What is The Human Variome Project?

Professor Cotton, from Melbourne, heads the international Human Variome Project, which has a bold aim: to collect all variations of genes that cause all diseases from every country. So far, he has rallied clinicians, geneticists and researchers from more than 30 countries to lend a hand.

Sharing data, reducing disease
The role that our genes play in our health and well-being is well known. The genetic makeup of an individual can cause a host of genetic disorders that can manifest from early childhood (cystic fibrosis, Prader-Willi Syndrome, Fragile X Syndrome) to adulthood (Alzheimer’s disease, polycystic kidney disease, Huntington’s disease) as well as significantly increase the risk of contracting more common diseases such as schizophrenia, diabetes, depression and cancer.

Furthermore the world is rapidly moving towards an era where it is both economically and scientifically feasible to sequence the genome of every patient presenting with a chronic condition.

But being able to sequence the genome of a patient cheaply and easily will be useless if we are unable to determine if the variations present in a sequence have an effect on human health. We are suffering from a critical lack of information about the consequences of the vast majority of the mutations possible within the human genome. And, even more concerning, is the fact that even when that information exists, it is not being shared and captured by the global medical research community in a manner that guarantees widespread dissemination and long-term preservation.

The Human Variome Project is trying to change this. They strongly believe in the free and open sharing of information on genetic variation and its consequences. They envision a world where the availability of, and access to, genetic variation information is not an impediment to diagnosis and treatment; where the burden of genetic disease on the human population is significantly decreased; where never again will a doctor have to look at a genetic sequence and ask, “What does this change mean for my patient?”

Source: http://www.humanvariomeproject.org/
Accessed 7/7/2011
Message from the GSNV Team

Welcome to the new look GSNV newsletter. We hope you enjoy this quarterly publication which will include articles on where we have been, what we have seen, what our support groups are up to, network and research news and of course important calendar dates.

For the last 12 months the GSNV team and committee and have been working very hard to establish and consolidate our strategic vision for the next three years. This process has involved a major audit of our internal operations and external relationships. We have engaged the feedback of our members and collaborators with our recent member’s survey. This exercise has helped enormously in directing our goals and we thank all those who have sent theirs back to us.

You will notice some changes in our logo and style and we do hope you like what you see. Our logo which is a gerbera flower reflects the uniqueness and beauty of each and every individual. No two persons are alike, and just like the gerbera we each have our own distinct genetic make up. We are still building on this new branding and image so we hope to further improve on our overall corporate look. Our new website, support group listing and database under well under construction so please bear with us if we have not updated your details yet. Once we are fully integrated and have tested the waters we anticipated a significant increase in the efficiency and reliability of our data.

As most of you are aware by now we have some new faces in the office and our team has changed a little. As the new team leader I will be managing Tarli Bogtstra, Stephanie Shepley, Penny Dodds and Lynley Donohue. With a fantastic mix of skills and experience between us, we have genetic support, development and administration covered. We have however not had the added benefit of volunteers for a little while so in the month ahead we will look at how we may encourage volunteerism and perhaps attract your attention.

All the latest developments in the GSNV and where to from here will be reported at our forthcoming Annual General Meeting to be held this October (stay tuned for the date) so we welcome all members and interested parties to join us. A full strategic plan for the next three years will be presented and as a member of the GSNV, your feedback is encouraged.

As well as planning for our future we have continued to work with support groups and engage in advocacy and representation as much as possible. Highlights this year so far have included Rare Disease Day celebrations, an FSHD Vic Branch Support Meeting, attendance of the First rare Disease Symposium in Fremantle, participation in an NHMRC workshop to inform the development of an action plan on whole genome sequencing and related technologies and participation in the Human Genetics Society of Australia (HGSA) 35th Annual Scientific Meeting – ‘Genetics in the Sun’.

In the months ahead we will continue to focus on support groups and with our support group afternoon tea on September 16, will engage groups to tell us what they need from the GSNV.

On behalf of all the GSNV team we look forward to every opportunity to grow our strengths and work with all our members in the months ahead.

Louisa Di Pietro
Team Leader
Better Health Channel

The Better Health Channel provides health and medical information to help individuals and their communities to improve their health and wellbeing.

The information is quality assured, reliable, up to date and easy to understand.

Information on the Better Health Channel is provided to help people stay healthy or understand and manage their health and medical conditions.

It does not replace care provided by medical practitioners and other qualified health professionals.

