



Gene Advocate

Issue 66

2011

What is a PCEHR?

A personally controlled electronic health record (PCEHR) will be a secure, electronic record of your important health information.

The PCEHR will bring key elements of your health information together in a unified record. You control what information goes into your PCEHR and who gets access to this information.



As PCEHRs become more widely used and the PCEHR system matures, you will be able to access your own health information anytime, anywhere—and so will your authorised healthcare providers.

Your information in the PCEHR system will be accessible only by you and your authorised healthcare providers.

This information will allow healthcare providers to make better, more efficient decisions about your health and treatment.

The PCEHR will not hold all the information in your healthcare providers' records, but will

detail key and frequently used health care information.

You will also be able to nominate which members of your family (or carers) have access to your information. For example, if you are mature aged or chronically ill, you can nominate your partner (or a trusted person), to have access to your PCEHR.

"Yesterday I [turned] 64... and I'm certainly not going to remember everything that happens to me, so I'd like somebody else to take care of it and to know that it's available, correct, that I have access to it... I also want to know that it's private and confidential but available to those who need the information."

Caroline, Consumer

How will the PCEHR work?

If you choose to use a PCEHR, you will be able to set your own access controls and specify what information can be viewed and by whom.

With your permission, key pieces of health information can be viewed by participating healthcare providers across different locations and health care settings.

The security of your PCEHR health information will be protected through a combination of governance arrangements supported by information privacy and system security measures. These mechanisms will ensure healthcare organisations, providers, and other

representatives, can access or provide information to your PCEHR.

"I think it would take a lot of pressure off myself, and my father, who is often in a position where he has to try and remember things (for my mother)."

Kerry, Consumer

How will it affect my health care?

At the moment, your health information is stored across different locations with different providers—your doctor, your specialist, hospitals, and pharmacies, to name just a few.

If you chose to have—and use—a PCEHR, your key health information will be accessible online to whoever you authorise. The system will allow your key health information to travel with you through the health care system. This health information will be available to support your 'end-to-end' health care needs regardless of where and when you need it; even with a new healthcare provider.

A PCEHR will help reduce the frustrations of repeating information unnecessarily when you are unwell. The PCEHR system is about improving your health care by giving your healthcare providers the right information at the right time.

"After hours, when our doors are closed in the general practice, the ED can't get that (urgent) information. So to have a system where they can actually get that information, and speedily treat our patients, and vice a versa... that has to benefit the patient...and also me."

Karen, GP Practice Manager

What information will be available via my PCEHR?

Your PCEHR will contain summaries of your key health care 'events' (activities). More detailed information of these health care 'events' will remain in local health information systems.

It's expected that from July 2012, basic health information will be available to be shared and presented in the following forms on your PCEHR:

- shared health summary: a clinical document written by your nominated provider, containing information about your health status that is useful to a wide range of healthcare providers
- discharge summaries: information about your care in hospital
- event summaries: any participating organisation, such as an after-hours GP clinic, hospital or allied health professional can create an event summary following a health care event for posting to your health record.

It is proposed that additional information that can be added to your PCEHR may include:

- pathology reports
- diagnostic imaging
- prescriptions.

What information can I include on my PCEHR?

It is proposed the PCEHR system will provide three avenues for you to enter information into the PCEHR via the consumer portal. These avenues are:

- a way of providing 'key information' about any allergies and adverse reactions or medications (including over the counter medications) you would like your healthcare providers to be aware of
- a way of providing information about the location and custodian of your advance care directive (if you have one)

- a notes area where you, and your representatives, can record information about your health care you want to remember.

How will it improve the health care system?

The key objective for the PCEHR system is to support health care quality and safety for all Australians, while benefiting healthcare providers and contributing to the sustainability of the health care system.

“I believe it will contribute to better chronic disease management, not just in my area of (Indigenous) mental health service delivery but (also in) the broader issues of chronic disease.”

Mental Health Nurse, NT

How will it help my healthcare providers?

The PCEHR system will enhance your healthcare providers’ ability to access important health care information. It can help to improve your medication management, avoid duplication of tests and procedures and help reduce the incidence of adverse outcomes.

The PCEHR system will be integrated into the business and practice of health care. Healthcare providers and organisations that choose, and are ready, to participate in the PCEHR system will have the opportunity to:

- access health information more easily: the PCEHR system provides secure, quick and easy access to a consolidated view of your key health information from other participating healthcare organisations
- ensure safer health care: the PCEHR system provides easy access to key information about you such as allergies and previous adverse reactions as well as your medicines and related health care conditions
- deliver more effective health care:

easier access to information can be crucial to improved prevention, early intervention and greater continuity of care.

“I had a gentleman come to see me, and as he’s leaving the consultation he says ‘Oh, by the way, I was in Perth two weeks ago and I had this really bad allergy...’ I had no discharge summary, I didn’t know what he was allergic to... it’s just not good enough.”

GP, Melbourne

Health care for the 21st century

Like many other countries worldwide, the Australian health care system is facing an increasing set of challenges which threaten the sustainability of current health care practices.

The PCEHR system, as part of the national e-health program, is being designed to meet these challenges by bringing customer-focused electronic health capabilities to the Australian health system. This technology is similar to that used in other customer-rich environments such as banking and travel.

As a key work stream of the Government’s national health reform agenda, the PCEHR system will support:

- reductions in preventable hospitalisations due to lack of relevant information

“Any nurse will tell you that anything that takes them away from spending time on paperwork, that frees them up to spend time with their patients and clients, is what we’re all looking for.”

CEO Australian College of Nursing

- delivery of better coordinated health care across distributed healthcare organisations and reductions in the number of avoidable adverse events and costly unnecessary treatment activities

- improvements in health sector efficiency through streamlining of health care information processes

- clinicians and clinical support staff by providing patient information they need in a more timely, convenient and reliable fashion.

