

# genetic support network of victoria

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**ANNUAL  
REPORT**  
2013 – 2014



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WELCOME TO THE GENETIC  
SUPPORT NETWORK OF VICTORIA INC. (GSNV INC.)  
ANNUAL REPORT FOR THE FINANCIAL YEAR  
ENDED 30 JUNE 2014

As we look at the year in review 2013-2014 we will showcase some highlights, achievements, challenges and outcomes of the GSNV Inc. including our Governance and Financials.

## EMPOWERING THE GENETICS COMMUNITY

From those early years of helping with the establishment of face to face support groups and building a database to connect people impacted by a genetic diagnosis, our work continues today helping the genetics community and engaging in psychosocial research. The GSNV hasn't stopped for almost 17 years.

This annual report for the period 1 July 2013 to 30 June 2014 is a celebration of the power of the genetics community through the commitment and care that our people and supporters provide every day in Victoria and into other states.



### OUR VISION

Purpose, dignity and choice for people with genetic conditions

### OUR MISSION

To influence direction and policy that empowers people, organisations and institutions that serve people touched by genetic conditions.

To develop the leadership of support services and stakeholders.

### THE GSNV IN CONTEXT

The GSNV has operated in a changing and challenging environment. This continued in 2013-14 – what we did change was the way we chose to embrace the fluid nature of the environment.

We concentrated on how we could meet the information needs of the community who now have increased expectation about what can be delivered in genetics health because of new technologies and increases in testing and diagnosis capability.

The landscape the GSNV now physically operates in includes major research institutes and clinical facilities in the Parkville/Melbourne precinct. This is building upon a focus on genomics and genetic expertise in Melbourne and developing a geographic centre of excellence.

For the GSNV, it is envisaged that this growth will indicate an upward trend in demand for our services and increasing pressure on our resources.

The concentration of clinical expertise further increases the focus on GSNV as having a vital role representing consumers and establishing a firm base from which their voice can be heard.

The GSNV will continue to focus on the important synergy between clinical service areas, research and consumers; we recognize our role is clearly to represent the people and to ensure continuous improvement in all areas of genetic services including service, care and support.

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## THE YEAR IN REVIEW

IN THE 2013-14 FINANCIAL YEAR,  
OUR VISION AND MISSION HAVE LEAD  
TO OUTSTANDING ACHIEVEMENTS.  
HERE'S A SNAPSHOT...



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## ABOUT THE GSNV

At all times the GSNV is guided by a set of core values and the overarching principle that we serve the Victorian genetics community.

### OUR VALUES

#### Integrity

We are ethical, tolerant and strive to deliver

#### Respect

We respect diversity and promote diverse opinion and approaches

#### Empowerment

We strive to empower people to overcome the challenges of living with a genetic condition or caring for someone with a genetic condition and share their lived experience to inspire others

#### Connectedness

We are committed to fostering ongoing partnerships and relationships in support of our vision

To fulfill our mission we aim to:

#### Empower

- Empower individuals and their families to reach positive health and life outcomes
- Empower people to act as community representatives
- Represent the interests and views of individuals affected by genetic conditions to the community as well as the State and Federal governments
- Promote consumer participation and feedback
- Provide educational opportunities to individuals and their families, health professionals and the wider community
- Advocate on behalf of others

#### Connect

In order to support people affected by genetic conditions the GSNV aims to connect:

- Individuals/Families
- Service Providers
- Community
- Government, and
- Health Professionals

#### Support

The central aim of the GSNV is to provide a point of contact for individuals and families seeking information and support.

We do this by:

- Providing timely, accurate and balanced information
- Referring to support groups and other community services
- Referring to clinical services
- Facilitate peer support
- Assisting with the establishment of new support groups and the expansion of existing groups
- Providing educational opportunities such as workshops, seminars, patient information sessions and peer support training
- Supporting clinical services and the delivery of quality healthcare

#### WHO DO WE SERVE?

- People who are affected by a genetic condition
- Support Groups and Stakeholders who represent people who are touched by genetic conditions

#### WHAT DO WE NEED TO BE TO SERVE SUCCESSFULLY?

- Strong, clear and consistent
- Connected to influential people
- Representative: informed by lived experience
- Smart, discerning and current: informed by research and best practice
- Respectful and grateful, active and engaged
- Know when to lead and when to follow



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THE GSNV SUPPORTS VULNERABLE PEOPLE IN OUR COMMUNITY

For almost two decades we have been focused on improving the lived experience of Victorians impacted by a genetic condition or genetic testing and diagnosis.

In striving to reduce isolation and vulnerability in the genetics community we connect families and individuals sharing a common experience, maintain a support group and network database, provide education and support at the community and professional level.

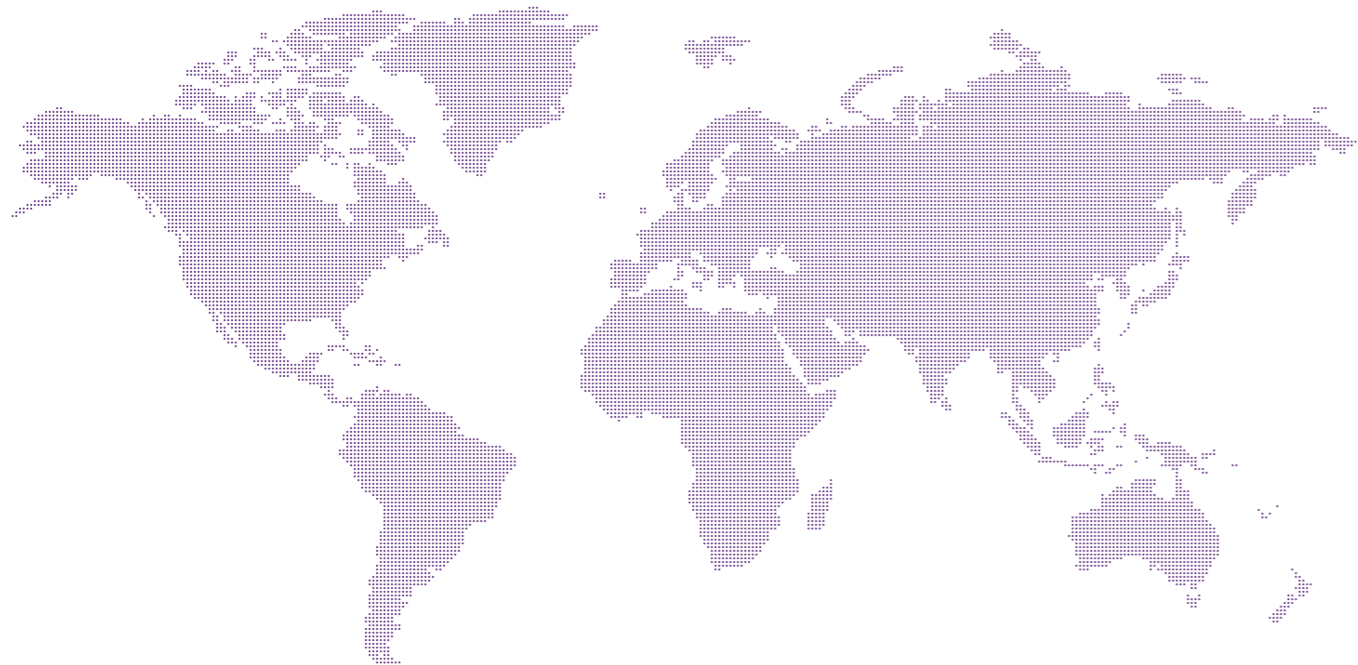
We help people in crisis, empower those motivated and working hard to represent a condition specific community, advocate and represent on behalf of our members. We help people prepare, respond and recover from a new diagnosis and work with those caring for others.

The GSNV is a member of the Human Genetics Society of Australasia (HGSA) and Special Interest Group (SIG) and the HGSA Victorian/Tasmania branch.

The GSNV is one of hundreds of genetic support and professional societies around the world.

- International Congress Human Genetics (ICHG)
- American Society of Human Genetics
- European Society of Human Genetics
- NZOrd
- EuroDis
- UK Alliance
- Genetic Alliance US

As a Victorian peak body, we work with interstate and international networks, government, research and public organisations in the genetics sector to provide our services.



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OUR PEOPLE

GSNV ORGANISATIONAL CHART

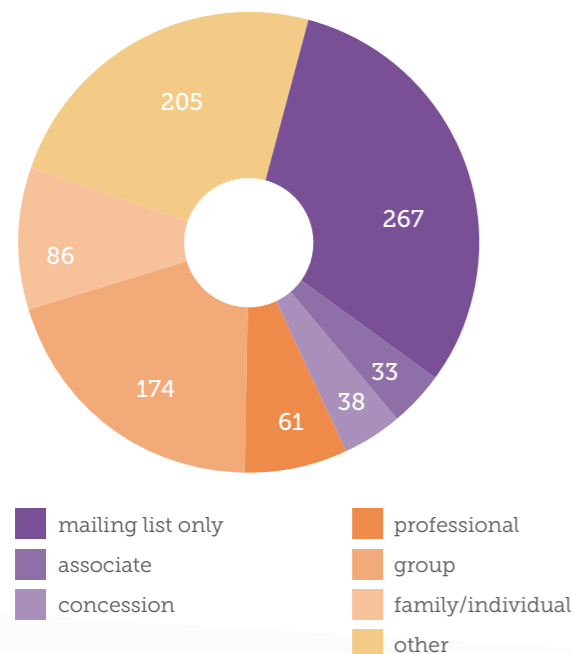


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## OUR MEMBERS

GSNV members are the backbone of our organization, providing feedback, ideas and contribution to enhance and support our work. The GSNV would like to say thank you to all members and we look forward to a continuing positive relationship.

### MEMBERS COMPOSITION 2014



The GSNV has a wide range of members, consumers and professional affiliates. These include:

### GENETIC AND ALLIED HEALTH

- Clinical Geneticists
- GPs
- Researchers
- Social workers
- Genetic counsellors
- Allied Health Professionals

### GOVERNMENT BODIES

- Victorian Health Department (DoH)
- Public Hospitals
- Local Councils
- General Practice Divisions
- Funding Bodies
- DHS

### COMMUNITY

- Individuals
- Families
- Carers
- Students
- Community groups
- Maternal Health Centres
- Early Intervention Education
- Corporate business
- Charitable Organisations
- Philanthropic societies
- Culturally and linguistically diverse groups

The GSNV currently has 865 members

### NETWORKS

- Not for Profit Organisations
- Support and Advocacy Groups
- International Advocates and Alliances
- Health Networks and Organisations

GSNV membership offers the opportunity to gain access to information on research, support group activities, patient information, peer connection, advocacy and education and to be represented on issues relating to genetic health.

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## OUR COMMITTEE

### PRESIDENT'S REPORT

On behalf of the Executive and Committee of Management of the GSNV Inc. I offer up the follow report to the members.

*"There is no immediate answer for every genetic illness out there. Without the research, support and dedication it will not happen on its own... we need help where ever possible. As they say... more hands make light work of things."*

Kay Timmins

It has been a busy year to date, with lots of changes and tweaks and the introduction of more efficient processes within GSNV.

A big focus this year has been to improve our financial position and work towards maximising the benefits of a membership fee structure which we will update in the New Year. We've put in the time to apply for grants and established a financial sub-committee (FSC) with terms of reference developed to focus purely on money matters.

The FSC has worked hard and in support of our goals is determined to secure a viable and prosperous future. I thank the FSC for their generous commitment of time and skill. I particularly thank Hanna Leslie for all her hard work on identifying grant opportunities and excellent grant applications. Hanna has set the bar high and established a foundation we can build on for future applications.

We have had a good working committee, all very committed the vision of the GSNV and working through a long list of actions to realise our goals. I thank them all. Along with the staff, also working hard we are all in the same boat and have paddled hard (the boat has rocked but no chance of sinking)...we share a vision.

Unfortunately we have had to say goodbye to 2 of our committee members, Charlotte Stockwell and Margherita Coppolino. Both have stepped down due to work and other commitments but we thank them for all their hard work, contribution and dedication to the GSNV.