The Better Health Channel was established in May 1999 by the Victorian (Australia) State Government. The site does not have any commercial advertising and they do not accept any corporate sponsorship.

IndieGoGo helps you raise more money, from more people, faster.

IndieGoGo is the world's largest self-serve open-funding platform and can be used by community groups to raise much needed funds.

Since it's founding in 2008, the site has distributed millions of dollars to over 30,000 campaigns in 196 countries.

Anyone in the world can easily create a campaign to raise money, offer perks and maintain 100% ownership.

Anyone in the world can then donate to them.

The company has been featured on "Oprah," ABC's "Good Morning America," and the BBC, as well as in the Wall Street Journal, The New York Times, and Techcrunch. IndieGoGo is headquartered in San Francisco, CA.

For more information, visit www.indiegogo.com and follow at www.twitter.com/indiegogo and www.facebook.com/indiegogo.

www.indiegogo.com
Early Intervention Centre Program

GSNV has noticed the need for more education and support for families of young children who are currently undergoing genetic assessment.

This can often be a long process for all involved. GSNV receive a number of enquiries from both families and health/education workers associated with young children going through this process.

Mixed messages are often received by staff at Early Interventions Centres from families undergoing genetic assessments. GSNV sees the need to support families in this process. As this is often a difficult time for families, their understanding of what is occurring can sometimes be incorrect and confusing. This can also be seen in many enquiries that GSNV receive.

GSNV are initiating an educational program that can be conducted at all Early Intervention Centres in Victoria. A three hour educational presentation is given to staff, covering topics like basic genetics, what is covered in a genetic counselling appointment, issues that may arise for families and much time for questions. Appropriate information will be left at each centre and GSNV staff will be available for further consultations with both staff and families.

This Program has the support of Genetic Health Services Victoria and the Early Childhood Intervention Australia Victorian Chapter.

PEER SUPPORT

The GSNV works hard to connect individuals and families interested in sharing their experiences and insights with others.

Persons interested in contacting others in the same boat can advertise their details through the GSNV and we will assist in making connections.

Support group information sessions are also a fantastic way to connect people with shared experiences and prove to be an important networking opportunity, particularly for those travelling from rural and regional areas.

The GSNV will be hosting a number of support groups and research sessions over the coming months so please watch this space!

Enquiries

Since the start of the year we have had a steady increase in the number of enquiries to the GSNV office, in particular from health professionals.

Enquiries are predominantly from individuals and families, support groups and health professionals, and are generally about specific conditions.

Peer support in the form of support group information and making contact with others ‘in the same boat’ continues to be the main reason for contacting the GSNV along with enquiries about starting support groups, equity and access and general enquiries related to community services and general genetic information.

We expect that with the increasing number of joint projects occurring between the GSNV, our support groups and other community and clinical services, and raised awareness of the GSNV and its activities, the number of enquiries will continue to grow.

Support Group Afternoon Tea

Friday 16 September
1.30-2.30pm

Please join the GSNV team for an afternoon tea designed to support you, in supporting others.

We look forward to connecting with all our Support Groups, coordinating a visit to your organisation from our team and sharing ideas about planning for the future.

Enquiries will continue to grow!
Martin came home from school the other day with a black eye and broken glasses. Another boy had called him a freak and punched him.

Martin is an albino, which means that his skin has no color. He is very pale and his hair is white, and he doesn’t see very well.

Martin’s mother loves her son very much just the way he is. But when she sees other children tease him, she wishes he were not so different.

The world is filled with nearly 6 billion people, but each and every one of us is different from everybody else. Only you have your combination of looks, personality, and behavior. At the same time, you have traits, or ways of looking, thinking, and being, that you share with some other people on earth.

One way that scientists know this to be true is by studying our genes. Genes are units of information inside the cells of your body. They contain the instructions for making cells and for doing the work that goes on inside them. It is through the genes that traits are handed down from parents to offspring, in a process called heredity.

Discoveries are teaching us a great deal about these instructions that construct and operate the human body. This new information will give us new opportunities to control the destiny of our bodies. But at the same time, it will force us to face new and sometimes difficult choices.

Questions Raised by the New Genetics
To get an idea of the many choices that come with the new genetic information, consider Martin. Martin is an albino because his genes do not give the right instructions for his body’s production of pigment, the dye that colors the skin, eyes, and hair. The result is that Martin is very pale. He must avoid the sun because he is at high risk of sunburn and skin cancer. Strong light hurts his eyes, and his vision is poor, so he needs glasses.