Other countries—such as Canada, United States, Denmark, and France—are integrating e-health solutions into their health care systems to ensure they are adaptable, efficient and meet future needs.

The PCEHR system will draw from these experiences and wide-ranging research and consultation. The PCEHR system will be a key part of Australia’s health care system in the 21st century.

When will the national PCEHR system be available?

Introducing a national PCEHR system is a complex undertaking with many levels of change needed involving people, processes and technology. Recognising this environment, the initial focus will be to deliver the core national elements of the PCEHR system for implementation from July 2012.

The implementation program will include a number of e-health sites across Australia. These e-health sites will deploy elements in controlled “real world”, practical, health care settings. Lessons learnt from these sites will help inform the priorities for national rollout. Once the national infrastructure is implemented, the PCEHR system will enable secure sharing of health care information. It will also have strict standards and governance arrangements.

Healthcare providers and organisations will be able to connect to the system whenever they are ready.

How can I register for my PCEHR?

By July 2012, all Australians who choose to participate will be able to register online for their own PCEHR. Registration is voluntary and

will not be required to receive health care. If you do not have internet access, you will be able to register by alternate means.

“I hope that a mother who’s got a child with a brain tumour doesn’t have to carry a compendium for what has happened to that child...so she doesn’t have to worry about taking the kids to Queensland (because) the information is transferrable.”

Nurse, Barwon Victoria

Continuing development

The growth of the national PCEHR system from July 2012 will be driven by its adoption by you, the consumer. It will be driven by your healthcare providers and their developing needs. It will also be driven and developed through market based initiatives, government considerations and lessons learnt from the first phase of the program.

Consultation continues to progress with key stakeholders ranging from professional associations for doctors, nurses, specialists and allied health professionals to information and communication technology groups, consumer groups, special interest groups and many others. The vital information and feedback gathered from these consultations is helping to drive and inform the design of the system and how the PCEHR will function in its first phase of implementation.

Further information can be found at

<http://www.yourhealth.gov.au/internet/yourhealth/publishing.nsf/content/pcehr>

Parkinson's disease: Global Study Includes Australians

A global study to uncover early signs of Parkinson's disease has been expanded to include Australians.

Researchers working with Hollywood star Michael J. Fox, a Parkinson's patient, have criss-crossed Australia this week eager to recruit people to join the study, which has already been launched in the US and Europe.

The researchers need Parkinson's patients, people without the disease, and scientists to help their study into identifying those most at risk of being stricken.

The study focuses on a search for Parkinson's "biomarkers", biological substances which can alert doctors to early signs of the degenerative neurological disorder.

Common examples of biomarkers in other diseases include high levels of cholesterol for possible heart disease.

In most cases, Parkinson's patients are diagnosed only when they show obvious symptoms such as body tremors, slowness of movement and muscle pain.

More than 80,000 Australians are estimated to be affected by the incurable disease.

What the Parkinson's Progression Markers Initiative (PPMI) study hopes to do is find biomarkers which can tip off doctors to a person's chance of developing the disease before those obvious symptoms begin to show.

Once a biomarker is unearthed, work can begin on improving treatments to either stop or slow the progress of Parkinson's.

Lead researcher Kenneth Marek, a clinical professor of neurology at Yale University, said data collected as part of the study would be made available to scientists around the world to help them identify Parkinson's biomarkers.

"The big issue for Parkinson's disease and Alzheimer's is that these are progressive disorders that get worse over time and the question is can you develop a drug that slows them down as opposed to treating the symptoms of the disease," Dr Marek told AAP.

"The issue that has been the show-stopper in trying to develop these drugs is that we don't have the tools to monitor what happens over time.

"This study is focused on developing those tools."

Parkinson's develops when nerve cells in the substantia nigra section of the brain die or become impaired.

The nerve cells stop producing dopamine, a chemical which allows smooth, coordinated function of the body's muscles and movement.

But it is only when 70 per cent of those cells are damaged that symptoms of Parkinson's disease appear.

Australians interested in taking part in the study must be at least 30 years old, newly diagnosed with Parkinson's and not be taking standard medications for the disease.

Healthy participants are also needed.

All must be willing to undergo a series of clinical tests and provide blood and spinal-fluid samples.

The study is expected to last five years and is being sponsored by the Michael J. Fox Foundation for Parkinson's Research.

Australians interested in taking part in the study can find out more information from the **Shake It Up** foundation's website at: www.shakeitup.org.au

Parkinson's Australia:
www.parkinsons.org.au

Genetic Support Council Annual General Meeting

The Genetic Support Council
AGM will be held on
Tuesday 18th October 2011

Guest Speaker:

Phil Gleeson

Principal — Maurice Blackburn Lawyers
“Insurance and Genetics”

Insurance is an area that can cause concern for individuals and families living with a genetic condition. In this relevant and informative presentation Phil will discuss life, total and permanent disability, trauma and income protection insurances as well as superannuation issues

**12.10 pm to 1pm at Oasis
Lotteries House 37 Hampden
Rd Nedlands 6009**

Light luncheon & Refreshments
will be provided

All members and friends welcome

RSVP: Kristina 9485 8999
info@geneticsupportcouncil.org.au

AGM
ANNUAL GENERAL MEETING

Wolf Hirschhorn Syndrome



4P Aussie Kidz was founded in 2010 by Melissa and Luke Young, after their daughter, Ellie, was diagnosed with Wolf Hirschhorn Syndrome at the age of 11 months.

4P Aussie Kidz is the only Australian organization that exists to provide financial assistance to families with children who have Wolf Hirschhorn Syndrome (WHS).

4P AUSSIE KIDZ aims to raise public awareness about WHS and money to assist with the financial burden associated with this condition.