The GSNV staff have been very busy with lots of movement regionally. Maree Kinniburgh (committee member) and GSNV Staff (Louisa and Keri) have been touring the state giving talks to health professionals in Sale, Mildura, and Swan Hill. These talks have been an important development in Health Education and Learning in regional Victoria, giving health professionals a fuller understanding of genetic referral pathways and how clinics run in the city.

A highlight for the year was an overseas trip undertaken by Louisa (Group Leader) to represent the GSNV at the European Society of Human Genetics (ESHG) and the Psychosocial Aspects of Genetics (EMPAG) meetings in Milan.

This was an exciting opportunity for Louisa to be presenting author of research conducted by Emma Swain, who is a current GSNV committee members and Graduate of the Master in Genetics Counselling (Melbourne University) course. Emma's research provided an opportunity to represent the Victorian, Australian context in genetics support.

Overall the GSNV staff have been kept very busy with communications, education, training, providing support, new staff inductions and training, an office move (temporary) and keeping up with everything in general. It's a busy office!

The GSNV website is an excellent portal with lots of information on our work, what our networks and support groups are doing and particularly what's on and where. The GSNV staff update the GSNV events calendar weekly so if you want to know what support groups are up to and what events to attend, please go to [www.gsnv.org.au](http://www.gsnv.org.au)

Kay Timmins

Kay Timmins  
GSNV President



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WHO WE ARE

The members of the Executive and Committee who held a position at any time during or since the previous AGM are:

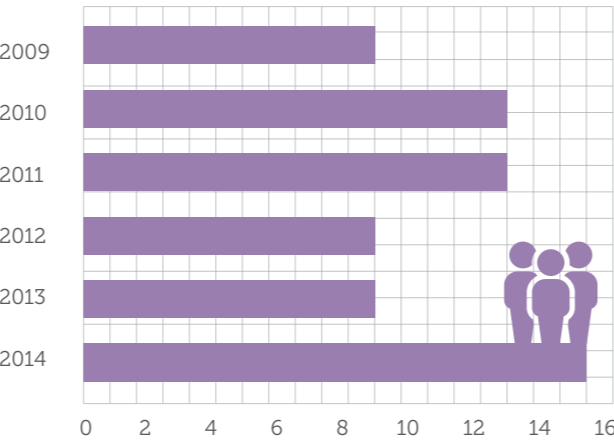
Kay Timmins	President
Shona Malberg	Vice President
Rachel Pope-Couston	Treasurer
Monica Ferrie	Secretary
Abbie Kinniburgh	Committee
Charlotte Stockwell	Committee (17/10/2013 – 19/06/2014)
Christine Williams	Committee
Doreen Floyd	Committee
Emma Swain	Committee
Hanna Leslie	Committee
Katarina Radonic	Committee
Maree Kinniburgh	Committee
Margherita Coppolino	Committee (17/10/2013 – 08/08/2014)
Marie Dunn	Committee
Moiray Rayner	Committee

The Committee of the Genetic Support Network of Victoria (GSNV) Inc. operates through an Executive and general committee with delegated authority and terms of reference according to the Associations Reform Act 2012 Model Rules. In line with our essential role in the community and our status as an Incorporated Association, GSNV Inc. is committed to maintaining the highest standards of corporate governance.

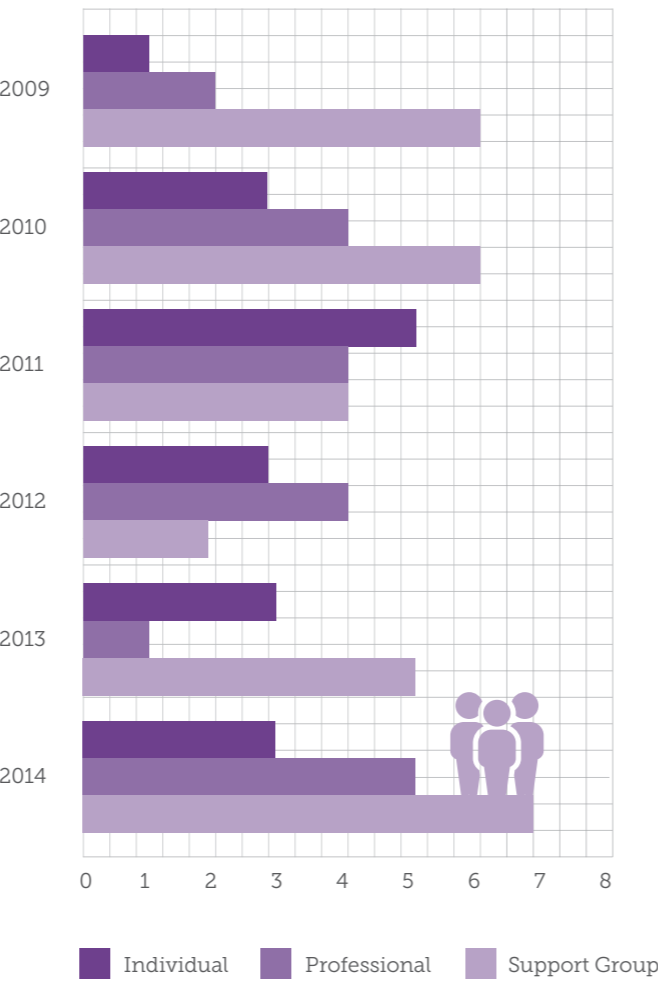
The GSNV Committee is responsible for the governance of GSNV Inc. and establishes the key strategic priorities and organizational performance indicators. Key business objectives are delegated to the GSNV Group Leader and staff, who work under an operations and line management process facilitated by the Victorian Clinical Genetic Services (VCGS).

Thank you and goodbye to Charlotte Stockwell and Margherita Coppolino who have both retired from the Executive during the past financial year.

COMMITTEE MEMBER GROWTH



COMMITTEE COMPOSITION 2009-2014



CORPORATE GOVERNANCE

The 2013-14 year has been a year of corporate and operational consolidation.

The GSNV adopted the Associations Reform Act 2012, New Model Rules in November 2013 providing an even stronger governance foundation.

The Committee of Management expanded to include 14 members bringing increased representation, expertise and resources. It remained a key goal of the Committee to ensure diverse representation including individuals living with and affected by genetic conditions, health professionals, researchers, educators and interested members of the broader community.

In the period since the previous AGM, the GSNV Executive and Committee have met on the third Thursday of each month at the Murdoch Childrens Research Institute (MCRI). Each month a key theme, supporting the achievement of strategic and business objectives, has been addressed and actions taken to deliver relevant and required documentation, focus and outcomes.

In 2013-14, these have included:

- Advocacy: Influence and Reputation
- Financial and Risk Management
- Service and Accessibility
- Marketing, Fundraising, Engagement and Image
- Leadership Development for Support Groups
- Rural and Regional Victoria

The agenda for Committee meetings is prepared in conjunction with the Group Leader and the Secretary. Reports and documents are distributed to Committee members in advance of the meeting and each Committee member is free to raise agenda items and general business.

On occasion in the previous year the GSNV Committee has invited external parties to attend meetings and has sought or taken external advice, when considered appropriate.

A Finance Sub-Committee has been operational in 2013-14, bringing increased financial rigor and reporting, risk assessment and management as well as a focus on fundraising through grants.

We have consolidated our governance base and look forward to a productive and exciting year in 2014-15.



"I could not have been happier with the meeting and am really looking forward to working with you... I must thank you too for your excellent manners and the way you looked after me with the driving directions, carrying my computer back to my car ALDS is yet again blessed!"

"It was a great experience, very rewarding and enriching. Although confronting, it has only strengthened my resolve to try to help out and make a difference. I've subscribed to the MDA's newsletter too"

STAFF AS AT NOVEMBER 2014

The GSNV has a recruitment policy that focuses on meeting our salary budget and maximising the staff profile to access diverse skills and experience. We seek opportunities to employ and build an expanding range of skills in core functions.

In the period between October 2013 to November 2014, the GSNV has continued to experience staff and workforce planning change. This year we have welcomed Keri Pereira (Genetic Support and Education Coordinator), Anna Jarmolowicz (Administration) and Nandini Somanathan (Administration).

We are most pleased to introduce Keri Pereira as a new genetic support and education coordinator. Keri has completed the Master in Genetic Counselling and is MHGSA qualified. The role of the genetic support and education coordinator includes processing and responding to enquires that are received by the GSNV. It also involves delivering presentations to a variety of different audiences on topics related to genetics.

Keri has a concurrent research role as a clinical research assistant on the Collaborative Autism Study, a project investigating the genetics of autism. These two roles complement each other as the skills required for a clinical research role are quite similar to those required for the GSNV role and vise versa. Both roles require empathy and understanding of how a genetic condition can impact on an individual or family.

The GSNV currently operates with 1.8 EFT (equivalent full time) with all staff members classified as part-time and/or casual. The GSNV continues to actively recruit graduates and students from the Master of Genetics Counseling Course (University of Melbourne) resulting in a high staff turn over, managed to positively impact operations. Our staff members often move on to pursue career related roles in Genetic Counselling.



The GSNV Committee recognises the transient nature of the GSNV workforce and appreciates the enormous contribution and support all current staff have made over the past year. Their dedication, passion and commitment to the work of the GSNV and consumers has ensured outstanding outcomes. Thank you.

We also take this opportunity to extend a formal thank you to former GSNV staff; Marleen Susman, Emily Higgs, Emma Swain and Catherine Beard for their contribution and fantastic work with the GSNV. Emma, Catherine and Emily continue to have an active role with the GSNV with Emma on the GSNV Committee of Management and Emily and Catherine liaising with the GSNV in their new roles as genetic counsellors. We particularly thank Emily and Catherine for continuing to be available to GSNV staff for ongoing professional support.

The GSNV follows policies and procedures as established by the Victorian Clinical Genetics Services (VCGS) for recruitment, performance reviews, evaluations and all other HR matters. GSNV staff come under the line management of Associate Professor David Amor – Director VCGS. We would like to take this opportunity to say thank you to David and his Team, particularly Suzanne Watson-Hayes for their support and assistance in operational matters.

Professional development remains a key focus for all GSNV staff and in 2013-14, this focus has provided many positive outcomes.

EFT Allocation as at 20 November 2014			
NAME	POSITION	CLASSIFICATION	EFT
Louisa Di Pietro	Group Leader	PT	0.8
Keri Pereira	Genetic Support Coordinator	PT	0.5
Anna Jarmolowicz	Administrative Assistant	CS	0.2
Nancy Amin	Administrative Assistant	CS	0.2
Nandini Somanathan	Administrative Assistant	CS	0.1
TOTAL			1.8
TOTAL Budgeted EFT			2.2

PROFESSIONAL DEVELOPMENT

The GSNV recognises the value of the ongoing professional development of our staff and committee. Examples this year have included:

Staff Member	Program
Louisa Di Pietro	<b>5 day initial OHS training for managers</b>
Louisa Di Pietro	<b>DNA of management, update training</b>
Louisa Di Pietro (Presenting Author)	<b>European Society of Human Genetics European Meeting on the Psychosocial Aspects of Genetics Conference (Milan)</b> <i>It's a very lonely path: Exploring experiences of establishing a genetic support group in Victoria</i>
Keri Pereira, Anna Jarmolowicz, Nancy Amin	<b>HGSA 38th Annual Scientific Meeting (Adelaide)</b> The theme of the conference was <i>The Changing Face of Genomic Medicine</i> , which explored treatments of genetic disorders, the changes in practice of genetic medicine through risk prediction and genome wide detection technologies.
Keri Pereira	<b>6th National Paediatric Bioethics Conference (Melbourne)</b> The theme of the conference was <i>Things that keep us awake at night</i> . The GSNV was interested in presentations including <i>Sequencing a child's genome – what should we do with the 'grey' findings?</i> and <i>Moral distress, ethical uncertainty and mere tragedy</i> .
Keri Pereira	<b>Melbourne Genomic Health Alliance Seminar</b> The seminar focused on the impact of genomics on clinical care now and in the future. Discussion of the fundamentals of genomics and next generation sequencing and the impact these technologies can have on diagnosis, prognosis and treatment was facilitated in an interactive and entertaining debate style.
Keri Pereira, Anna Jarmolowicz	<b>Grand Rounds</b> Grand Rounds is a weekly seminar held at the Royal Children's Hospital. It is a flagship educational meeting designed to be informative and dynamic, with weekly guest speakers from around the world.
Keri Pereira, Anna Jarmolowicz, Nancy Amin, Nandini Somanathan	<b>VCGS Friday Functional Genomics Seminars + Clinical Laboratory Interface Meetings</b> Fortnightly seminars are organised through the Victorian Clinical Genetics Services, aimed at the clinicians and laboratories who work within the genetics sector. The meetings raise thought and discussion around particular cases, new research, new approaches as well as community issues.