Suppose researchers discover a way to treat Martin’s genes so that they give the proper instructions for producing pigment. It would mean that Martin’s skin and eyes would regain color. He no longer would have to stay out of the sun all the time. Plus, he wouldn’t stand out from other children. These changes could make a big difference in Martin’s life.

Do you think Martin should have the genetic treatment? In other words, do you think being an albino is a medical problem that needs fixing?

Think about the choices Martin’s mother would have to make. If she loves Martin the way he is, how does she explain a decision to have him treated? But if he is unhappy with the way he is, how does she explain a decision not to treat him? Also, many medical treatments have side effects. What level of risk is acceptable?

Perhaps when Martin grows up, he will decide that he wants to prevent his children from having the problem he has had. He may decide to have any baby of his tested before it is born, to make sure it is not an albino. If it is, he and his wife could choose to have an abortion and try again.

It’s possible that when Martin grows up, he will be comfortable with how he looks. He may not care whether his children are born albino. In fact, he may even prefer it because then they would look more like him.

Do you think it is wise to let children into the world with problem skin and poor eyesight if we know how to keep this from happening? Another way to ask this is, Should Martin be prevented from having children who are albino? Who are we to say no to him?

Finally, there is the question of where society should put its time and money. Perhaps along with research into the treatment of genetic conditions, we should put equal effort into teaching children (and adults) to accept those who are different?

These questions are just the “tip of the iceberg” when it comes to genetic research. There are many more.

Source: Chapter 1 Your Genes, Your Choices by Catherine Baker. Exploring the issues raised by genetic research. Your Genes, Your Choices describes the Human Genome Project, the science behind it, and the ethical, legal, and social issues that are raised by the project. This book was written as part of the Science + Literacy for Health project of the American Association for the Advancement of Science (AAAS) and funded by the U.S. Department of Energy. http://ehrweb.aaas.org/ehr/books/1_martin.html
Join the NDIS campaign

We encourage you to sign up to the ‘Every Australian Counts Campaign’.

Every Australian Counts is the campaign for the introduction of a National Disability Insurance Scheme.

The NDIS will revolutionise the way people with a disability, their families and carers are supported in this country. It will transform the way services are funded and delivered, ensuring people are better supported and enabling them to have greater choice and control.

The goal of the campaign is to demonstrate to the Australian government how much support there is to realise a National Disability Insurance Scheme.

Over the next few months the Productivity Commission will report to the Government on the findings of its inquiry into a long-term disability care and support scheme.


Where GSNV has been in 2011

Feb 28 – Rare Disease Day 2011

April 1 – Illoura Early Intervention Presentation – Genetic Testing

April 17 – Charcot Marie Tooth Information Session

May 4 – Facioscapulohumeral Dystrophy (FSHD) Victorian Branch Meeting

May 10 – Melbourne University Principles of Clinical Practice Patient Information

May 20 – Neurofibromatosis Association of Australia (NFAA) Information Session

July 21 – City of Boroondara Free Service Expo (this Expo was to give people with a disability and the ageing community the opportunity to learn more about free services that are available to them).

FSHD (Facioscapulohumeral Dystrophy) Global Research Foundation – Vic Branch Meeting

On 4 May, GSNV hosted the FSHD Global Research Foundation’s Victorian Branch meeting.

International researchers Professor Alexandra Belayew from Belgium and Professor Melanie Ehrlich from the US presented their latest insights into the causes of FSHD and potential treatment options. Several executive members of the FSHD group from Sydney attended as well as members affected by the condition and their families from Victoria, many of whom travelled great distances.

GSNV helped to produce flyers to advertise the event as well as organising location and catering.
**INTERSTATE & INTERNATIONAL FOCUS**

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### Orphan Drugs

"Orphan drugs" are medicinal products intended for diagnosis, prevention or treatment of life-threatening or debilitating rare diseases.

They are “orphans” because the pharmaceutical industry has little interest under normal market conditions in developing and marketing drugs intended for only a small number of patients suffering from very rare conditions.

The cost of bringing a rare disease medicinal product to the market is not be recovered by expected sales of the product.

However patients affected by rare diseases need to be informed about scientific and therapeutic progress. They have the same rights to care as any other patient.