About Wolf Hirschhorn Syndrome

Wolf Hirschhorn Syndrome refers to a condition that is the result of a genetic error on chromosome 4. It is caused by a deletion of genetic material near the end of the short (p) arm of chromosome 4. This chromosomal change is sometimes written as 4p-. It is estimated to occur in 1 in 50,000 births. This condition was first reported in 1965 in reports published by Wolf and Hirschhorn.

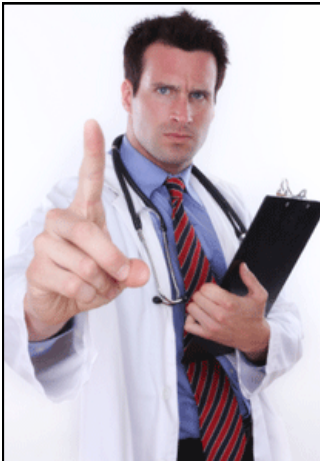
4P AUSSIE KIDZ aim to raise funds by holding an annual gala golf day/dinner, smaller things each month as well as government funding, philanthropic trusts, private donations and various activities.

Funds raised will contribute towards costly items that could help improve the life of a child with WHS, such as standing and walking frames, car seats, communication devices, wheel chairs, & private therapy.

4P AUSSIE KIDZ are having their first annual fundraiser on the 22 October, 2011 in Victoria at The Mentone RSL. To find out more details or to purchase tickets, visit their website (www.4paussiekidz.org.au) and see the events page.

You can contact 4P AUSSIE KIDZ at 4paussiekidz@gmail.com or follow us on facebook at www.facebook.com/4PAussieKidz or visit our website at www.4paussiekidz.org.au

Can a Patient Ever Know Too Much?



A recent QuantiaMD.com educational webinar for physicians was entitled, “The Patient Who Knows Too Much” as part of their “Difficult Patient” series.

Indignation from patient advocates and a CNN story appears to have prompted a name change to “The Cyberchondriac.”

You can register for free to view this presentation; however, you can't see the comments unless you are a healthcare professional. To get a full understanding, I suggest viewing the presentation.

It turns out that this case is about a patient with a mental illness who is expressing it via repeated doctor visits using information from the internet.

Unfortunately, the physicians who present this case do not clearly express this and it comes across as a bit of an attack on empowered patients.

Leonard J. Haas, PhD, ABPP, starts off by saying: “Patients who present their expertise as telling you how to practice medicine are implicitly discounting your expertise.”

I disagree. Often, patients are trying to understand what is going on with them or trying to find a diagnosis or new treatment.

Rare Disease Patients Drive Diagnosis

One of Siren's key insights, through years of working in rare disease therapies, is that rare disease patients are the primary drivers of diagnosis and treatment.

With the huge volume of information, there is no way physicians can stay current, especially on diseases which they may never see in their practice.

As Dr. Donald Lindberg, director of the National Library of Medicine, said, “If I read and memorized two medical journal articles every night, by the end of a year I'd be 400 years behind.” These patients and caregivers are uniquely motivated to research and try to find out what's wrong.

While by their very nature rare diseases are rare, taken as a whole they impact 1 in 10 Americans. I've heard story after story of patients and caregivers who played a critical role in their diagnosis. Here's an example: Julia Smit was only diagnosed after her grandmother, who worked at a lab, had Julia's blood tested. Siren's collection of stories about rare disease caregivers, *Uncommon Challenges; Shared Journeys*, offers more examples.

Information Source:

Children's Rare Disease Network. 1/08/2011.
Available: <http://www.crdnetwork.org/blog/can-a-patient-ever-know-too-much/>

The Interactive E-Health Experience

HealthBeyond is a FREE interactive experience aimed towards healthcare professionals and consumers. Take a step into the future by entering this virtual e-health world and experiencing the latest technologies and innovations relating to health and wellness.

HealthBeyond will provide visitors with an immersive experience that is relevant, educational, fun, entertaining, interactive and inspiring. Visitors will be able to gain an understanding of related healthcare settings including the home, GP clinic, hospital and aged care facility along with a strong focus on preventative/wellness.

HealthBeyond will present Australia's e-health vision to a national audience; to educate and inform them as to the scope of e-health and the future of healthcare.

HealthBeyond is a FREE major Australian exhibition with no requirement to pre-register, guaranteed to reshape how you think about and experience healthcare. Coming to a town near you soon!

The Roadshow

HealthBeyond will drop by the major metropolitan spots and go 'off the beaten track' to EDUCATE and INSPIRE tens of thousands of Australians.

The Roadshow will set off from Melbourne in 2012, and head North, stopping in all capital cities and regional centres across the great wide land (well, maybe not all regional centres, but we will get to as many as we can afford to!).



Visit: <http://www.healthbeyond.org.au>

Doctors renew call for mothball ban to prevent brain damage in babies

The risks are so extreme, the Australian Pesticides and Veterinary Medicine Authority have decided to suspend the sale of naphthalene flakes for domestic use.

This move has been welcomed by three leading professors concerned with that the link can be difficult to diagnose.

“Professor William Tarnow- Mordi, Director of the WINNER Centre for Newborn Research at Westmead Hospital and Head of Neonatal Trials at NHMRC Clinical Trials Centre, University of Sydney (<http://www.usyd.edu.au/news/84.html?newsstoryid=7135>), said babies, particularly if affected by the genetic condition G6PD, can develop massive breakdown of their red blood cells within hours of being wrapped in clothing stored with moth repellents containing naphthalene.”

Information Source: Hospital & Aged Care, 01/08/11, General News, Page 8.

Additional Information

What is the G6PD gene?

The G6PD gene provides instructions for making an enzyme called glucose-6-phosphate dehydrogenase. This enzyme, which is active in virtually all types of cells, is involved in the normal processing of carbohydrates. It plays a critical role in red blood cells, which carry oxygen from the lungs to tissues throughout the body. This enzyme helps protect red blood cells from damage and premature destruction.

