on the web.

BioMedSearch also provides advanced account features that allow saved searches, alerts, saving documents to portfolios, commenting on documents and portfolios, and sharing documents with other registered users.

Registering for BioMedSearch is free.

Visit: <http://www.biomedsearch.com>

Privacy Policy & Terms of Use:
www.biomedsearch.com/privacy.html

New policy Template Legislative Compliance

A Legislative Compliance Policy template is available at Our Community PolicyBank.

The template policy sets out a framework for compliance with the law (including OH&S, tax and privacy laws), governance structures, responsibilities and the processes established to give effect to that policy. Visit:

<http://www.ourcommunity.com.au>

New online training: putting diabetes guidelines into practice

Diabetes guidelines and their application in general practice is the focus of a new active learning module (ALM) from the Royal Australian College of General Practitioners (RACGP).

The online activity has been designed to help provide optimal primary care management to patients with type 2 diabetes mellitus.

This activity aims to improve the duration and quality of life in patients with type 2 diabetes by examining and implementing the new Diabetes Guidelines for General Practice 2009-10.

The learning tool is now available on the RACGP's online education portal, gplearning at www.gplearning.com.au

Free Financial Help Guide

Westpac and Our Community have just released the Guide for Community Board Members: Understanding Finances, the second in the community financial literacy series.

Download your guide now or request a hard copy.

Visit: http://www.ourcommunity.com.au/financial/financial_article.jsp?articleId=1043

Centre for Genetics Education's Info Sheets

Our information sheets, regarding the rarer and complex genetic conditions, are updated upon request. Some of the most recent ones updated include:

- Kabuki syndrome
- Oculopharyngeal muscular dystrophy
- Branchio-oto-renal (BOR) syndrome
- Spondyloepiphyseal dysplasia congenita (SEDC)
- Pyruvate dehydrogenase deficiency (PDD)
- Batten disease
- Cardiofaciocutaneous (CFC) syndrome
- Carbamyl phosphate synthetase (CPS)

To request an information sheet, please

email: contact@genetics.com.au

Web: <http://www.genetics.com.au>

Windsor Non-profit Survey #3

The survey was designed to capture information about non-profit employee satisfaction and performance in Queensland non-profit organisations.

A total of 348 employees from 22 non-profit organisations responded to the employee survey. In addition, 99 volunteers from eight different non-profit organisations took part in the volunteer survey.

Participants were drawn from organisations in a wide range of activities including disability, community services, animal welfare, member associations, and foundations.

Reports Available:

<http://www.windsor-recruitment.com>

Joint Pain Relief for Children and Young Adults

This is an independent Parent to Parent website for parents of children and young adults who have a rheumatology condition.

The purpose of this site is to record our experiences for the benefit of other parents where ever you are.

You can contribute from either a professional or personal perspective.

Post Categories

- Arthritis Story (2)
- EhlersDanlos & Hypermobility Disorder (7)
- Hospital Stays (4)
- Hospital Systems & Procedures (3)
- Managing Emotions (13)
- News (8)
- Pain in Young People (1)
- Pharmacy (1)
- Physical Therapies (3)
- Research (1)
- Resources (2)
- Rheumatology Issue Explained (2)
- School Days (1)
- Siblings (1)
- Support Groups (2)

Visit: <http://jointpainrelief.com.au>



Information accessed through the World Wide Web is of varying levels of quality and accuracy. The material supplied is for information purposes only & is not to be used for diagnosis or treatment.



Grants

ANZ Staff Foundation

Closing Date: 15 July 2010

The ANZ Staff Foundation was established in Australia in 1988 to help meet the needs of Australian communities. It is jointly funded by ANZ staff in Australia and by ANZ.

The Foundation has distributed \$1.5 million to more than 150 charitable organisations throughout Australia since its inception.

Aim

The ANZ Staff Foundation funds small projects (usually up to \$5,000) in the following areas:

- Skills and independence: giving people the skills to manage their lives and provide them with independence.
- Environment: assisting communities to conserve resources and protect the environment.
- Local initiatives: innovative projects from local community organisations.
- Capacity building: assisting organisations to build their capacity (especially in rural areas).

To access the guidelines and/or an application visit: <http://www.anz.com.au/about-us/corporate-responsibility/community/community-involvement/giving/>

Child Care Rebate

The Child Care Rebate helps working families with the cost of child care. The Child Care Rebate covers 50 per cent of out-of-pocket child care expenses for approved child care, with a rebate of up to \$7,778 (indexed) per child per year, for eligible families.