For this reason, governments and rare disease patient advocacy organisations such as Eurordis (Rare Diseases Europe) have emphasized the need for economic incentives to encourage drug companies to develop and market medicines for the many neglected and "orphaned" rare disease patients.

In order to stimulate research and development in the sector of orphan drugs, authorities have implemented incentives for health and biotechnology industries.

The Australian orphan drugs policy was set up in 1997.

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### Medical funding may be stopped

The future of Ari Karambatos, who has a rare medical condition, depends on a government decision. The Murrumbeena father-of-three has Pompe disease, an inherited disorder which progressively weakens the heart, muscles, liver and nervous system.

Mr Karambatos is one of 16 adult Australians with the disorder who rely on Myozyme — a drug costing $500,000 a patient a year — which is supplied free of charge by manufacturer Genzyme.

But newly diagnosed patients over the age of two will be excluded from the program after Genzyme announced it can no longer sustain it, prompting calls for government funding. There are fears the 16 patients will be unable to get ongoing treatment.

The Government’s Pharmaceutical Benefits Advisory Committee has rejected putting the treatment on the life-saving drugs program*. It will review another submission from Genzyme. The decision is expected by next month.

Australian Pompe Association president Helen Walker said the drug was financed in the healthcare systems of 44 other countries. “That patients around the world being treated with Myozyme have seen tremendous results highlights the need for the drug to be subsidised in Australia,” Ms Walker said.

Mr Karambatos was diagnosed in 2004. “Before I started (taking Myozyme) I was deteriorating ... since I received it I feel much better,” he said. “If they stop it, we (patients) won’t last long.”

Source: Caulfield Glen Eira Leader Tuesday July 12, 2011. Reporter Jenny Ling

*Life Saving Drugs Program (LSDP)

(Through the LSDP, the Australian Government provides subsidised access, for eligible patients, to expensive and potentially life-saving drugs for very rare life-threatening conditions.)

Source: www.orpha.net/consor/cgi-bin/Education_AboutOrphanDrugs.php?lng=EN

Pompe’s disease is an example of a rare disease where access to lifesaving medication is hampered due to the cost of providing it for such a rare condition as seen in the following article.
NF Report

Assisted by a grant from GSNV, NF Australia held a Neurofibromatosis Information session on Friday 20 May 2011 with guest speaker Professor Kathryn North.

Professor North from the Institute for Neuroscience and Muscle Research at the Children’s Hospital at Westmead, Sydney, began with a PPT presentation and continued with questions from the audience. Afternoon tea and conversations followed.

The overall feeling was of enthusiasm, satisfaction (at hearing someone who really knew about NF), and also frustration at not having enough time with Professor North. There is a real need for more of these types of sessions. People travelled from Adelaide, Bendigo, Geelong and Shepparton, as well as all parts of Melbourne.

NF Australia wishes to thank the GSNV for the wonderful assistance they gave as well as the grant which enabled us to hold this session.

Somebody to Listen

Raising children can be challenging and isolating at times. Sometimes it is good to have someone with whom you can talk things through.

The Parent Support Program is able to offer free counselling sessions with an advanced diploma student at Hartnett House on Wednesdays.

Free counselling will be available from Patricia Yeung, Advanced Diploma Student of The Australian Institute of Family Counselling. Please call Thea on 9385 9235 to book a session.

Support Group News

My Story | Anna Hickey - Gorlin Syndrome

I was diagnosed with Gorlins Syndrome at Peter MacCallum hospital in Melbourne in 1979. Fortunately the Plastic Surgeon on duty, Mr. Simon Donahoe made the connection between the bccs and the jaw cysts I had as a teenager in Ireland.

The first BCC appeared when my first son was one year old. I had skin cancers removed by a Dermatologist in his rooms for ten years.

It was only when he administered radium to a particularly difficult spot on my nose that warning signs appeared. The area did not heal and after a year he referred me to a plastic surgeon who gave me my first skin graft and warned me to be careful.

A colleague at work suggested I go to Peter MacCallum.

My doctor had a friend, who was a surgeon studying under Professor Gorlin in America and these two gentleman operated on me the day after Mothers’ Day in 1979. I was sore physically, emotionally and mentally. I organized my life round family, work and three monthly visits to hospital for more surgery.

Unfortunately in July 1981 I was asked to resign from my job because of needing so much sick leave.