Information Source: Genetics Home Reference. <http://ghr.nlm.nih.gov/gene/G6PD>

Rare-disease studies seek online giving

Global Genes Project

Launched in January 2010, this collaborative initiative began as a grass roots awareness campaign, and has grown from 5 disease groups to over 250 global organizations in its first year.

An Umbrella Initiative

The Global Genes Project has a big challenge. This campaign needs to act on behalf of over 7,000 rare diseases and over 1200 patient advocacy groups, creating unity.

The trick is to create a campaign that can be embraced by all without losing site of the individual diseases, their unique needs and identities.

We have worked hard to create a program, promotional opportunities, and soon to be launched – the R.A.R.E. Fund™, that provides value for individual disease groups while being part of the larger campaign.

For more information, please contact us at info@globalgenesproject.org

'Hope. It's In Our Genes'

Charleston Orwig, developed this slogan for the Global Genes Project, and in 8 weeks of launch it was translated into over 20 languages including Braille.

We encourage our partners to help spread the word by incorporating the denim ribbon, global genes logo or the slogan into their website, social media, online communities to help spread the word about rare disease and help unify this community of millions!

About Us

Rare Disease affects over 350 million people worldwide, and the largest percentage are children. There are over 7,000 rare diseases that have been identified all with very unique

needs, but many with little to no support. Although each individual disease may only impact a few, together the impact is in the millions. Rare disease is not so rare.

The Global Genes Project is broadly promoting the needs of the rare disease community, engaging the general public, garnering corporate support, under the "unifying symbol of hope" – the blue denim ribbon.



The Global Genes Project is a program of R.A.R.E. Project, a non-profit 501c3. R.A.R.E.'s leadership is committed to ensuring that its programs support its mission, are relevant and impactful. R.A.R.E. exists to:

- Raise Rare Disease Awareness and Education
- Unify and Empower a vibrant rare disease community
- Fund innovations that support in-their-lifetime rare disease research

Facebook: <http://www.facebook.com/globalgenesproject?sk=wall#!/globalgenesproject?sk=info>

Website: www.globalgenesproject.org

Future of general practice applauds tech-savvy health

General Practice Registrars Australia (GPRA) has welcomed the use of technology to deliver improved health outcomes for patients and to help address GP workforce shortages and working hours.

The endorsement follows two recent Government technology initiatives – the Medicare Benefits Scheme (MBS) item numbers for video consultations and the launch of an after hours GP helpline.

GPRA says video consultations will enable patients in rural, regional and **outer metropolitan areas to 'see' their specialist close to home** without the time and expense of travelling to major cities.

Meanwhile, the after hours GP helpline, staffed by nurses and GPs, will provide after hours medical advice to patients who cannot access their usual doctor.

"It is inevitable that technology will transform the way healthcare is delivered in Australia by removing distance, time and cost as barriers to accessing care," says GPRA Chair Dr Wicky Wong. "As future GPs, we will be at the forefront of this change, and we're hopeful that this will be a positive for our profession, as well as for our patients."

"These new primary care initiatives may also have the potential to positively impact on the work hours of GP registrars, particularly those based

in settings of workforce shortage," says Dr Wong.

GPRA recently released a discussion paper, which focused on the issue of safe working hours for GP registrars. The research was prompted by reports that due to workforce shortages, some registrars were consulting in normal hours, while also managing hospital or emergency presentations. This was sometimes coupled with registrars **being 'on call' overnight, and then continuing to consult the next day with minimal recovery time.**

"GPRA has long advocated for creative methods of addressing workforce shortages and patient access," says Dr Wong, "Innovation drives the advancement of science including medicine, and as the future leaders of general practice, current trainees have an imperative to embrace new technologies that may impact positively on healthcare delivery."

Information Source:

GP Australia. 13/07/2011. <http://www.gpaustralia.org.au/content/news/future-general-practice-applauds-tech-savvy-health-initiatives>

Myriad retains BRCA gene patents in latest US legal ruling

By Dr Philippa Brice
PHG Foundation 5/08/2011

The US Federal Court of Appeals has ruled that the company Myriad Genetics is, after all, entitled to US patents held for the BRCA1 and BRCA2 genes (see previous news), mutations in which are associated with hereditary breast-ovarian cancer syndrome.

This epic legal saga has hinged on whether it is possible to patent human DNA sequences; last year, the US government Justice Department said such patents should not be granted because all DNA sequences are a product of nature. Conversely, the new ruling from a panel of three judges supports the current policy of the US Patent and Trademark Office by stating that isolated DNA is patentable because it is markedly different from the DNA that exists within the body and is therefore not merely a product of nature, after all.

A third patent claim over the analytic process for examining genes to identify cancer-associated mutations was ruled invalid on the basis that it **involved 'patent-ineligible abstract mental steps'**. Myriad was nevertheless buoyant after the decision, but – unsurprisingly – the plaintiffs were said to be considering appealing to the full Court of Appeals or the Supreme Court.

Full Article Available: <http://www.phgfoundation.org/news/9422/>

Chronic Disease Management Plan

People with a chronic medical condition may be able to access Medicare benefits for eligible allied health services to assist in the care and management of their condition.

If you have a chronic medical condition, your doctor may suggest a GP Management Plan (GPMP). If you require treatment from two or more healthcare providers, your doctor may also put a Team Care Arrangements (TCA) plan in place for you.

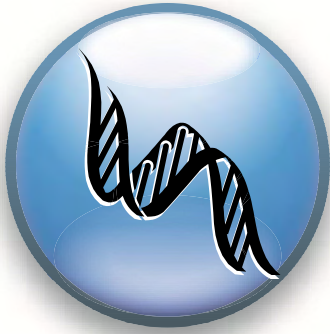
A GPMP is a plan of action agreed between you and your GP. It identifies your health care needs, sets out the services to be provided by your GP and lists the actions that you need to take.