Committee Member	Program
Rachel Pope-Couston	<b>HGSA 38th Annual Scientific Meeting (Adelaide)</b> The theme of the conference was <i>The Changing Face of Genomic Medicine</i> , which explored treatments of genetic disorders, the changes in practice of genetic medicine through risk prediction and genome wide detection technologies.
Shona Malberg	<b>Getting to Yes workshop</b> The course delivered skills for negotiating agreements and resolving disputes. Shona has a template, tips and articles which can be shared across GSNV and with any groups who would like some new techniques or tips when negotiating outcomes.

\* All GSNV new recruits must successfully complete compliance training modules under the i-manage and i-know systems under the on-boarding process

OUR VOLUNTEERS

The GSNV launched a successful Volunteer Program in May 2013. A success of the program has been its addition and recognition under the Masters of Genetic Counselling (MGC) program (University of Melbourne) accreditation.

This program is an opportunity for support groups to source suitable volunteers to help them with various small tasks. Volunteers are students in the Masters, or students interested in applying for the Masters and seeking some hands-on experience in the genetic health and support community.

THE GSNV VOLUNTEER PROGRAM

We currently have 11 volunteers and 9 support groups involved. Potential volunteers apply through the GSNV website and are then invited for an orientation at the GSNV. Volunteers are then matched with a support group based on mutual skills and interests. Some examples of volunteer-support group matches are:

Volunteer organisation	Volunteer match	Activity
Australian Leukodystrophy Society	1	Assistance with newsletter
Friends of Sammy Joe Foundation	1	Assistance with database
Muscular Dystrophy Association	1	One day on-site
Niemann-Pick C Disease Foundation	1	Writing condition-specific information to provide health professionals
CleftPals	1	Assistance with mail-out
Syndromes Without a Name	1	Assistance with mail-out
Australian Alopecia Areata Foundation	1	Promoting awareness in hairdressers
Aus. Gorlin Mutual Support Group	1	Website design
PKU	1	Assistance with fundraising

The GSNV has received encouraging feedback from both parties and is continuing to recruit more participants.

"The volunteers have been professional, punctual and a great resource for us. We are extremely grateful for your support and encourage other smaller organisations to speak with the GSNV about your needs as you are not alone"

- President of Support Group on the Volunteer Program

"The personal and professional growth attained during volunteer experiences are unique and valuable"

- Student on the Volunteer Program

"Please thank our volunteer for the wonderful work she has done on our web page: It looks so much better."

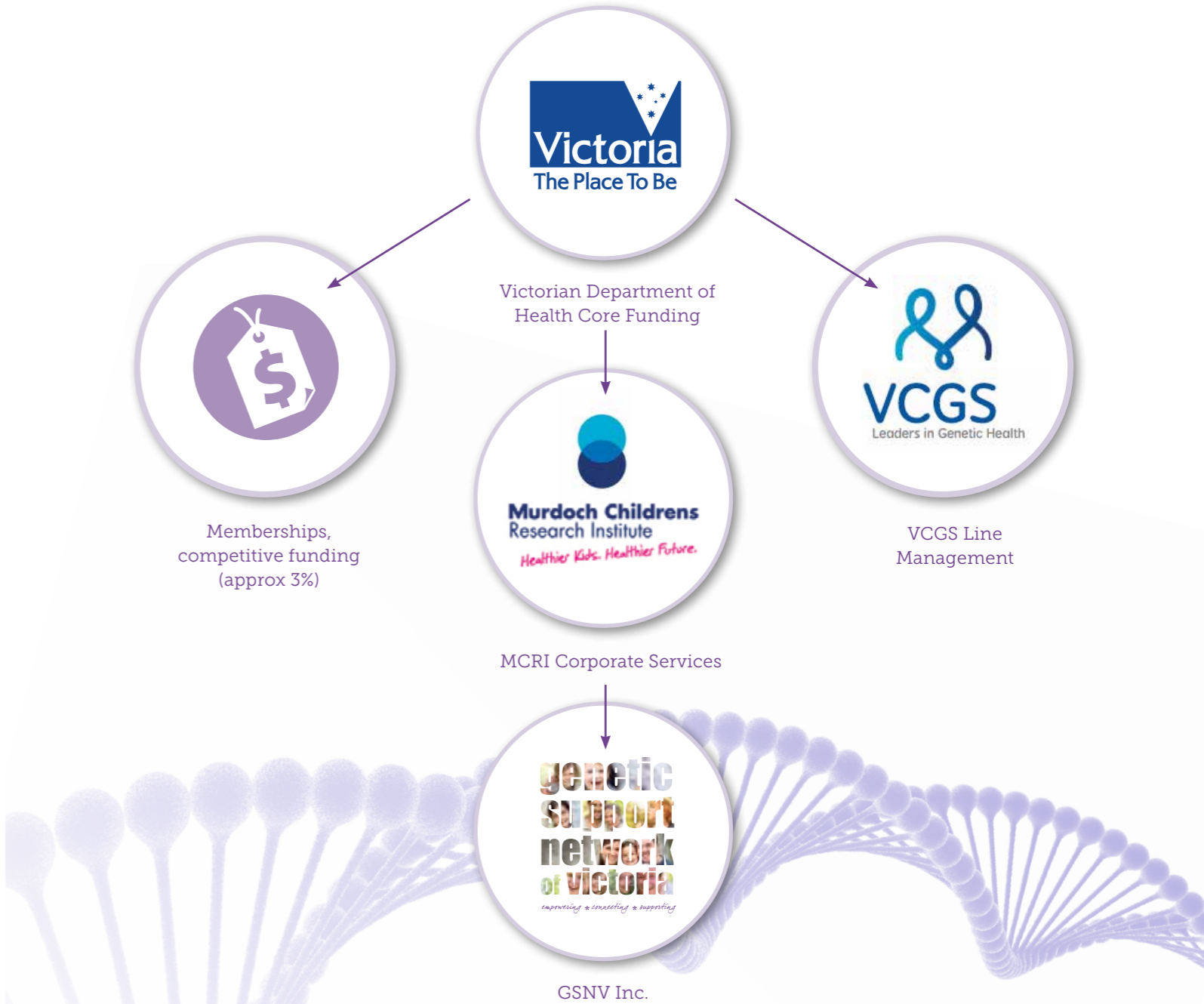
"The banners you assisted us with are absolutely fantastic. We really have gone up market now. Thank you very much."

STAKEHOLDERS

The GSNV team has continued to work closely with the Murdoch Childrens Research Institute (MCRI) and the Victorian Clinical Genetics Services (VCGS) and the Department of Health (DOH) Victoria to create open, transparent and effective corporate relationships that are mutually beneficial. As major stakeholder groups, each plays an essential role in the sustainability of the organization and the execution of key business functions and operations.

We work very hard to secure sufficient and sustainable funding to meet our commercial obligations and expand our work form year to year. This is a particular challenge in the coming years as our work has become increasingly more expansive to run and the demand on our services has expanded exponentially.

STRUCTURAL/FUNDING ARRANGEMENTS





## MESSAGE FROM THE GROUP LEADER

Over the last 12 months, the GSNV has continued to support Victorians impacted by a genetic condition and engaged with our interstate counterparts to ensure all Australians are getting the support they need.

With demand for our services increasing, our everyday work wouldn't have been possible without the generous support of our members, stakeholders, volunteers, staff and supporters who have given their time, resources and energy.

The GSNV has been working in Victoria for nearly two decades and we have a long and full history of people helping people. This year we renewed our focus on volunteering, rare diseases and genomics, all of which will inform our future direction in this important era of community engagement in research and support, in order that we provide ethical and quality services.

As we review the year that has passed I am already focused on all that our organisation can do to continue to build our strengths, encourage community engagement and empower vulnerable people impacted by a genetic condition next year and beyond.

### HOW WE DO THINGS

How we do things is very important to the GSNV, perhaps just as important as what we do. Our strategic objectives apply and inform our work all of the time but are guided by our underlying philosophy:

- Acknowledging our vision and mission
- Working effectively with our stakeholders
- Working for the Victorian community

- Building our strength and capacity
- Working strategically
- Efficient use of our resources
- Empowering the genetics community and support groups
- An evidenced based approach
- Advocating for change and improvement
- Mobilising people
- Inclusive and engaging

### OUR GOALS

The GSNV works toward a sustainable future and we want to remain strong and influential. How we organise ourselves to do this ongoing is by looking to our strategic goals:

- To strengthen our reputation as an expert voice and advocate effectively
- To have a constant living relationship with people with genetic conditions
- To strengthen support groups and stakeholders
- To be a committee of excellence
- To be a best practice organisation

In addition to our ongoing traditional work in key areas such as education, support and advocacy we have also undertaken additional pioneering work in a range of areas. These will have influence on strategic and operational matters in the coming months.



### RECOGNISING CULTURAL AND LINGUISTICALLY DIVERSE COMMUNITIES (CALD)

In view of changes in immigration policy and increasing numbers of asylum seekers in Australia, the GSNV has considered its policy on cultural and linguistically diverse communities and how we might address one of the most vulnerable groups of people in our society.

In developing the basis for a full CALD policy, the GSNV committee has reflected on the fact that our services are based on need, should be without discrimination and irrespective of culture.

We have developed the basis for a CALD strategy that helps us deliver our services with sensitivities applicable to all cultural and religious groups. We see it as our responsibility to respond to this issue of diversity and will continue to seek to improve and benchmark our services in order that we remain inclusive and responsive.



### INVESTING IN THE SUPPORT OF PEOPLE SUPPORTING EACH OTHER

The GSNV is committed to the empowerment of support groups in order that we can increase the capacity of the genetics community to support each other. With the facilitation of support group education and information sessions and the opening up of our Small Grants Scheme again this year, the GSNV has invested in our most important stakeholder group and contributed to the improvement and delivery of support group outcomes.

In May this year the GSNV and the Victorian/Tasmanian branch of the Human Genetics Society of Australasia (HGSA) financially supported the visit of Anne C. M. Smith (genetic counsellor who helped first describe and diagnose Smith-Magenis Syndrome) to Melbourne.

Anne's visit was designed to coincide with the Camp Breakaway SMS family camp held in NSW. Anne Smith conducts this camp every year in NSW, but had approached the GSNV last year in relation to a visit to Melbourne which had not occurred before. This was important event for SMS families in Victoria made possible by a grant provided by the GSNV and the HGSA Vic/Tasmanian branch. In bringing this important visitor to Melbourne, the GSNV also opened up the awareness of SMS and engagement with families.

Every year the GSNV offers Small Grants to members for projects that assist the work of support groups. Small grants are usually awarded to financial members of the GSNV as memberships provide the pool of funding for the grants scheme.

Small Grants may be used for a once-off project or activities that support groups wish to undertake and deem as a benefit to the community. Examples from past years include printing brochures, venue hire, event organisation, event catering, and purchase of resource material and library books to contribute to a program.

Small Grants are not awarded for the purchase of aids and equipment. This funding is designed to provide once-off financial assistance to support groups who find it difficult to obtain funding via other grants schemes.

GSNV grants may also be used towards administration costs for unfunded agencies. We look forward to announcing our Small Grant recipients in December of this year.