In May 1988 I secured a part time reception job for a Chiropractor who was understanding and supportive as I turned up looking the worse for wear. Patients sometimes asked me if I had been in a fight, gone through a windscreen or fallen over!

Medicare twice questioned the amount of treatment I was having. The first time I had to ask the Ombudsman for help, the second time the pathology reports alone were sufficient to persuade them to agree to pay. If Medicare does not pay, then no private health insurance company will pay.

In recent years a new treatment called Photodynamic Therapy has proved very effective in the treatment of BCCs.

As Medicare will not contribute towards this treatment I was advised to have the treatment at Peter Mac. This treatment is far less invasive than surgery and enables a person to continue their work schedule provided it is not outside. Although is not a cure it helps keep the BCC’s under control.

I have often wanted to talk with others who suffered from Gorlin Syndrome. Eighteen months ago I started the Australian Gorlin Syndrome Mutual Support Group. We have 19 members spread over Queensland, NSW, Victoria and South Australia. The Adelaide group also started “Gorlin Syndrome In and Beyond Australia” on Facebook and meet regularly for lunch.

My husband and I travelled to Perth in May to represent these groups at the Annual Dermatology Conference. We are in touch with the American BCCNS Life Support Forum and the UK Gorlin Syndrome Support Groups. It is very affirming to be part of this network of support.

There are exciting new trials of drugs being done in the US and Switzerland. An organization called Cart Wheel is taking information from people who might be interested in taking part in trials.

If anybody would like to know more about us they can contact Margaret Emery in Adelaide on pmemory@bigpond.com. And/or me annahickey43@gmail.com. Thank you for taking time to read my story.
Grants open
A great range of community grant programs and funding opportunities are currently available including training workshops to assist you in your applications.

THE CITY OF MELBOURNE GRANTS
International Day of People with Disability 2012 grants
International Day of People with Disability 2012 grants are awarded to programs designed to celebrate International Day of People with Disability on 3 December 2012.

On this day, the Melbourne community has the opportunity to acknowledge the integral and valuable contribution people of all abilities make in our community.

Your project must support one or more of the following themes from:
- promoting the diversity and talent of people with disabilities
- raising positive community awareness of disability
- providing learning and development opportunities for people with disabilities
- creating sustainable and positive outcomes for people with disabilities.

To find out more and to book, visit the website. www.melbourne.vic.gov.au/CommunityServices/Pages/CommunityOrganisationTraining.aspx
Contact: Shawn Neilson, MetroAccess Project Officer on 9658 8587 or 0413 011 832 for further information on either of these opportunities.

DEPT OF HUMAN SERVICES (VIC)
Victorian Aids and Equipment Program
Grant G03183 (Category: Disability)
On ongoing grant to assist eligible people to access subsidised aids, equipment and home modifications to enhance their safety and independence, support their family and carers and prevent premature admission to institutional care or high cost services. Contact your local Aids and Equipment issuing centre.

You can apply if you are a child or adult with a long-term or permanent disability, are frail, and are a permanent resident of Victoria, or on a Permanent Protection Visa (RoS) (subclass 851) or an asylum seeker, you may be eligible for aids and equipment. Apply via mail.

For further Information visit www.dhs.vic.gov.au/disability/supports_for_people/living_in_my_home/aids_and_equipment_program
Contact: Disability Services, DHS, disability.services@dhs.vic.gov.au
Freecall: 1300 650 172
TTY: (03) 9096 0133

PETER BROCK FOUNDATION GRANTS
On ongoing grant providing assistance to various organisations and worthwhile causes. The Foundation also has a strong commitment to assisting the community and individuals in times of need.

For further Information visit www.peterbrockfoundation.com.au

YARRA COUNCIL COMMUNITY SKILLS AND TRAINING
These ten sessions run from February to November 2011, and provide groups with practical advice to assist in everyday activities, such as working with financial statements, recruiting volunteers or planning an event.

All sessions take place on the first Wednesday of the month from 6pm - 7.30pm at the Richmond Town Hall. Bookings are essential as places are limited.

Contact: Nina Collins, Community Planner on 9205 5146 or email Nina.Collins@yarracity.vic.gov.au

This grants information is sourced from the EasyGrants monthly newsletter, with permission from OurCommunity.com.au. If you would like to receive a complete monthly update of all grants that may be available to you, please go to www.ourcommunity.com.au/easygrants
GSNV Small Grants are open to all current financial members of the GSNV for projects that support the work of a support group.