A TCA plan enables your doctor to collaborate with, and refer you to, at least two other health care providers who will provide treatment or services to you.

If you have both a GPMP and a TCA prepared for you by your GP, you may be eligible for Medicare rebates for specific allied health services.

Information Source:

<http://www.humanservices.gov.au/customer/services/Medicare/chronic-disease-management-plan>



Member Profile

CleftPALS

The Cleft Palate and Lip Society

What is CleftPALS?

CleftPALS is a national organisation of parents and professionals involved in the treatment of the cleft condition. We all volunteer our time and efforts to provide information and support to families and professionals dealing with a cleft condition. We have members throughout Australia and exchange information with other parent groups around the world.

CleftPALS Newborn Programme

We offer support and guidance to new families all over Australia. We have contact parents who can visit you while you are still in hospital or once you get home. Ask your doctor or midwife to call us if you are still in hospital or call us yourselves. It can be very helpful to talk with another parent who has been through it all already.

For Parents of Older Children

CleftPALS is not just for parents of new babies. We are there at the end of the phone any time you want some advice or someone to talk to.

Reliable Information

We also send newsletters throughout the year to members and professionals, with articles by specialists, letters from other parents sharing experiences and ideas and sometimes letters from children and adults who were born with a cleft themselves. Information from the Internet: while this can be an extremely useful information tool, parents need to be wary of websites. Be aware that the information stated may not be applicable to your child.

What is a Cleft?

Early in pregnancy at about 6-8 weeks, the usual fusing, or joining up of tissues of the lip and/or palate does not occur, leaving an opening or "cleft". **A cleft sometimes occurs** in the lip, or the palate (soft and/or hard palate), or the lip and palate together. It can occur on one side of the lip/palate (called a uni-lateral cleft) or on both sides (called a bi-lateral cleft).

A cleft lip may sometimes be called **a 'hare lip'**. **This term dates back to** medieval times when it was believed a mother gave birth to a baby with a cleft lip because a hare had jumped across her path. 'Cleft

lip' is a more meaningful term to describe the condition today.

Do Many Babies Have a Cleft?

Clefts are one of the more common congenital conditions, affecting one in every 700 babies born in Australia.

Detection of a Cleft

Advances in technology have meant that more clefts are now being detected during a routine ultrasound in pregnancy. There are many reasons why a cleft may not be detected by ultrasound: position of the baby, lack of amniotic fluid, maternal tissue density and scarring, time of **gestation, baby's limbs moving across** the face. Clefts of the palate only are difficult to detect by ultrasound.

Diagnosis by ultrasound means you have time to prepare emotionally and practically for the arrival of your baby. Clefts not detected by ultrasound are usually detected at or soon after birth.

Why Did It Happen?

In a small number of cases, there is a relative also born with a cleft, therefore there is a genetic link. For the remainder there is no family history. The cause can be: unknown, multifactorial, chromosomal, or environmental. It is important to realise that clefts are not caused by feelings or actions of one or both parents, and research has shown that there is no one simple reason. Genetic counselling is available if you wish to discuss causes and the possibility of future children being affected by a cleft.

Medicare Australia's Cleft Lip and Cleft Palate Scheme

Medicare Australia recognises that the treatment for a person with a cleft may last into their adult life. The Cleft Lip and Cleft Palate Scheme helps families to meet treatment costs for specialised services for cleft lip and cleft palate conditions. Card holders are entitled to claim additional benefits from Medicare, to offset some of the dental costs associated with having a cleft.

In addition, Medicare Australia provides an information service and call centre to assist families in understanding the process, and what can be claimed. More information on the Cleft Lip and Palate Scheme is available on their website: www.medicareaustralia.gov.au

Or call the HOTLINE 1300 652 492

CleftPALS Australia presented a united case to the government a few years ago, successfully appealing for an extension to the eligible age from 22 to 28 years old.

Information Source:

www.cleftpals.org.au

[Article Reviewed By Neeva Stephen]

Contact Details

President, Neeva Stephen (WA)

Email: keeva@inet.net.au

Website: <http://www.cleftpalswa.org>





Coming Events

Western Australian Association for Mental Health

Date: 9 - 15 October 2011



Western Australian Association
for Mental Health

Mental Health Week raises community awareness about mental health issues and is held every October to coincide with World Mental Health Day.

Contact: 9420 7277

Website: www.mentalhealth.wa.gov.au/events/mentalhealthweek.aspx

National Down Syndrome Awareness Week

Date: 16th October 2011

The **National Buddy Walk** will take place at Lake Monger, Wembley, commencing at 10am. The walk will be followed by a Family Fun Day. Why not get a team together and join in the fun, help raise funds for Down Syndrome WA, and promote acceptance and inclusion of people with Down Syndrome in the community?



Down Syndrome WA

Contact: (08) 9368 4002

Toll free: 1800 623544

Email: dsawa@upnaway.com

Website: <http://sawa.asn.au>

Occupational Therapy Week

OT Australia WA

Date: 23 - 29 October 2011

This week promotes the role of Occupational Therapists in the community.

Contact: 9388 1490

Website: www.otauswa.com.au

National Bandanna Day

CanTeen WA

Date: 28th October 2011

National Bandanna Day is a great opportunity for everyone in the community to show their support for young people living with cancer. You can do this by selling bandannas throughout October and buying a bandanna to wear on National Bandanna Day, Friday 28 October.

Website: <http://bandannaday.com.au>

Movember Foundation

Date: 1-30 November 2011

Movember is an annual, month-long celebration of the moustache, highlighting men's health issues, specifically prostate cancer and depression in men.

Mo Bros, supported by their Mo Sistas, start

Movember (November 1st) clean shaven and then have the remainder of the month to grow and groom their moustache.