Can I get the Child Care Rebate?

There are certain requirements you must meet to get the Child Care Rebate.

You must have:

- used approved child care during the year;
- been eligible for Child Care Benefit (entitled at a rate of zero or more)*;
- passed the Child Care Benefit work, training, study test (for the purposes of the rebate).

*** Note:** There is no income test for the Child Care Rebate.

If you are eligible for Child Care Benefit, but your Child Care Benefit entitlement is zero due to income, you are still eligible for the Child Care Rebate.

How much Child Care Rebate can I get?

If you meet the eligibility criteria, you can get 50% of your out-of-pocket expenses for approved care up to a maximum of \$7,778 (indexed) per child per year.

Out-of-pocket expenses are the total fees you had to pay for child care expenses for approved care, less the amount of Child Care Benefit and Jobs, Education and Training Child Care fee assistance (if applicable) you received.

How do I meet the test for registered care?

For registered care you only have to participate in work related commitments at some time during a week or have an exemption. No minimum number of hours is required.

How do I meet the test for the Child Care Rebate?

You only have to participate in work related commitments at some time during a week or have an exemption. No minimum number of hours is required.

What is a work related commitment?

This is one or more of these activities:

- paid work or self employment
- setting up a business
- training or studying
- looking for work
- voluntary work to improve your work skills*

* **Note:** The time you spend in work related activities can be combined with other work related activities to meet the 15 hours per week requirements.

Voluntary work that does not improve work skills cannot be combined with other activities and must be undertaken for at least 15 hours a week or 30 hours a fortnight.

Can I satisfy the work, training, study test in any other way?

You will satisfy the work, training, study test if you are:

- on annual leave and long service leave
- on sick or other paid leave
- on paid or unpaid parental leave
- on self employment leave
- on self employment sick leave
- receiving Carer Payment
- receiving Carer Allowance
- caring for a disabled person on carer leave and carer sick leave

Are there any exemptions from the work, training, study test?

You may be exempt if:

- you or your partner get Carer Allowance or Carer Payment from Centrelink for a child
- you or your partner have a disability (the other partner must still meet the work, training, study test)
- your or your partner is overseas or in prison
- your or your partner are a grandparent with primary care for your grandchild
- you are facing exceptional circumstances

For further information visit: http://www.familyassist.gov.au/Payments/familyassistance/child_care_rebate/Pages/default.aspx

Grants for communities and local governments in regional and metropolitan Western Australia

The 2010 Grants Directory provides information on wide range of grants available to communities and local governments in regional and metropolitan Western Australia.

Visit: <http://grantsdirectory.dlgrd.wa.gov.au>



GSCWA can assist with grant applications and resources for your group!



Link Line

Genetic support groups are an important resource for families or people in a similar situation. The Link Line provides a supportive and confidential means of connecting individuals and families for whom no known genetic support group exists. If any individual is seeking contact with others in these circumstances, The Link Line is available to you for this purpose.

To date, there appears to be no specific support group for the following conditions/syndromes'.

Opitz G/BBB syndrome

A Western Australian family living with Opitz G/BBB syndrome are seeking contacts with others living with this condition.

Synonyms of Opitz G/BBB syndrome:

- BBBG Syndrome
- Hypertelorism with Esophageal Abnormalities and Hypospadias
- Hypertelorism-Hypospadias Syndrome
- Hypospadias-Dysphagia Syndrome
- Opitz BBB Syndrome
- Opitz BBB/G Compound Syndrome
- Opitz BBBG Syndrome
- Opitz G Syndrome
- Opitz Hypertelorism-Hypospadias Syndrome
- Opitz Oculogenitolaryngeal Syndrome
- Opitz-Frias Syndrome
- Telecanthus-Hypospadias Syndrome

Monomelic Amyotrophy

A person from Western Australia who is living with monomelic amyotrophy would like to make contact with others living with this condition.

Synonyms: of Monomelic Amyotrophy:

Benign Focal Amyotrophy, Hirayama syndrome, O'Sullivan-McLeod syndrome, Sobue disease and Single limb atrophy.

If you would like to contact with one of these families or individuals, please call Kristina at the Genetic Support Council for further details on 08 9389 6722 or email: info@geneticsupportcouncil.org.au

Kennedy's Disease

Russell is seeking contact with anyone affected by Kennedy's disease.

Please contact Russell via email to RussellAnderson@grocon.com.au.

Congenital Disorder of Glycosalation

Melissa is seeking to contact anyone affected by the Congenital Disorder of Glycosalation.