### WORKING IN PARTNERSHIP TO INCREASE OUR RESEARCH PROFILE

Working in partnership with our national and international partners underpins much of our work in the delivery of support services and in keeping abreast with best practice and research in genetics. This year we have taken significant steps to profile our work in international forums and to showcase the GSNV model abroad.

I am very proud to have had the opportunity to present at the European Society of Human Genetics (ESHG) and European Meeting on the Psychosocial Aspects of Genetics (EMAG) meetings in Milan, Italy. Both meetings provided an opportunity to learn, network and engage with professionals from all over the world.

As presenting author of a research paper developed by Ms Emma Swain, I presented research that looked specifically at the experience of setting up a support group in Victoria and the experience of interacting with the GSNV in doing so.

A particular focus for the coming year will be on the development of a significant research paper developed by the GSNV team and presented at national and hopefully international meetings.

The GSNV is keen to publish independently and will work over the coming months to get ethics approval and begin our research project. Collaboration with expertise proximate to the GSNV and external is an important element in our capacity to develop quality work and we will seek to engage expertise as required.



# genetic support network of victoria

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## RECOGNISING OUR LOGO AS AN EMBLEM OF OUR WORK AND WHO WE REPRESENT

The GSNV logo which is a montage of faces and real people has become a symbol of the significance of what we do and the importance of who we do it for...the community. More than just a logo, we have developed all our collateral with the 'faces of the GSNV' at the forefront and drawing attention to our organisation. It has been a great success, and translates very well on all media applications, electronic and print.

This year we introduce 'GSNV George' our well travelled GSNV vehicle. As a branding tool, GSNV George will lead and headline our social media campaign for a genetic support awareness week (GSAW) culminating in Rare Disease Day (RDD) and a RDD event. We are very excited about the information and flurry of activity that will be delivered during the last week in February. We have MCRI on board as a partner this year and look forward to Kathryn North, Director MCRI opening our RDD event and addressing our guests.

We have travelled 1,148km in Victoria and 1,877km Nationwide



*empowering \* connecting \* supporting*



## STRENGTHENING OUR REPUTATION AS AN EXPERT VOICE AND ADVOCATING EFFECTIVELY

The GSNV has continued to embrace every opportunity to provide a voice for, and representation of individuals and families affected by genetic conditions.

In our work providing consumer feedback, comment and position statements on a wide range of issues including access, equity, health reform, management, support and systemic reform, our aim is to represent the genetic perspective in the overall approach to health, health delivery and the quality of life.

As part of our work at the governance level we have reflected on the current context and the changing landscape guiding the debate in genetics health and the needs of consumers. We look forward to rolling out an updated advocacy policy in 2015.

Advocacy involves:

- Representing the interests and views of people and their families affected by a genetic condition to the community, State and Federal Government
- Providing "arms length" representation when needed
- Empowering GSNV existing members and the general community
- Raising issues and providing feedback on a range of topics



## KLINEFLETER SYNDROME (KS) NATIONAL SUPPORT AND ADVOCACY

In 2013 funds were redirected from the KS Victorian Support Group to the GSNV by the Department of Health under a Band 1 condition-specific support grant.

The GSNV has taken on an active role in facilitating the coordination of KS support in Australia. The GSNV has an interest if professionally leading KS support, as existing peer support arrangements and information is lacking and/or inappropriate.

The GSNV website now has a dedicated KS section and this will be developed further and in consultation with key stakeholders.

The GSNV has taken a very active role in facilitating the coordination of KS support in Australia and the response has been very positive. We can report a marked increase in calls to the GSNV for KS support and information, all of which we are responding on a case by case basis but also building up a body of knowledge and ideas that is populating our plan to develop a full project brief around KS support.



The GSNV has an interest if professionally leading KS support, as existing peer support arrangements and information is lacking and/or inappropriate. The GSNV website now has a dedicated KS section and this will be developed further and in consultation with key stakeholders.

Over the last year the GSNV has provided consultation and expertise in other areas including:

- Health Innovation and Reform Council (HIRC) Vic
- Human Genetics Society of Australasia (HGSA) Victorian/Tasmanian Branch
- HGSA Ethical and Social Issues Committee (ESIC)
- Consumer Health Forum
- Cystic Fibrosis Victoria Carrier Screening Reference Group
- Melbourne Genomics Alliance Consumer Advisory Group
- Individual letters of support for families seeking services and financial assistance
- Office of Population Health Genomics, Public Health and Clinical Services Division WA, Australian Survey of Adults living with rare diseases development
- Rare Voices Australia Rare Diseases Survey
- Application to Medical Services Advisory Committee (MSAC) to Have Pre-implantation genetic diagnosis listed on the Pharmaceutical Benefits Scheme

*empowering \* connecting \* supporting*



## OUR FUTURE – STRATEGY 2015

The GSNV strategic plan is the path forward for the next year. Comprising our core organisational goals, our ways of working, our enabling goals and our priority areas, it is a stand up plan. This year, we have continued working towards the goals we set in 2013, conducting an ongoing review on our progress, identifying our key opportunities and priorities and making significant steps towards enhancing engagement.

### Reflecting on our progress and moving forward

In addition to ongoing review, the GSNV has scheduled a review of Strategy for late November 2014. This is designed to reflect on our progress to date in enacting our strategic goals, and develop a plan in moving forward. An important focus at the meeting will be to assess any new and emerging internal and external factors affecting our operating environment and ways to maximise our efficiency and develop some cost recovery for our activities.

### Anticipated Recommendations

It is anticipated that the forthcoming review will see recommendations that will apply to priorities and recommendations confirmed through ongoing reflection and will include:

- Enhancing member engagement and membership growth
- Improving volunteer engagement



- Increasing our capacity to deliver services throughout Victoria
- Incorporated rare diseases into our philosophy and key messages
- Reaffirming the importance of the GSNV as a consumer representative body
- Investing in our staff and committee
- Developing effective partnerships with partner agencies, governments and stakeholders to build our network
- Strengthen our financial position

This review will also analyse any further risks to our organisation and any subsequent mitigations. Amongst these we will focus on risks to funding due to governmental change, our operating environment, or a reduction in the number of members, volunteers or supporters of or work.

OUR SUCCESSES

The energy and professionalism which has gone into improving the GSNV’s governance processes, operating procedures and strategic planning process has been the backbone of our success over the past twelve months.

There have been significant contributions from the Executive and the newly established Finance Sub-Committee, allowing me to dedicate more time to the organization.

I thank the incumbent GSNV Executive and Committee and particularly the Finance Sub-Committee for the generation of ‘real outcomes’. These achievements have provided a great platform to deliver the work of the GSNV.

Our successes have included:

- A streamlining of governance processes
- An opportunity to reflect, review and improve current practice
- Developed an advocacy strategy
- Developing a CALD policy
- Capacity analysis of staff and Committee
- Focused internally on our financial situation
- Developed a risk management strategy
- Completed a full review of our representation on external boards
- Developed a grants ‘hit-list’ and made applications
- Development of a research project pending ethics application
- Participation on the Victorian Health Innovation and Reform Council (HIRC)
- Voting membership at the Consumer Health Forum
- Successful nomination to the Human Genetics Society of Australasia (HGSA) Victorian/Tasmanian

Branch committee and invitation to the Local Organising Committee (LOC) of the HGSA Annual Scientific Meeting (ASM) Tasmania, 2016

We are also now 18 months into a formal line management arrangement with the Victorian Clinical Genetics Service (VCGS) and I see the major benefits delivered through this to include the following:

- GSNV is now operating in a highly supporting environment with supported decision making
- GSNV has access to operational systems including all corporate services
- GSNV has structured support allowing focus to be on the consumer – the confidence in the systems and structures of the VCGS allows up to focus on our core role.

With the development of a simple and practical strategic plan, a committee planner, a business actions plan and a full risk assessment, the GSNV has in the last 12 months operating with greater clarity and efficiency. The development of good business basics and a clear understanding of our organizational performance indicators and measures has strengthened our capacity.

The GSNV operating environment is however not without its challenges. A degree of change within the MCRI in terms of its corporate policy, pressures on accommodation options for the GSNV and mounting pressure to increase our budget in order to met the obligations of infrastructure and corporate services charges as a ‘Group’ in the broader VCGS structure has offered up some nail biting moments for me. I remain optimistic that we will be rewarded by the significant work put in recently for a fundraising and donations project. I also remain hopeful that we will attract competitive income with a grant or two.

Congratulations to the committee and GSNV staff for a year of successes and for the development of even stronger foundations for an improved services for years to come.

*I remain focused on improving the lives of all those impacted by a genetic condition in Victoria and mobilising the power of the GSNV network to make a difference.*



Louisa Di Pietro  
Group Leader

EDUCATION AND SUPPORT

EDUCATION

Education is a vital part of the core work of the GSNV. Education activities are grouped as follows:

- Professional
- Community
- Student
- Facilitated education, support and information

PROFESSIONAL

Health Education Australia Limited (HEAL) presentations

The GSNV presented for Health Education Australia Limited on three occasions this year. The HEAL talks were held in 3 rural locations, Mildura, Swan Hill and Sale over the course of two months.

The presentations covered the role of the health professional in genetic health, the reasons for referral to a genetics service and the ethical issues around genetic information. These talks aimed at increasing the attendees’ knowledge around genetics and why referrals to genetic services are required.

The audience for these talks included paediatricians, general practitioners, nurses, allied health professional, students and doctors specialising in other areas.

Feedback from these sessions suggested that the people that attended the session found the presentation informative and useful. Maree has shared her trip experiences with us....

*“We arrived at Gippsland Base Hospital, Sale, where I had spent many anxious weeks, decades ago when my daughter’s rare condition was discovered and treated. With a warm welcome from the local doctors and nurses, the GSNV team shared with them the services we offer to the community across Victoria. We provided an update on the*



*super science of genetics and answered questions from the floor. I presented my lived experience as a parent of a daughter with a genetic condition and reaffirmed that the doctors must trust us as parents to ensure that we all work together as an effective team.*

*A couple of weeks later Keri and I were sponsored to give a repeat performance at Mildura Hospital where we were again well received by medical professionals, in addition to a local parent. After our talk, the mother gave us an agonising account of the frustrations and difficulties of isolation and the impact on her child with a rare genetic condition. In this technological age we are so lucky to have the GSNV at our fingertips for support and while nothing beats the personal touch, it offers a very reassuring umbrella to collect people from all corners of the state. I truly hope we were of some help to her.”*

*“The feedback received was that you provided an interesting talk; covering information that health professionals can reflect upon and utilise as professionals working in the health industry. Your willingness to take the time to come and talk about Genetic Illness was greatly appreciated.”*

HEAL Coordinator

Maternal and child health nurse seminars

The GSNV was invited to present at two Maternal and Child Health professional development sessions in the last year.

The maternal and child health nurses are at the forefront of infant care therefore are an important audience in terms of raising the awareness and understanding of genetic and rare conditions.

The GSNV presented *Supporting the pathway to genetic diagnosis*, which discussed the role of genetics and its importance in primary health. The session also gave a number of case examples looking at particular conditions so the audience had a better understanding of how to assist their clients.

Genetic counsellor afternoon tea

An informal get together and chat with genetic counsellors was organised by the GSNV. This event was held at the Royal Melbourne Hospital. The intended purpose was to strengthen the relationship with counsellors and encourage referral to the GSNV.

This was a successful endeavour with measurable results after the meeting.

## COMMUNITY

### Pre-Pregnancy Planning Presentations

This year the GSNV continued to participate in the education of couples prior to pregnancy through the Pre-Pregnancy Planning seminar. These education sessions are run by the Epworth Freemasons Hospital and were held three times over 2014.

The GSNV presentation focused on genetic screening during pregnancy including the new non-invasive prenatal screening and informed decision making.

Other topics presented at the seminars included preparing your body for pregnancy, preparing your finances and infertility.

Prior to the sessions this year we reviewed our 'Pre-Pregnancy Planning' brochures and updated them to include more information about testing options during pregnancy. These brochures were distributed in show bags to all that attended the evening.