Grants may be used for any once-off project a group may undertake that benefits people affected by genetic conditions.

Examples from past years include printing brochures, venue hire, and event catering and library books. GSNV grants may also be used towards administration costs for unfunded agencies.

Amount Available

In order to share the funds amongst as many groups as possible, projects of $50 to $400 will be considered.

The total funding pool available is limited to our total membership fees for one financial year.

If applications received exceed the total funds available, the GSNV will do its best to fund as many eligible groups/projects as possible by offering partial funding.

If your project cannot go ahead without full funding, please indicate this on your application form.

Criteria

- All applicants MUST be current financial members of the GSNV ($22 per year).
- Applications will be judged according to how they meet GSNV’s objective, which is to empower people to overcome genetic challenges through information, education, support and advocacy.
- Groups who received funding in 2010 are welcome to apply, however in the event that there is more money requested than total funds available, priority will be given to groups who did not receive funding last year. Partial funding may be awarded where there are several worthwhile applications.
- Successful applicants must acknowledge GSNV’s Small Grant Program as appropriate.
- DGR Status is not required. For accounting reasons, successful applicants will be asked to either invoice the GSNV or provide confirmation on official group letterhead.

Description of Project

In your proposal outline, please provide a basic description of your project. You may wish to include the following:

- Your project details
- Some aims and objectives
- Why this project is important
- What the money will be spent on (fee free to list some items)
- How you may wish to acknowledge the grant and your association with the GSNV (e.g. in your newsletter, on signs at an event, your brochure, web site etc).

Process

Application – Due Monday September 12, 2011 by 5pm.

GSNV understands that you have many competing demands on your time. The application process has therefore been streamlined to a simple 2 page form. You are welcome to attach any additional documentation or letters of support that may assist our decision. (Excessive documentation may not be read.)

All applications MUST be on the application form.

Successful Applicants

Our accounts system requires documentation from successful agencies before we can raise cheques – either a tax invoice or an ‘official’ letter on your letterhead. Cheques will be raised once we have received a tax invoice or letter from successful applicants.

The Decision – October 2011

GSNV’S Management Committee will appoint a panel, who will review applications and make a decision. The Panel’s decision is final.

Our “kitty’ is small, so if several worthwhile applications are received, partial funding may be awarded to some (or all) applicants.

Applications will also be checked against the criteria (see above).

The outcome and Small Grant recipients will be announced at the GSNV Annual General Meeting in October, date to be announced.

All applicants will be advised of the decision in writing by October 7, 2011.

Successful Applicants

Our accounts system requires documentation from successful agencies before we can raise cheques – either a tax invoice or an ‘official’ letter on your letterhead. Cheques will be raised once we have received a tax invoice or letter from successful applicants.

Applications close Monday 12 September, 2011.

Submissions may be made by:

- Fax to (03) 8341-6390;
- Mail to PO Box 1100, Parkville 3052;
- Email to louisa.dipietro@gsnv.org.au

More Information

For more information, phone the GSNV team on (03) 8341 6315 or email to louisa.dipietro@gsnv.org.au
RARE DISEASE DAY

Rare Disease Day: Rare but Equal

People living with rare diseases should be entitled to the same access and quality of care as any other patients. But today the reality is far from that.

Paradoxically, rare diseases are common. There are 8,000 known rare diseases which collectively affect 7-10% of our population, three quarters of which are children. That equates to somewhere between 1.2 million to 2 million Australians – similar numbers to those suffering from diabetes. Up to 400,000 are estimated to be children.

30% of children with a rare disease die before the age of 5. 80% of rare diseases have identified genetic origins. Obtaining a definitive diagnosis is often difficult and delayed. Rare diseases are severe to very severe, chronic, often degenerative and life threatening.

Neurological and intellectual disabilities occur in about half of all cases regardless of disease type and lead to a loss of independence and opportunities. Most begin in childhood and continue throughout life. Many rare diseases have no cure.

In Europe a disease or disorder is defined as rare when it affects less than 1 in 2000 people. One rare disease may affect only a handful of patients in the EU, and another touch as many as 245,000. On the whole, rare diseases may affect 30 million European Union citizens. (eurordis)

History of Rare Disease Day

February 29 2008, a “rare day”, was the First Rare Disease Day in Europe, organised by EURORDIS (European Organisation for Rare Diseases). For the first time patient groups from different countries and representing a variety of diseases collaborated on a large-scale awareness raising campaign in favour of rare diseases.