Please contact Melissa via email to Melissa@baf.org.au



GSC Members

Full Members

Acoustic Neuroma Association of Australia, WA Branch
Alzheimer's Association of WA
Angelman Syndrome Association
Arthrogyrosis Support Group
Australian Cystinosis Support group
NEW Australian Dyspraxia Association
Australian Huntington Disease Association
Australian Leukodystrophy Support Group
NEW Australian Mitochondrial Disorder Foundation
Australian Pituitary Foundation WA Branch
Australian Tuberous Sclerosis Society Inc.
Australasian CHARGE Syndrome Association
NEW Charcot-Marie-Tooth Association Australia
Cleft Lip and Palate Society of WA
Coeliac Society of WA
Cornelia De Lange Syndrome Support Group
Cushing's Disease Support Group
Cystic Fibrosis WA
Diabetes Australia - Western Australia
Down Syndrome Association of WA
Dyslexia SPELD Foundation WA Inc
Epilepsy Association of WA
Even-Keel Bi-Polar Support Association (Inc)
NEW Familial Hypercholesterolemia Support Group WA
Fragile X Support Group WA (Inc)
Haemophilia Foundation WA Inc.
Heart Kids WA
Klinefelters Support Group
Learning and Attention Disorders Society of WA (LADS)
LQTS Support Group WA
Lupus Group of WA
Lymphoedema Association of WA
Mental Illness Fellowship WA
Motor Neurone Disease Association of WA
Mucopolysaccharide & Related Diseases Society (MPS)
Muscular Dystrophy Association of WA
Neurofibromatosis Association of WA.
Parents of Children with Disabilities
Periodic Paralysis Society of Australia
Perth Tourette Syndrome Support Group
PXE Support Group of WA
Rett Syndrome Association of WA
Senses Foundation Inc.
Short Statured People's Association WA Branch

SIDS and Kids Western Australia
Spina Bifida Association of WA
Thalassaemia Association of WA
Turner Syndrome Association of Australia (WA Branch)
Support Organisation for Trisomy and Related Disorders of WA (SOFTWA)
Usher Syndrome Support Group
Western Australian Retinitis Pigmentosa Foundation

Corporate Associate Members

ARAFMI Western Australia
NEW Androgen Insensitivity Syndrome Support Group Australia
Association of Genetic Support of Australasia
Australian Kidney Foundation
Carers Association of WA Inc
ConnectGroups
Ectodermal Dysplasia Support Group - OzED
Genetic Support Network of Victoria
Health Consumers Council WA
NEW Myasthenia Gravis WA Friends and Support Group
Office of Population Health Genomics
People with Disabilities WA
NEW The Chromosome18 Registry & Research Society
The Kalparrin Centre
The Neurological Council of WA Inc
Western Australian Deaf Society Inc.

Individual Associate Members

Linda Bovill
Mark Bovill
Anja Hermann
Terry Keating
Sindhu Kurup
Amanda Samanek
Kristina Sengotta
Professor Charles Watson
Robyn Hendriks
Darren Webb

Membership Forms are available on the Web!
<http://geneticsupportcouncil.org.au>



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Attach mailing
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Contact Us

Genetic Support Council WA Inc. (GSCWA)
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East Perth WA 6009

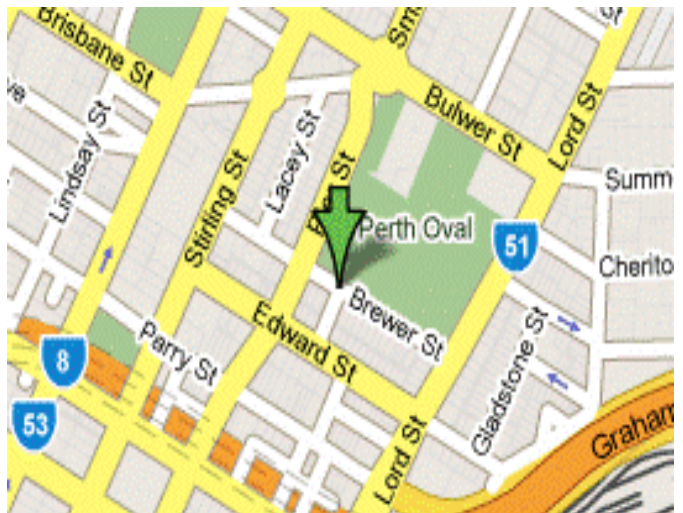
Phone: (08) 9389 6722

Email: info@geneticsupportcouncil.org.au

Web: www.geneticsupportcouncil.org.au

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