There were between 80-100 attendees at all sessions and the genetics presentations were well received. These education sessions will continue to run in 2015.

### Peer support training

Professional Counsellors are helpful, but there is nothing more powerful than talking to someone who has been "in the same boat" as you. Peer support training is available for all individuals, including staff from support groups.

The GSNV runs an annual half day workshop aimed to help individuals feel empowered to listen and talk to others who are facing genetic challenges, and to share their wisdom and experience. Training is free to GSNV financial members.

Upon completion attendees receive a certificate as well as a Peer Support contract with the Genetic Support Network of Victoria.



"Thank you for your time and commitment to the Epworth Freemasons PPP Program during 2014 – I have valued the support of the GSNV."



## STUDENTS

### Ave Maria High School Presentation

The GSNV was asked to deliver a presentation to a group of year 9 student at Ave Maria Collage. The students were investigating the concept of "designer babies" and what is currently possible and what is not. They enlisted the GSNV team to aid their research in to the topic.

The GSNV presented basic genetic concepts and raised some question around the ethics of "designer babies". A group activity was also carried out, getting the students to explore the ethical dilemmas which genetic information and engineering can raise.

The students were very engaged in the presentation and at the end created a futuristic "60 minutes" story outlining the positives and negatives of designer babies.



### Monash University – Biomedical sciences

The GSNV was again invited to present a guest lecture to the Monash University Biomedical Science students. This year the GSNV presented on the impact of genetics in society and possible career paths in genetics. We discussed with student the impact of genetic disease and testing on individuals, families and the wider community. More importantly this presentation pointed out the issues that the increased use of genetic technology may raise.

The presentation included examples of enquiries that the GSNV received which illustrated "real life" scenarios so the students could better understand the work that the GSNV carries out.



"We really appreciate the time and effort you put into preparing and giving your talk. It was a really good illustration for them of the relevance of a lot of the material they had covered in the unit."

It also gave the students a great deal of insight into what you do, which will be very helpful for them in planning their careers."



THE UNIVERSITY OF  
MELBOURNE

### University of Melbourne – Masters of Genetic counselling

The GSNV supports the University of Melbourne Master of Genetic Counselling program each year and has established a synergetic relationship with the course coordinators and student population.

Each year we conduct a number of tutorials and use the opportunity to provide students with the unique GSNV perspective on elements of their training.

- **Rapport tutorial**

GSNV presented on the topic of building rapport with health professionals. The session was greatly supported by a parent's perspective, Nicole Millis from the Mucopolysaccharidoses Society (MPS).

- **Public health genomics**

The GSNV participated in the University of Melbourne Public Health Genomics course 2014 by assessing student blogs designed to inform an newly launched blog under the MCRI banner. The GSNV will be a regular contributor to the MCRI blog and will work on the development of interesting and relevant topics for discussion.

- **Genetics and the community**

The GSNV facilitated a tutorial with the 1st year MGC students around genetics and the community. Students discussed the role of support groups and how they formed part of the community. They engaged in activities to illustrate both the positive and negative aspects of support groups in genetic health.

### University of Melbourne – Societal Issues and Personal Genomics

The GSNV participated in the University of Melbourne Societal Issues and Personal Genomics course 2014.

With a focus on personal genomics the course explored the history of genomics, eugenics, what is personal genomics, what do consumers want to know about their genomes, direct to consumer testing and the future of health services with employment of sophisticated genetic technologies.

FACILITATING INFORMATION AND SUPPORT

The GSNV offers support to individuals, families and groups in a number of different ways:

- Support, information and assistance to an average of 19 people from the general community per month
- Includes connecting individuals in similar circumstances
- Referring people to specialised service and organisations
- Health professional enquires
- Service enquiries
- Network enquiries

Through the work of our genetic support team we aim to provide a point of contact for families and individuals affected by genetic conditions to:

- find the most appropriate support group,
- ensure that they are supported in all areas,
- have appropriate support and information available to them immediately to reduce feelings of isolation and confusion and
- facilitate the mechanics of lobbying to address specific issues.

Providing support for support groups is a significant part of our role. We can aid support groups in the following ways:

- Assisting new groups in setting up
- Incorporation
- Finances
- Communications
- Corporate Governance
- Peer Support
- Information dissemination

Over the year we will have assisted the following groups:

- Rare Diseases Day – February 2014
- MPS family information day – March 2014
- Fabry/Genzyme education session – March 2014
- Syndromes without a name, undiagnosed children awareness day – March 2014
- Vascular anomalies family day – April 2014
- Syndromes without a name, Ambiguous Loss workshop – September 2014
- Neuromuscular support group webinar – September 2014
- AusDoCC parent information day – October 2014

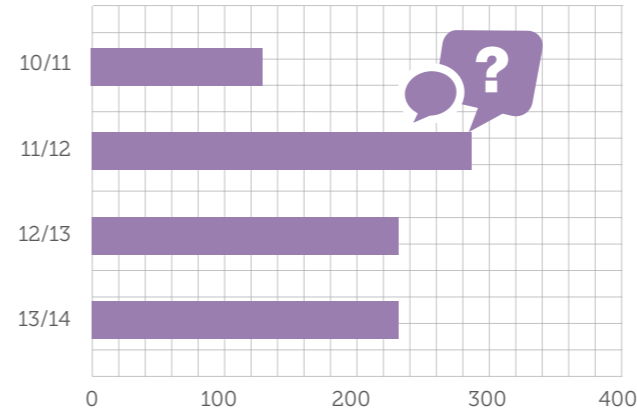
"I would like to thank you very much for the comprehensive information that you have provided me on 15q13.3 microdeletion syndrome. I have now passed this information on the parents who I am sure will be grateful. Thank you for providing such an excellent service."

"One of the strengths of the GSNV is its effective, empathetic, collaborative approach, which is often lacking in other medical/health organisations."

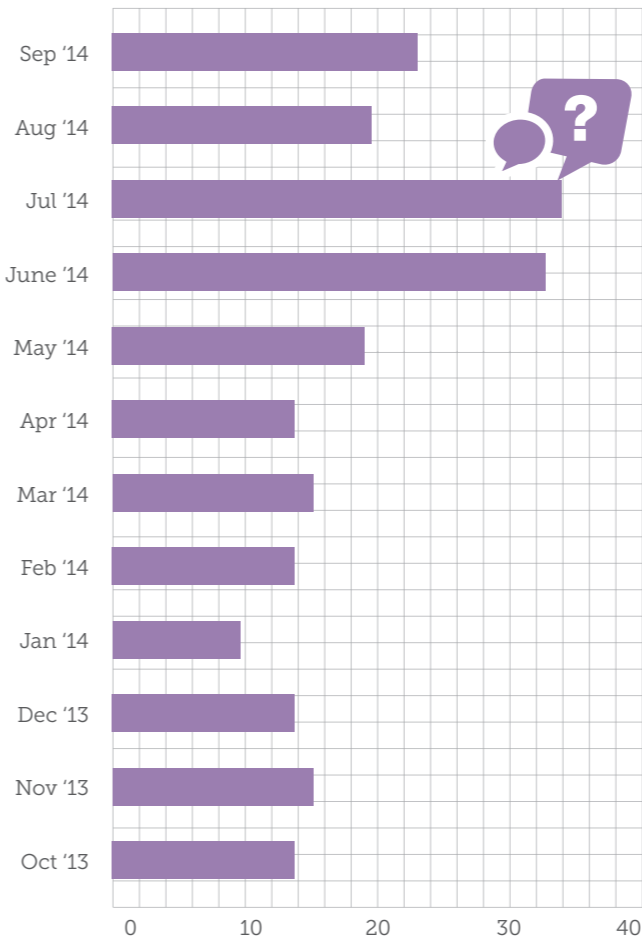
"Thank you so much for your ongoing support and commitment to AusDoCC. Having this day of information and support for parents of those with Corpus Callosal conditions and those with the condition mean so much because it is so rare in Australia."

INFORMATION REQUESTS

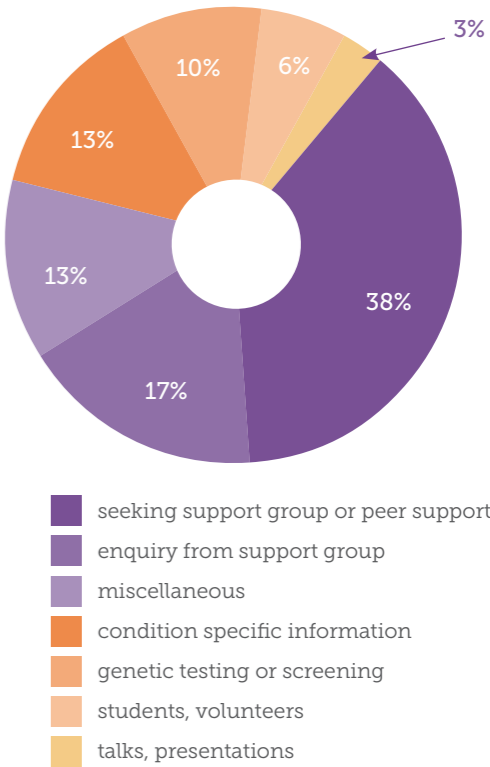
ANNUAL ENQUIRIES



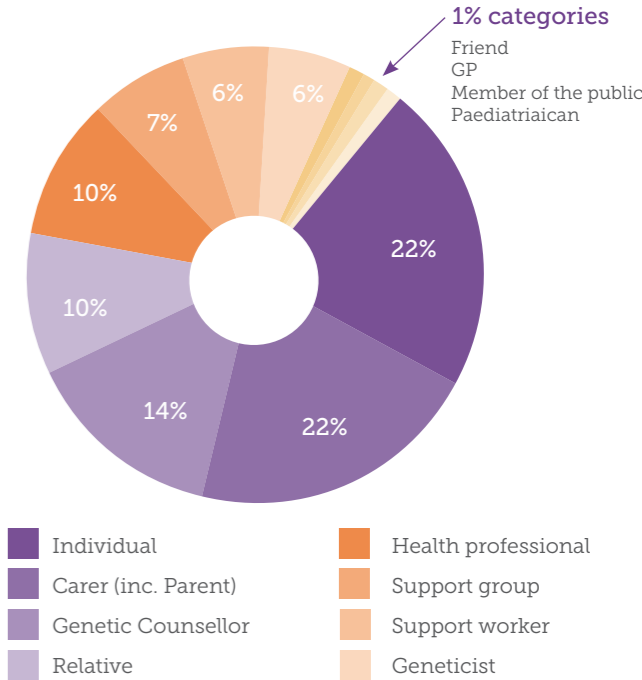
2014 ENQUIRIES BY MONTH



TYPES OF ENQUIRIES RECEIVED



WHO IS SEEKING SUPPORT?



# SNAPSHOT

## VASCULAR ANOMALIES

Supporting parents and children through improved communication and vascular anomalies service delivery.

by Zerina Lokmic

Vascular anomalies are a group of conditions that are best described as localised collections of abnormally formed blood and/or lymphatic vessels. Some are present at birth but they can appear at any stage during childhood.

Vascular anomalies are divided into tumours, such as the benign tumour infantile haemangioma, also known as strawberry birthmark, and malformations such as capillary, venous, lymphatic and arterio-venous malformations. Both types of conditions can occur anywhere in the body. Some are quite large and can have serious impact on body function and appearance.

Vascular Anomalies Clinic is a specialised unit devoted to clinical management of these conditions. Our multidisciplinary team is based at the Royal Children's Hospital in Melbourne.

The full team meets at the Royal Children's Hospital once a month to discuss patient cases and plan treatments. This is necessary as children with vascular anomalies sometimes present with complex conditions which may need more than one treatment.

In May 2014, we held our first vascular Anomalies family Day with an aim to increase an awareness of our services but also to connect the families with vascular anomalies to each other. On the day we provided basic information on vascular anomalies, how they are caused and what treatments might be available. We also asked families what we could do better to improve our services and what support services are available to them.