During Movember, each Mo Bro effectively becomes a walking billboard for men's health and, via their Mo, raises essential funds and awareness for Movember's men's health partners - The Prostate Cancer Foundation of Australia and beyondblue - the national depression initiative.

At the end of Movember, a series of Gala Partés are held to thank Mo Bros and Sistas for their support and fund raising efforts. The idea for Movember came about in 2003 when a few mates were having a beer in a small bar in Fitzroy, Melbourne. Inspired by the women's health movement, it was recognized that men were lacking a way to engage and actively involve themselves in their own health. During a conversation about fashion and past trends, the idea came up to bring the moustache back for one month, and in doing so, have some fun, raise a small amount of money and hopefully encourage men to talk about their health with each other.

Phone: 1300 GROW MO (1300 4769 66) within Australia

Website: <http://au.movember.com/events/>

National Cleft Awareness Week

CleftPALS

Date: November 8-14 2011



Each year during November, CleftPALS celebrates National Cleft Awareness Week, with family days, seminars, fund-raising and publicity. Each state organises its own events, but we share ideas, merchandise and support.

Website: <http://www.cleftpalswa.org>

International Data Linkage Conference

Date: Tuesday, 1 May 2012 - Friday, 4 May 2012

Location: Perth Convention Exhibition Centre

The conference will bring together a wide range of exciting leading international speakers, researchers, data linkers, practitioners, policymakers and consumers to discuss and exchange ideas relating to the full spectrum of utilising data linkage to advance knowledge for better health and social outcomes.

We are pleased to offer a comprehensive four-day package that will include a structured conference program comprising invited keynote presentations and presentations from submitted papers, as well as a series of half-day and full-day workshops covering cutting edge theory and practice relating to a wide range of elements critical to successful data linkage.

For more information on this event and to register, visit: www.datalinkage2012.com.au

Public Liability Insurance & Events

If Genetic Support Council Member groups are planning an event and require Public Liability Insurance the council may be able to assist. For further details and eligibility criteria contact us on

- 9485 8999 or
- email: info@geneticsupportcouncil.org.au

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

RareSpace



RareSpace is a collaborative project to share and advance knowledge about rare diseases among medical professionals, researchers, patients, parents, advocates and the general public.

Visit: <http://www.crdnetwork.org>

National Carers Strategy

The National Carers Strategy, announced on 3 August 2011, includes the following initiatives:

- increased eligibility for Bereavement Payment for some Carer Allowance customers
- changes to Carers Supplement
- extension of Carer Adjustment Payment, and
- inclusion of insulin dependent Diabetes on the List of Recognised Disabilities (LORD) for children aged 10 to 16 years.

The changes to:

- Bereavement Payment take effect from 1 January 2012
- Carer Supplement and Carer Adjustment Payment take effect from 1 July 2012.
- Inclusions of insulin dependent Diabetes take effect from 3 September 2011.

Visit: http://www.centrelink.gov.au/internet/internet.nsf/individuals/national_carers_strategy_faq.htm

Better Start for Children with Disability initiative

From 1 July 2011 the Better Start for Children with Disability initiative created new Medicare items to provide early intervention services from eligible audiologists, occupational therapists, orthoptists, optometrists, physiotherapists, psychologists and speech pathologists for children with sight and hearing impairment, cerebral palsy, Down syndrome and Fragile X syndrome.

Further information is available at: www.health.gov.au/children-disability

Our Community Templates

This webpage is designed to provide community groups with access to document templates that you can use in developing your community group's marketing materials.

Whether you are designing a direct mail letter, postcard or other form of marketing, this site will provide you with layouts to make your communications sing!

All the layout and template sites that we link to allow groups to download layouts free of charge.

Visit: http://www.ourcommunity.com.au/marketing/marketing_article.jsp?articleId=1413

A Brief guide to developing a Patient Participation Group

WHAT IS A PATIENT GROUP?

A selection of patients and practice staff who meet at regular intervals to decide ways of making a positive contribution to the services and facilities offered by the practice to the patients.

Visit: <http://www.lewishampct.nhs.uk/documents/1321.pdf>

Templates for Microsoft Office products.



Deliver great newsletters and brochures with free templates.

Visit: <http://office.microsoft.com/en-au/templates/results.aspx?ck=1&ex=2&qu=newsletters&av=all>

Siblings Australia

Siblings Australia is a unique national organisation committed to providing support for brothers and sisters of people with special needs; including disability, chronic illness and mental health issues.



Visit: www.siblingsaustralia.org.au

Facebook: <https://www.facebook.com/home.php#!/pages/Siblings-Australia/78545022595>

Medicare - Rare diseases

Qualifying rare diseases

Imatinib mesylate is available as a PBS subsidised therapy for the treatment of patients with the following rare diseases:

- dermatofibrosarcoma protuberans
- hypereosinophilic syndrome or chronic eosinophilic leukaemia
- Platelet-Derived Growth Factor Receptor (PDGFR) B fusion gene-positive myelodysplastic or myeloproliferative disorder
- aggressive systemic mastocytosis with eosinophilia.

Visit: <http://www.medicareaustralia.gov.au/provider/pbs/drugs2/rare-diseases.jsp>

NEW Food and Health Dialogue Website

The Dialogue's primary activity is action on food innovation, including a voluntary reformulation program across a range of commonly consumed foods. The reformulation program aims to reduce the saturated fat, added sugar, sodium and energy, and increase the fibre, wholegrain, fruit and vegetable content across nominated food categories.

Visit: www.foodhealthdialogue.gov.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

Steve Waugh Foundation Grants

Criteria for funding

- Applicants must be between the age of 0-25 (applicant must not turn 26 in the year the application is submitted).
- Be an Australian citizen or Permanent Resident of Australia.
- Have an Australian Medicare Card.
- Have a recognised rare disease as identified on ORPHANET (An International recognised list of rare diseases and reference for medical health professionals) Refer to Rare Diseases Listing.
- The rare disease has been formally and officially diagnosed by a Paediatrician and/or Geneticist, and has a name.
- You have and can provide evidence of the diagnosis of the rare disease signed off by a Paediatrician or Geneticist.
- You have and can provide medical, health and other documentation or support material required as part of this application.