28 February, 2011 marked the fourth International Rare Disease Day. Hundreds of patient organisations from more than 40 countries worldwide organised awareness-raising activities converging around the slogan “Rare but Equal”.

The GSNV hosted an afternoon tea in support of Rare Disease Day 2011.

We invited our members, friends and colleagues to join us in thinking about rare disease as a public health priority.

The GSNV is well aware of the challenges rare disease patients and their caregivers face and wished to mark the day for them.

The event was a success with representatives from various support groups and a number of health professionals attending.

GSNV staff had the opportunity to make connections, discuss some of the issues surrounding rare disease, and think about how we can continue to best represent and support individuals and families affected by rare disease in the future.

**IN BRIEF**

**WANT TO BE ON OUR EXECUTIVE?**

Results from our online survey suggest you believe there are too many professionals on our executive committee.

If you are a member of a support group or someone with or a relative of someone with a genetic condition we would welcome your valuable input!

Please contact our office on 8341 6315 or email info@gsnv.org.au and you will be warmly welcomed.

**GSNV OFFICE UPDATE**

The GSNV has moved office and is in a temporary space on the 10th Floor of RCH until a major move in November 2011.

From November 2011 the GSNV will remain co-located with VCGS in larger office space. We look forward to reporting on our move.

**ACKNOWLEDGEMENT**

The GSNV is proudly supported by the Department of Health Victoria.

The GSNV thanks the Murdoch Children's Research Institute for their ongoing support of our work and the Lord Mayor's Charitable Foundation for the generous grant which has made much of our work possible.

### CALENDAR OF EVENTS

**Spina Bifida Awareness Week**

**Thursday 1 September to Wednesday 7 September**

**Spina Bifida Foundation Victoria**

Focusing on educating the community about the importance of folic acid in the diet leading up to, and during pregnancy.

**Patient Information Session, Huntingtons Victoria**

**Friday 2 September**

Hosted by GSNV

**Genetic Disorders Awareness Day**

**15 September 2011**

AGSA celebrates this day each year to draw attention to genetic disorders.

**2nd Annual National Disability Summit 2011**

Paving a future direction for disability policy reform in Australia

**15-16 September**

**Novotel Melbourne on Collins**

Hear the latest policy and funding issues from key government decision makers at IIR’s 2nd Annual National Disability Summit.

**International Keynote Addresses:**

- **UK Disability Services - From State Provision to Independent ‘For Profit’ Provision**
- **Angela Gifford**, Founding Member of the United Kingdom Home Care Association and a UK Law Society Care Expert with reference to personal injury claims since 1990.

**Keynote Addresses from:**

- **A Future Disability Support System for Australia**
  - John Walsh, Former Associate Commissioner, Productivity Commission, Disability Care and Support
- **DisCo DDA NDS and NDIS: The Landscape Post Productivity Commission Report**
  - Graeme Innes AM, Disability Discrimination Commissioner


**NFAA 2011 Family Camp**

**16-18 September**

**Stanwell Tops Conference Centre, near Wollongong**

Put the date in your diary and plan to come along. Every year the camp gets bigger and better.

**Down Syndrome Awareness Week**

**Begins 15 October 2011**

**Buddy Walk**

**16 October 2011**

Buddy Walk is an international walk-a-thon style event aimed at raising funds for and awareness of Down syndrome. There are more than 260 Buddy Walks all around the world. Buddy Walk – Melbourne will be held on 16 October at Princes Park. Horsham and Bendigo will also hold a Buddy Walk in 2011 (dates TBC). More information, including how to set up your Buddy Pages, will be available soon. For further information on any Buddy Walk event, phone 1300 658 873 or email fundraising@dsav.asn.au

**Kawasaki Disease Information Session**

**Saturday 16 October**

Royal Children’s Hospital, Melbourne

Guest speakers: Dr. David Burgner – Murdoch Childrens Research Institute, Dr. Nigel Curtis – Paediatrician R.C.H, Dr. Michael Cheung – Cardiologist R.C.H.

The afternoon will start at 12.30pm with lunch and close about 5.30pm following an open discussion/questions and answers session.


**Williams Syndrome Family Support Group**

**2011 Family Camp**

**21-23 October**

**Adanac, 47 Hoddle St, Yarra Junction**


**International Day of People with a Disability**

**3 December**