The most exciting outcomes from this meeting were families connecting with each other for the very first time and that we are now developing the Royal Children's Hospital Vascular Anomalies Website. Here the families will be able to find information on the conditions their child has, how to get in touch with us and how to get involved in our future work, including research.

In response to experiences the families have shared with us, we have also started developing research projects that will address some concerns families have expressed such as the adequacy of information on vascular anomalies conditions and treatments provided at the time of diagnosis, particularly during pregnancy.

Lucy Hallenstein, a Masters of Genetics Counselling student, will be joining our Vascular Anomalies team to examine experiences of parents who received their baby's diagnosis during pregnancy. Lucy will also examine what sort of information parents wished to have received at that time that would help them understand what is happening with their unborn child.

We will soon promote this project via the Vascular Anomalies Website to encourage families to connect with us and share their experiences. We hope the outcome of this study will lead to improved services for expecting parents.

If you would like to know more about our work, please feel free to get in touch with us by emailing [zerina.lokmic@mcri.edu.au](mailto:zerina.lokmic@mcri.edu.au)

## COMMUNICATIONS

At every opportunity the GSNV communicates directly with its members and networks and seeks feedback on what we are doing and how well we meet the needs of those we serve.

Feedback from our members is vital and from time to time we offer small incentives to encourage ongoing feedback and comment.

This has proven to be a great success and sample responses include:

"I always find the [GSNV] newsletter has really important bits of information or advice relevant to my situation. It's very user friendly and broadly informative."

"I enthusiastically read each addition of the GSNV newsletter. I just wanted to sincerely thank everyone that contributes to this important publication. It's such a vital service that is being provided and I'm truly grateful. I love learning about the latest research being conducted and services and support available. It makes a huge difference to families like mine and provides us with hope and a feeling of being more in control and able to handle our daughter's condition."

"Just a quick note to say well done, particularly like the article on the pros and cons of genetic testing"



### PRINT AND E FORMAT

The GSNV has over the last three years consolidated its communications into two main publications:

- A quarterly Newsletter
- Regular E-News Bulletin – 'Bits and Pieces'

Our communications are sent to more than 700 individuals, groups and professionals with our newsletter readership also extended through wider circulation amongst our networks.

Our print communications represent a significant expenditure in our budget given that the GSNV has opted for a highly professional production. We are considering cost recovery and better efficiencies for the future and will look towards strategies in 2015 to reduce our expenditure.

## We are going Green!



### NEWSLETTER COMMUNICATIONS

@ 64%  
by email

✉ 36%  
by post

64% of our communications have been electronically delivered via e-mail and 36% have been via post.

The GSNV encourage our members and community to help us become more environmentally friendly and receive our newsletter in electronic version.

Please email [info@gsnv.org.au](mailto:info@gsnv.org.au) and request to receive future newsletter editions by email.

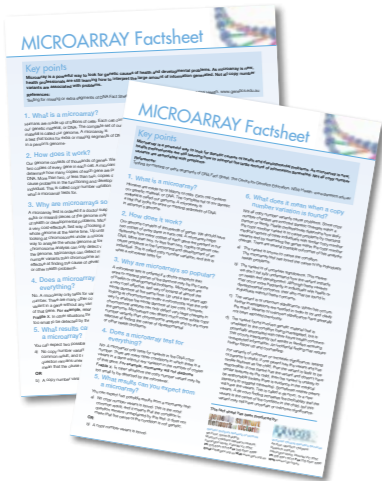
GSNV RESOURCES

The GSNV is committed to providing useful, practical and up to date information. The GSNV provide generic information packs for health professionals, disability professionals, and for the community.

The packs include a range of information sheets and brochures about genetic testing, pathways to genetic diagnosis, and access to services. These packs have played an essential role in raising the profile of the GSNV as a peak support organization. These information pack support our presentations to various health professional and community groups.

This year the GSNV along with VCGS has developed a microarray factsheet aimed at informing clients and health professionals about this increasingly common genetic test.

A full size version of the fact sheet is located on page 32.

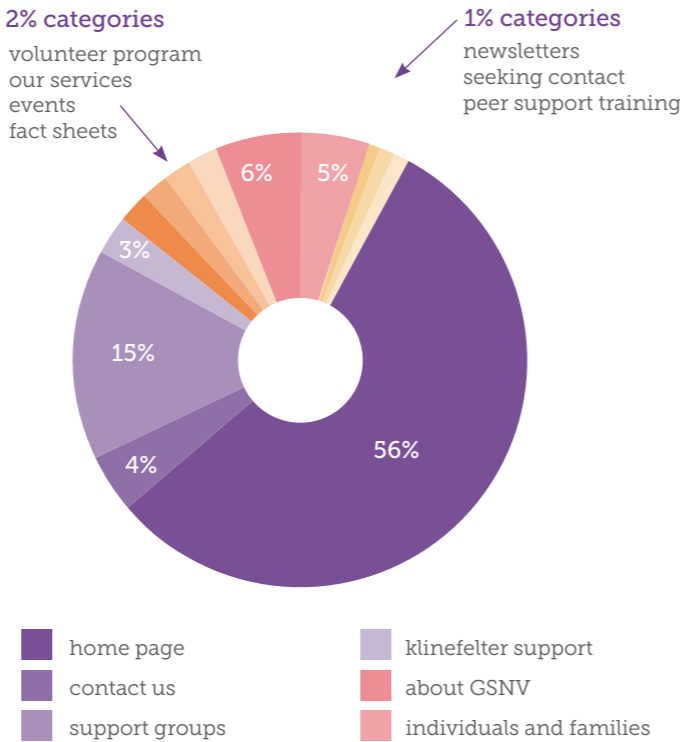


WEBSITE, DATABASE AND SOCIAL MEDIA

Social media remains an important tool in communicating to our members. In 2015 the GSNV will use social media campaigns and blogging to highlight our important events and information. Social media has been identified as an acceptable form of communication both formally and informally and is incorporated into the GSNV communications policy.

The GSNV seeks to follow trending on social media and subscribes to over 30 closed Facebook pages and blogs developed for condition specific support. It is anticipated that social media will require further time and resources in the future and therefore should be considered in future planning and project managements.

WEBSITE PAGE VIEWS



NUMBER OF WEBSITE VIEWS

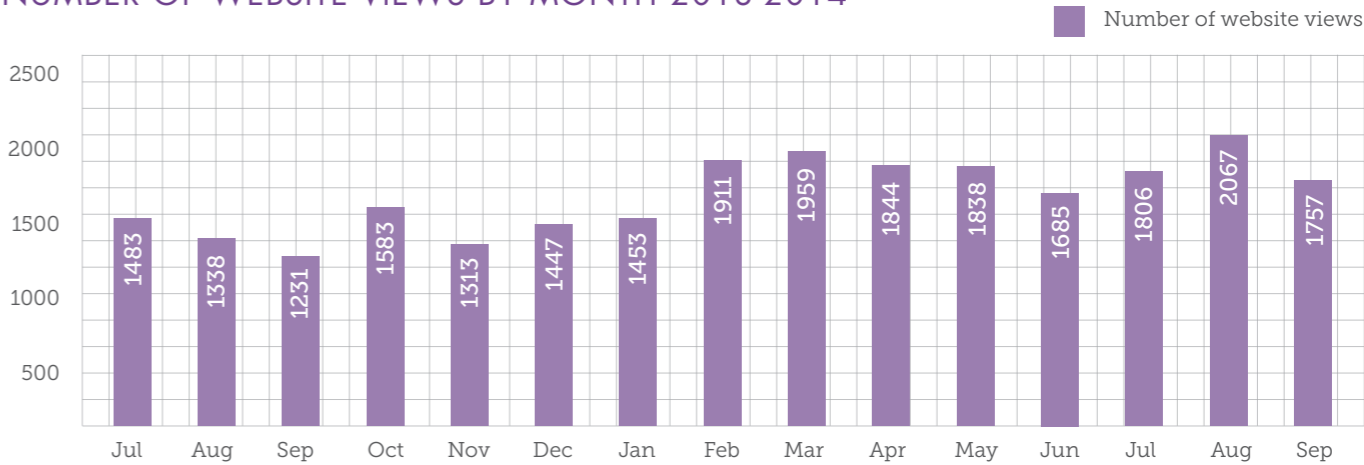


WHO ARE WE REACHING ON TWITTER?

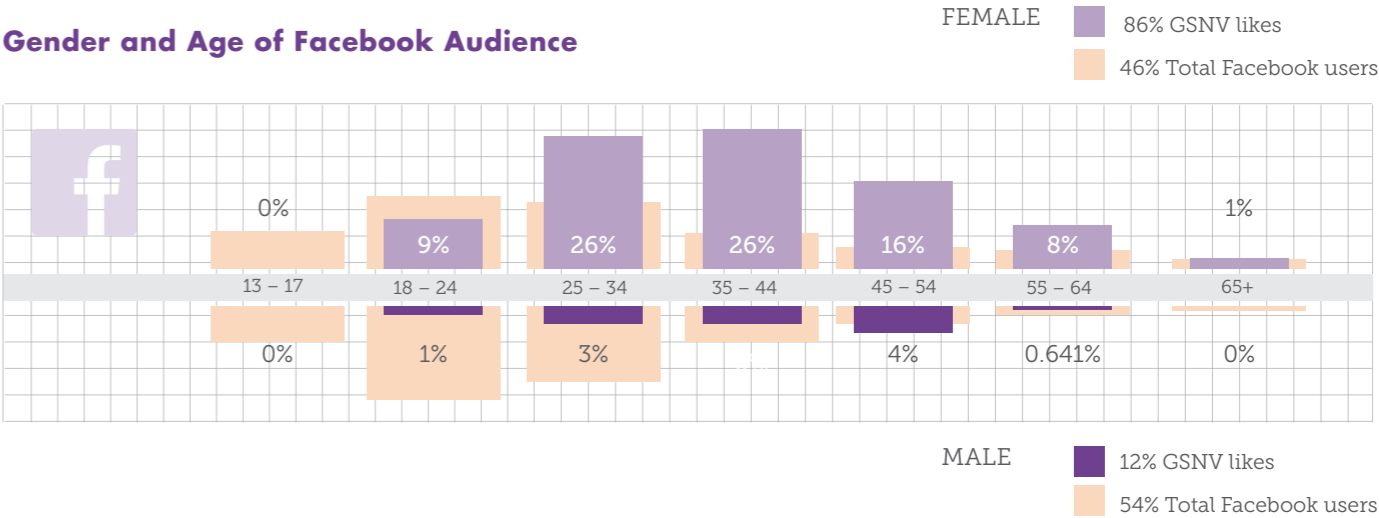


196 TWEETS | 427 FOLLOWING | 84 FOLLOWERS

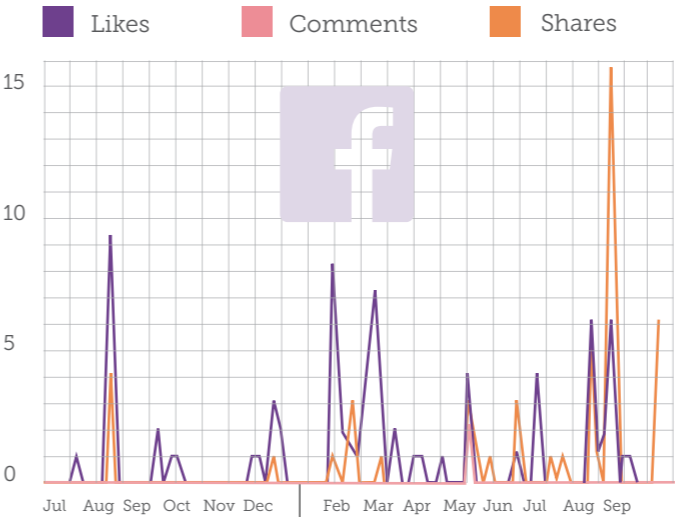
NUMBER OF WEBSITE VIEWS BY MONTH 2013-2014



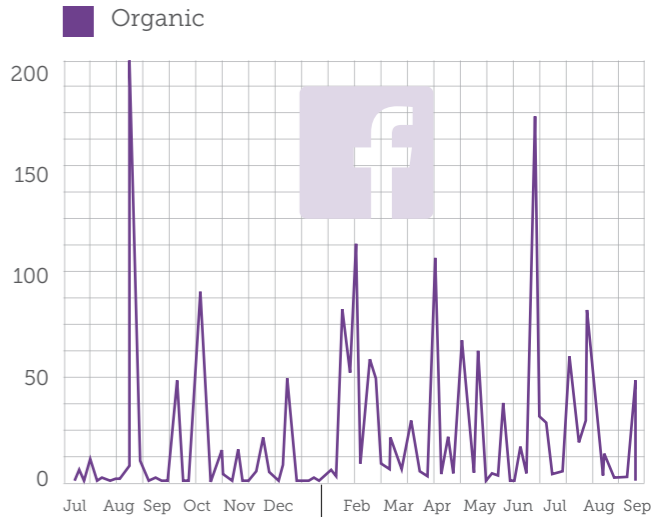
WHO ARE WE REACHING ON FACEBOOK?