Do I have to be an Australian citizen to get support from the Steve Waugh Foundation?

- Yes, you must be an Australian Citizen or an Australian permanent resident to gain assistance from the Foundation.

How do I apply for funding from the Steve Waugh Foundation?

- Check that you meet all Foundation criteria, refer to criteria list and follow ALL instructions carefully. You must complete ALL requirements of this application process.
- If you meet the Foundation's criteria please make sure you complete all sections in Part One and Part Two of the application form.
- Documents must be completed with every section filled in to be accepted. The Foundation will not accept partial or incomplete application forms.
- All invoices or quotes supplied must come directly from the supplier of the product and be valid at the time of the application and for a minimum of 3 – 6 months thereafter.
- You must ensure you provide a carefully considered and costed funding request and include everything that needs to be covered and specific to this application request. Should your application be successful, any additional or extra costs that have not been included or considered may not be covered by the Foundation.

ROUND 2:

1st October 2011 (applications open)
to 11th November 2011 (applications close)

ROUND 3:

1st February 2012 (applications open)

to 11th March 2012 (applications close)

- Please monitor website for the latest updates. Website:
www.stevewaughfoundation.com.au/content_common/pg-grants.seo

Does your non-profit need a server?

DonorTec now have the entire Microsoft Small Business Server 2011 range available through the Microsoft Software Donation Program.

Microsoft Small Business Server 2011 is designed for small organisations to easily and cost effectively share files, access servers remotely, host email and work collaboratively.

Nonprofits with less than 25 computers can now order Microsoft Small Business Server 2011 Essentials, which doesn't require CALs, with larger nonprofits able to order Microsoft Small Business Server 2011 Standard or Microsoft Small Business Server 2011 Premium Add-on.

Admin Fee: Au\$28.00 for Microsoft Small Business Server 2011 Essential

Microsoft Small Business Server 2011 Essentials provides home offices and small organisations with many of the features used by large organisations including network folder and printer sharing, remote access, support for mobile devices, and backup/restore tools in an integrated product that supports up to 25 users.

Requirements

Microsoft Small Business Server 2011 is available only as a 64-bit operating system and can be installed only on a server with a

64-bit processor.

Hardware

- 1.4-GHz (or higher) quad-core x64 or 1.3-GHz dual-core x64 processor (supports up to two sockets)
- 2 GB RAM; 4 GB recommended; 32 GB maximum
- 160 GB available hard-disk space
- DVD drive
- 800x600-pixel (or higher) display
- Ethernet interface card

Other

- For some server functionality - Internet access and a Microsoft Windows Live ID account
- For Internet access - broadband or high-speed -modem Internet connection

Supported client operating systems

- Microsoft Windows XP Professional SP2; Microsoft Windows Vista; Microsoft Windows 7; Mac OS X 10.5.8 or greater

To participate in the DonorTec program, Australian charities and nonprofit organisations must be non-government entities and meet the eligibility criteria outlined at: www.donortec.org/eligibility

For Further Information Visit: <http://www.donortec.org/microsoft/microsoft-small-business-server-2011-range>



GSCWA can assist members 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':

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 9485 8999 or email: info@geneticsupportcouncil.org.au

Dandy Walker Syndrome Support Group

Available: <http://www.facebook.com/group.php?gid=2357002701&v=info>

Basic Group Info

For people who have Dandy Walker Syndrome, or know someone who has it.

Privacy Type: Open: All content is public.

Contact Info

Website: <http://www.dandy-walker.org>

Online Tetrasomy/Pentasomy X Support Group

Online Tetrasomy/Pentasomy X Support Group and this website to provide support, friendship and information for all of them throughout the world and raise awareness of 48XXXX & 49XXXX, amongst the medical profession and the public. We have achieved our goal of creating a non-profit status within this group and we hope to be able to help our girls and their families.

Group Information

- Members: 129
- Category: Health Care
- Founded: Jan 28, 2000
- Language: English
- Membership requires approval

Visit: <http://groups.yahoo.com/group/tetra-x-list/>



Genetic Support Council WA

The Genetic Support Council WA Inc (GSCWA) is a not for profit organisation that acts as a peak body for genetic support groups in Western Australia. The Council provides information to individuals and families about genetic conditions including information on the services provided by Genetic Support Groups.

Genetic support groups can be a useful resource for individuals and families affected by a genetic condition. Support Groups can provide information about the condition and community resources, as well as an understanding and empathetic ear.

In Australasia, there are hundreds of such groups each focussing on a specific genetic condition. We can help put you in contact with these groups or provide the support and guidance to help you establish a new support group.

The Council may also be able to provide information and support for conditions so rare that there is no specific local support group.

The GSCWA maintains a database of Genetic Support Groups throughout Australia and with international links can provide up to date information on genetic conditions – including the latest information on research developments.

Information is also readily available on a range of associated topics - online resources, living with a family member with a genetic condition, parenting, and education in association with a genetic condition are some of these.

The Council conducts forums for members on issues of interest or concern to enable member's views to be represented to the wider community and State and Federal Governments. We also provide written and verbal submissions to Government and others in the field of genetics and participate with like bodies nationally to further represent Genetic Support Groups and their members.

We conduct forums and awareness events to enable participants to increase their skills and knowledge and access up to date information about resources for people affected by genetic conditions and their families. The Council maintains an up-to-date website and publishes a bi-monthly newsletter and information sheets for consumers.