Facebook page visits  
July 2013 – October 2013



Post Reach – the number of people our post was served to



# MICROARRAY Factsheet

## Key points

Microarray is a powerful way to look for genetic causes of health and developmental problems. As microarray is new, health professionals are still learning how to interpret the large amount of information generated. Not all copy number variants are associated with problems.

**References:**  
Testing for missing or extra segments of DNA Fact Sheet, The Centre for Genetics Education, NSW Health. [www.genetics.edu.au](http://www.genetics.edu.au)

### 1. What is a microarray?

Humans are made up of billions of cells. Each cell contains our genetic material, or DNA. The complete set of our genetic material is called our genome. A microarray is a test that looks for extra or missing segments of DNA in a person's genome.

### 2. How does it work?

Our genome consists of thousands of genes. We should have two copies of every gene in each cell. A microarray helps determine how many copies of each gene are present in our DNA. More than two, or less than two, copies of genes can cause problems in the functioning and development of an individual. This is called copy number variation, and this is what a microarray tests for.

### 3. Why are microarrays so popular?

A microarray test is ordered if a doctor suspects that extra or missing pieces of the genome may be the cause of health or developmental problems. Microarrays are a very cost-effective, fast way of looking at almost the whole genome at the same time. Up until a few years ago looking at chromosomes under a microscope was the only way to analyse the whole genome at low cost. However, chromosome analysis can only detect very large changes in the genome. Microarrays can detect much more subtle copy number variants than chromosome analysis and so are more effective at finding the cause of developmental or other health problems.

### 4. Does a microarray test for everything?

No. A microarray only tests for variants in the DNA copy number. There are many other conditions in which there is a variant in a gene without any variation in the number of copies of that gene. **For example, microarray will not diagnose Fragile X.** In other situations the copy number variant may be too small to be detected by the microarray.

### 5. What results can you expect from a microarray?

You can expect two possible results from a microarray test:

- a) No copy number variant is found. This is the most common result, and it means that the condition in question remains unexplained by this test. It does not mean that the cause of the condition is not genetic.

**OR**

- b) A copy number variant is found.

### 6. What does it mean when a copy number variation is found?

Not all copy number variants cause problems. Some copy number changes are innocent genetic changes within a person or family. Health professionals determine how likely the copy number variant is to cause problems by checking medical reports of other individuals with similar copy number variants and by examining the genes within the copy number change. There are several possible outcomes of this analysis:

- a) The variant is known to cause the condition. The microarray test has found the cause of the individual's health problems.
- b) The variant is of uncertain significance. This means we don't yet fully understand how the variant impacts on health and development. Although these variants may occur more frequently in individuals with health or developmental concerns, they also may be found in individuals without these concerns.
- c) The variant is of unknown significance. When this occurs, further investigations are needed in order to try and clarify the result. Variants of unknown significance have generally not been reported previously.
- d) The variant found involves genetic material that is unrelated to the condition being investigated, but is potentially associated with other future health concerns. This occurs infrequently but results in families receiving unexpected information. An incidental finding may require further follow up and counselling.

For variants of unknown or uncertain significance, testing of parents is useful. If one parent has the variant and has similar features as the child, then the variant is likely to be responsible. If one parent has the variant and doesn't have similar features as the child, then the variant is unlikely to be responsible (unless there is evidence in the medical literature to suggest otherwise). Sometimes neither parent will have the variant. This is called a de novo, or a new variant. A de novo finding increases the probability that the variant is the cause of the condition in the child, but this variant may still have uncertain or unknown significance.

This fact sheet has been produced by:



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**Web** [www.vcgs.org.au](http://www.vcgs.org.au)

# SNAPSHOT



## HUMAN GENETICS SOCIETY OF AUSTRALASIA (HGSA) CONFERENCE

This year the GSNV Inc. funded Keri Pereira, the GSNV's new Genetic Support Coordinator, and Rachel Pope-Couston (Treasurer), to attend the 38th annual HGSA conference which was held in Adelaide, SA. The theme for this year's conference was 'The Changing Face of Genetic Medicine'. This theme represented the focus of the 4-day conference which was on the rapidly changing way that genetic testing is being conducted in the laboratory, as well as the evolution in how genetic healthcare is delivered by all genetic healthcare professionals.

The HGSA represents many special interest groups (SIGs) including the Australasian Society of Genetic Counsellors (ASGC). Keri and Rachel are both members of ASGC and attended conference sessions organised by this SIG as well as the general conference presentations. Rachel also presented her research to the ASGC SIG as part of the 3 Minute Thesis competition.

Rachel's research is centred on exploring how clinical geneticists communicate with families when the genetic result is of 'uncertain clinical significance', particularly looking at what they write in the consultation summary letters clinician's send to families following their appointment. First place in the competition was awarded to another of our committee members, Hanna Leslie, who presented her research into how genetic specialists could be educating other health professionals about genetic testing and care for individuals impacted by a genetic condition. So congratulations Hanna!

Much of the focus of the conference was on the way that supporting individuals and families impacted by a genetic condition is changing in response to improving genetic technology and the increased availability of testing.

Both Keri and Rachel gained valuable knowledge and experience through attending the HGSA conference and that has flowed on to benefit their respective roles at the GSNV Inc. However, they also felt there were some disappointing aspects of the conference program this year. In the past, there had been a more integrated feel across the SIGs, which is beneficial because the HGSA conference is a unique opportunity for the various genetic specialist groups to interact, from laboratory to clinical staff as well as research specialists and international experts in disease treatment.

It was also disappointing that there was no representation for individuals impacted by genetic conditions, especially considering the theme of the conference. It seemed to be an oversight not to include the perspective of organisations such as the GSNV Inc. as in the past there have been open forums and panel debate sessions which allow these viewpoints to be presented. Our group leader has participated in these in the past and has spoken eloquently and passionately and we were saddened that there were no similar opportunities for the GSNV Inc. this year.

All conference attendees are asked to provide feedback to conference organisers and Rachel took that opportunity to highlight these few disappointing aspects (as well as all the positives). We hope that Rachel was not the only one to provide that feedback and that next years' program will include more opportunities for the GSNV Inc. and other groups to advocate on behalf of our members.

Keri and Rachel would like to sincerely thank the GSNV Inc. for the opportunity to attend the 2014 HGSA Conference.

**Keri Pereira & Rachel Pope-Couston**

## SNAPSHOT



### RARE DISEASE DAY IN VICTORIA – THE GSNV LEADS THE CAMPAIGN

Rare Diseases Day was held on 28 February. With over 50 attendees comprising a mix of campus staff, support group leaders and individuals living with rare conditions, it was a terrific opportunity to bring together all those in the rare disease community. Rare Disease Day raises the awareness of rare diseases and importantly focuses on the profound impact on people affected personally, their families and carers.

Two personal stories presented by Nicole Millis (MPS Society of Australia) and Mandy Jacobs (NPC Foundation of Australia) provided some insight on the experience during diagnosis and life thereafter for families such as theirs, in the rare disease community. Further presentations from Dr Sue White and Flora Pearce explored the clinical perspective of 'a heightened sense of responsibility regarding the genetic aspects of a diagnosis and feelings of isolation.'

The take home message from the presentations was that parental reaction and coping mechanisms around a new diagnosis vary greatly, but aspects such as a person's culture, lived experience, beliefs and knowledge along with their resilience and ability to adapt influences their journey over all.

The GSNV was pleased to see attendees 'raise their hands' in support of rare disease patients around the world. In a symbolic gesture, attendees had an opportunity to add their hand to a patchwork of hands, in colour creating a collage of joining hands. This very simple exercise indicated that both professionals and individuals impacted by rare diseases are driven to join together in finding effective cures and treatment, therefore embracing the international Rare Disease Day goal of 'joining together for better care.'

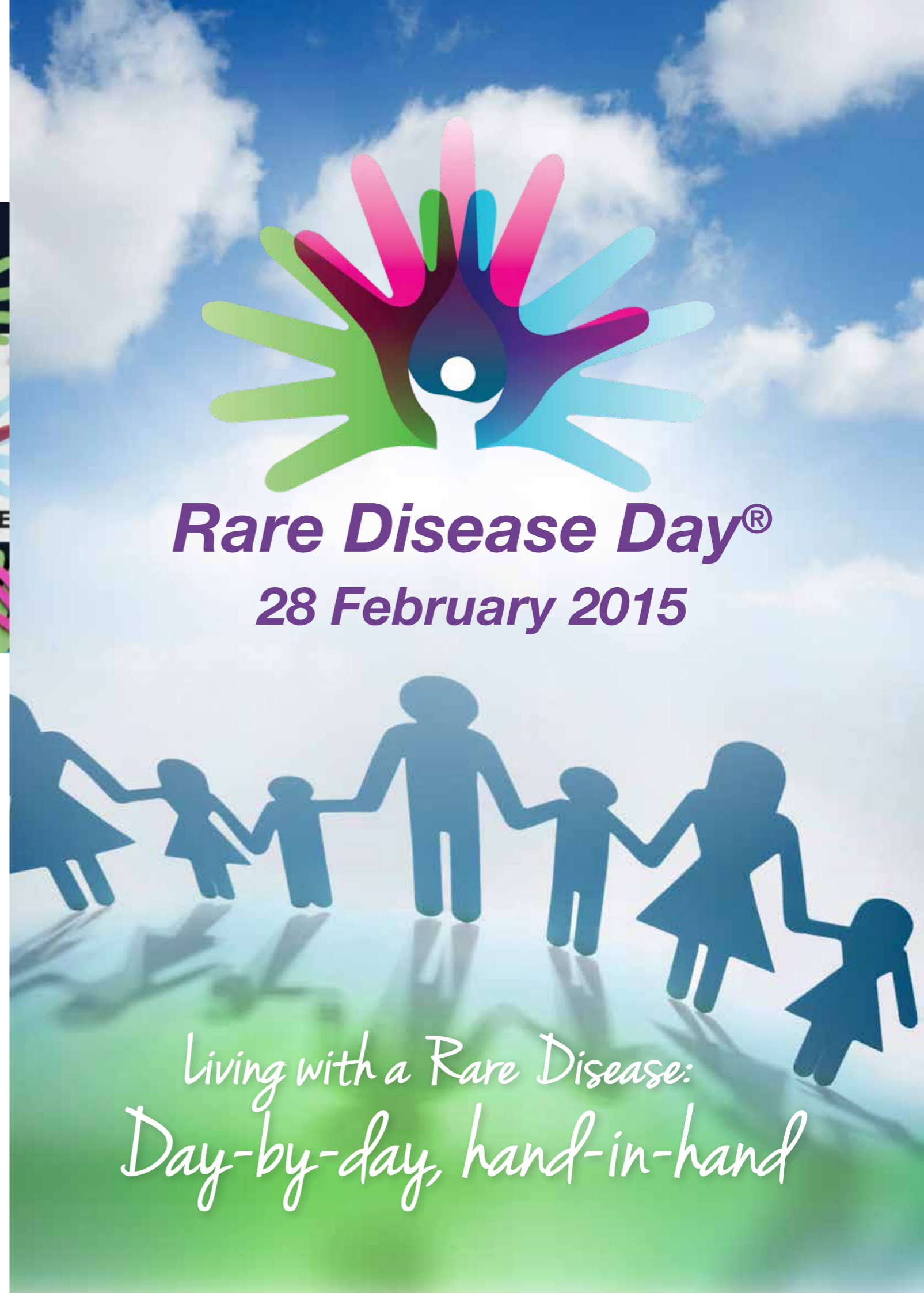
This year's celebrations were a true partnership, with great support from MCRI. The GSNV thanks all who supported Rare Disease Day.