The GSCWA actively supports the ongoing development of Genetic Support Groups. We are able to assist with the development of resources such as information packages, pamphlets and the printing of promotional material. Groups can also publish articles and advertise and promote awareness of their Support Group's activities on the GSC website and in our bi-monthly newsletter the "Gene Advocate".

Staff can assist groups and individuals in researching information regarding particular genetic conditions or predispositions and help keep members aware of matters of interest such as changes of legislation etc.

All GSCWA services are free to member groups and individuals. Please call us for further information or to access any of these services.

What causes genetic conditions?

There are three types of genetic conditions:

- Heritable conditions: Due to a mutation in a single gene
- Chromosomal conditions: Occurs when an individual is affected by a change in the number, size or structure of his or her chromosomes
- Multifactorial conditions: Due to the interaction of the genetic information and environmental factors such as diet, chemical exposure and lifestyle.

What can be done about Genetic Conditions?

Prevention

Some people are more at risk than others for developing a condition that is due to the interaction of environmental factors with their inherited genetic information. They are "genetically predisposed" to develop these conditions. However, the presence of an environmental "trigger" is necessary for the person to be affected with the condition. In some cases, prevention of the condition can be achieved by the person avoiding being exposed to the particular environmental factor that will trigger the condition.

For example, it is possible to prevent about 70% of the cases of spina bifida (a neural tube defect) in babies if women who are more likely to have a baby affected with spina bifida, because they are genetically "predisposed", take the vitamin folic acid before, and continue it during early pregnancy.

Early Diagnosis and Treatment

In some genetic conditions, early diagnosis, sometimes even before the symptoms appear, can lead to specific treatment. For example, all newborn babies in Australasia are screened for phenylketonuria (PKU) by a simple blood test.

Diagnosis and treatment within the first month of life are crucial to avoid intellectual disability.

Also, some cancers which have a genetic component, e.g. breast cancer, bowel cancer and melanoma, can be detected early enough to enable treatment to take place.

Checking the family health history may determine if a person or another blood relative are at risk for developing a genetic condition or for passing it on to their children.

Genetic Counselling

Genetic counselling is available to families and individuals that have concerns about a condition in their family which may have a genetic basis. A team of health professionals which may include clinical geneticists, genetic counsellors and social workers, work together to provide information and supportive counselling so that families may be better able to understand, and adjust to, the diagnosis of a genetic condition.

Genetic testing, if it is available and appropriate, can also be organised on the basis of informed consent. Genetics Services are available throughout Australasia and provide genetic counselling to assist in informed decision making regarding genetic testing.

Support Groups

Support groups provide affected individuals and families with information about the condition and community resources, as well as an understanding and empathic ear. There are over 200 genetic conditions for which there are over support groups/branches in Australasia.

Some points to remember when contacting a group

Many of the support groups contact numbers are home numbers, so please be considerate of the hours at which you call.

Many support groups do not receive funding and rely on their group members for income. Offering to pay for postage, photocopying and/or materials provided will be appreciated by many groups.



GSC Members

Full Members

Acoustic Neuroma Association of Australia, WA Branch
Alzheimer's Association of WA
Angelman Syndrome Association
Arthritis Foundation of WA
Arthrogyrosis Support Group
Australian Cystinosis Support group
Australian Dyspraxia Association
Australian Leukodystrophy Support Group
Australian Mitochondrial Disorder Foundation
Australian Pituitary Foundation WA Branch
Australian Tuberos Sclerosis Society Inc.
Cardio Facio-Cutaneous Syndrome Support
Charcot-Marie-Tooth Association Australia
CHARGE Syndrome Association of Australasia
Chiari & Syringomyelia Australia
CleftPALS WA
Coeliac Society of WA, Inc
Cornelia De Lange Syndrome Support Group
Cushing's Disease Support Group
Cystic Fibrosis WA
Diabetes Western Australia
Down Syndrome Association of WA Inc
Dyslexia SPELD Foundation WA Inc
Epilepsy Association of WA
Even-Keel Bi-Polar Support Association (Inc)
Fabry Support Group of Australia
Familial Hypercholesterolaemia Support Group WA
Fragile X Support Group WA Inc
Haemophilia Foundation WA Inc.
Heart Kids WA
Huntington's WA Inc
Klinefelters Support Group
Learning and Attention Disorders Society of WA (LADS)
LQTS Support Group WA
Lupus Group of WA
Lymphoedema Association of WA
Menieres Support Group WA
Mental Illness Fellowship WA
Mitochondrial Disease Foundation
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
PXE Support Group of WA
Retina Australia (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
Tourette Syndrome Support Group, Perth
Thyroid WA Support Group Inc
Turner Syndrome Association of Australia (WA Branch)
Support Organisation for Trisomy and Related Disorders of WA (SOFTWA)
Usher Syndrome Support Group

Corporate Associate Members

ARAFMI Western Australia
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
Kidney Health Australia
Learning Centre Link—Linkwest
Myasthenia Gravis WA Friends and Support Group
NZ Organisation for Rare Disorders
Office of Population Health Genomics
People with Disabilities WA
Self Help Organisations United Together (SHOUT)
Speak Easy Association Western Australia
The Chromosome18 Registry & Research Society
The Kalparrin Centre
The Neurological Council of WA Inc
Unique in Australia Chromosome Disorder Support Group
Western Australian Deaf Society Inc.

Individual Associate Members

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

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

Attach mailing
address label here

Contact Us

Genetic Support Council WA Inc. (GSCWA)
Oasis Lotteries House
Suite 7, 37 Hampden Road
Nedlands WA 6009

(08) 9485 8999

info@geneticsupportcouncil.org.au
www.geneticsupportcouncil.org.au

Office hours: 9am to 4pm Monday to Friday



- ▶▶ The views and opinions expressed in this newsletter are those of the individual authors and not necessarily those of the GSCWA. The material supplied is for information purposes only, and is not to be used for diagnosis or treatment.