**Rare Disease Day®**  
28 February 2015

*Living with a Rare Disease:  
Day-by-day, hand-in-hand*



*empowering \* connecting \* supporting*



## SNAPSHOT

### NEUROMUSCULAR SUPPORT GROUP FIRST MEETING

**By Hannah Pennington, 25 years old, Melbourne  
suffers from (currently un-diagnosable) form of Spinal Muscular Atrophy**

The first neuromuscular support group informal meeting was held on Wednesday 3 September at the Royal Children's Hospital in Parkville.

Louisa Di Petro the Group Leader of the Genetic Support Network of Victoria began the discussion by welcoming the attendees and introducing the first speaker, Clinical Geneticist Associate Professor Paul James who ran through some recent testing and treatment updates and also a general scientific explanation on neuromuscular conditions. Paul also touched on an exciting update of a new nerve deterioration treatment option which has recently been cleared for human participation – this is yet to be introduced in to Australia but it is an absolute possibility.

This was followed by my talk – which was a speech describing my personal experience as an adult living with a neuromuscular condition. I outlined the stages of my diagnosis over the past 4 years to now – including my first symptoms to a few misdiagnosis experiences, how it has affected and still affects my day to day life and relationships.

Following my (emotional) speech was a presentation given by Adrienne Sexton, a genetic counsellor (and my personal genetic counsellor) at the Royal Melbourne Hospital. Adrienne's presentation included questions and experiences of young adults in general who had visited the genetics service and also some young adults' experiences taken from a few international websites who suffer from a neuromuscular condition. The presentation touched on how the condition impacted work, home, socially and also in relationships and concluded by reminding us that while physically we were perhaps restricted, mentally and emotionally we can still do a whole lot in our lives.

The meeting was finished by thanks and comments from Louisa and me and also a big thank-you to Adrienne, who

without her support the creation of the group would not have been possible.

I look forward to future meetings and anticipate possible participation and involvement from other young adults living with neuromuscular conditions. Personally, I believe that a large part of dealing with this particular type of condition is that because there are currently no medical physical treatments available – sharing your experiences and being able to relate to the similar experiences of other young adults is perhaps a method of mental and emotional treatment in itself.

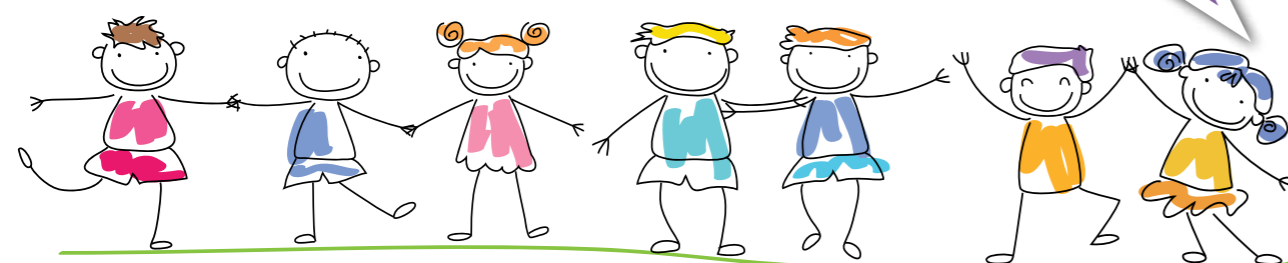
To see the video recording of the session go to [neuromuscularsupport.weebly.com/meetings.html](http://neuromuscularsupport.weebly.com/meetings.html)

I would like to thank those associated in organising the informative evening I attended on 3.9.14 for neuromuscular social support group for young adults.

I am very proud to have been a part of such a structured and informative session. I am grateful for Professor Paul James' talk and found it very informative and helpful.

I am very proud of my daughter's speech on her insight with coping with SMA. Hannah delivered herself well and it was comforting to be with Hannah and her friends and the staff at such a heart wrenching event. I do hope that other people will try and attend in the future. It really does help talking to others. Well done!!

Ann Pennington



*empowering \* connecting \* supporting*



## THANK YOU

THE GSNV WISHES TO THANK THE MANY PEOPLE, INCLUDING MEMBERS AND VOLUNTEERS WHO HELP DELIVER OUR WORK IN VICTORIA AND THOSE WHO HAVE GIVEN A GIFT OF THEIR TIME, EXPERTISE AND KNOWLEDGE.

We thank those who have donated and paid a membership due in order that we can provide small grants and give back to support groups. We are equally grateful for the ongoing support of the Department of Health Victoria who continue to provide recurring funding for our important work. We thank in particular Dr Paul Fennessy and Margaret Howard (DoH) for an ongoing supportive relationship with the GSNV and for their wisdom year in, year out.

We acknowledge the in kind support and advice from many VCGS and MCRI staff without whom, we would not be able to what we do. We thank Assoc. Prof David Amor for support of the GSNV staff as line manager and providing that all important sounding board to the Group Leader.

# FINANCIAL REPORT

ON BEHALF OF THE EXECUTIVE AND COMMITTEE OF THE GSNV INC. WE ARE PLEASED TO REPORT ON THE FINANCIAL DETAILS FOR THE FINANCIAL YEAR 2013-2014.

We have continued support from the Department of Health (DoH) in the form of an ongoing grant managed by VCGS and through this grant and other small avenues of income, the GSNV Inc. remains a viable and solvent organisation.

As part of VCGS, the accounts for GSNV are audited by KPMG who are our External Auditors. They report that for the Financial Year 2013/2014 the GSNV Inc. recorded a surplus of \$8,028.39. This surplus combined with the DoH funding means that the GSNV Inc. carries forward a total of \$167,171.61 to the current 2014/2014 Financial Year.

However, the GSNV Inc. is continuing to consume the existing surplus with increased salary and infrastructure costs and so a major focus for the committee has been to find new ways to secure our financial future.

## FINANCIAL SUB-COMMITTEE

As part of that focus, a Financial Sub-Committee (FSC) has been formed which includes myself as Treasurer, our President, Kay, along with several other committee members; Hanna, Shona, Christine, Doreen and Katarina.

We have had a very busy year finding our feet as a newly formed sub-committee but we have also been very productive. Our main goals were to brainstorm and implement ways of recovering daily running costs and securing grants for special programs such as regional outreach and conducting a member survey. The FSC has clear terms of reference as outlined here below:

### Responsibilities

- Be involved in the development of the GSNV's annual budget with the help of the president and treasurer
- Ensure that the annual budget supports the GSNV's strategic and business plans
- Present the recommended budget to the GSNV Committee for approval and adoption
- Regularly monitors the budget against revenue and expenditure and report significant variances to Committee
- Ensure there is an adequate internal control system (as per MCRI financial systems and GSNV processes) to promote operational efficiency and to minimise financial risk
- Ensure that the GSNV has an appropriate policy in relation to any assets/investments and that this policy is approved by Committee annually
- Monitor the status of any assets/investments (this might be monies on a term deposit, etc)
- Make recommendations to Committee on fund-raising activities and liaise with the GSNV regarding these activities
- Review the annual audited financial statement and the auditor's report and assist with follow-up where appropriate
- Assist the treasurer in ensuring that the GSNV operates within the annual budget and that all liabilities and financial commitments are brought to account in the relevant year
- Full and transparent reporting as per GSNV AGM requirements and in relation to responsibilities under the amended Associations Act

## Membership

- At least four people – including but not limited to the Group Leader, the Treasurer, a member of the general Committee and any Committee member or interested person who has a interest in supporting the GSNV financial position

## Meetings

- The finance sub-committee will meet at least once a month prior to every GSNV Committee meetings to consider the financial operation of the GSNV and to ensure that appropriate reports and recommendations are prepared for Committee
- If the convenor of the finance sub-committee is absent or unable to preside at the meeting, a member other than the leader elected by the members present at the meeting must preside

## GRANT SEEKING AND APPLICATION

One of the major achievements of the FSC this year was to prepare a 'Grants Opportunities' document which is a working list of relevant grants that is reviewed at each FSC meeting. Grants that are suitable for the GSNV Inc. are then highlighted for discussion with the GMC to determine capacity and feasibility of preparing an application.

The main grant application prepared by the FSC for 2014 (with many thanks to Hanna Leslie who was the primary author) was for the Lord Mayor's Charitable Trust Foundation (LMCTF) Exploration Grant, and we are eagerly awaiting the outcome, with notifications due in November. This grant would provide the GSNV with \$20,000 to develop and complete a survey of all GSNV members which would provide the GSNV with important information, particularly about what services could be added or improved.

The FSC also investigated the feasibility of applying for a Helen McPherson Smith Trust, which is now prioritised for 2015. The Grants Opportunities document also notes online community resources which provide help for grant-makers through all stages of preparing a grant application.

Developing the Grants Opportunities document is part of the FSC's efforts to harness the learnings from its first year of operation, in order to start building its processes and knowledge base, with the intention of gradually streamlining FSC operations. In its report on applying for the LMCTF grant, the FSC recognised the value of existing expertise within the FSC and GMC which can be drawn on in preparing applications, and the value of a collaborative effort, but also recognised members' busy schedules and existing time commitment to the GSNV.

The FSC and GMC have been exploring ways to ensure appropriate expertise and time can be invested in preparing grants, given the potential rewards of preparing a comprehensive application.

Our goal is that eventually all committee members and general members of GSNV Inc. will have the opportunity to be involved in grant applications on behalf of the GSNV Inc. as well as their own support groups. This would involve the GSNV Inc. supporting members in up-skilling through education and access to people experienced in preparing grants.

MEMBERSHIP AND FEE STRUCTURE REVIEW

The FSC also took on the task of reviewing the current GSNV Inc. membership and fee structure. We did this by investigating the membership structures and fee schedules of other organisations that provided services comparable to the GSNV Inc.

We would like to reassure members that there will be no changes in service delivery or fees for individual members however, the FSC have identified some opportunities for securing more professional memberships as well as the possibility of developing a corporate sponsorship/partnership model.

The FSC recognises that any changes to the membership structure and fee schedule will need to be made transparently and with full accountability in place for any sponsorships or partnerships the GSNV Inc. may form. We will be seeking endorsement from members at this AGM in order to proceed with our investigations.

CHANGE TO FINANCIAL REPORTING DATES

One of the important changes implemented by the FSC this year was to move our financial reporting from Financial Year to Calendar Year meaning that our end of financial year date will change from the 31 June to the 31 December each year.

The GSNV Inc. utilises the MCRI financial tracking and reporting computer system as well as the MCRI financial management team which all operates to the Calendar Year so moving the GSNV to this format enables greater ease of access to reports as well as making the reports easier to understand for non-finance specialists.

The change to financial reporting dates was put to our membership as a special resolution in a special meeting held on the 18 September 2014 and has now been approved by members and Consumer Affairs Victoria. This means that the next AGM following this AGM will be held by the end of May 2016 and will encompass financial reporting from 1 July 2014 to 31 December 2015.

OTHER ACHIEVEMENTS FOR THE FSC

The FSC contributed to the GSNV Inc. risk register by developing a financial risk register. This was done by identifying potential risks to our financial future as well as the steps we could take to mitigate those risks. This includes the risk of depleting our surplus and one of the ways we can mitigate that risk is by applying for grants to supplement the DoH funding. The FSC is pleased to report that we have implemented all possible steps to mitigate financial risks for the GSNV Inc.

As a new sub-committee, one of the major achievements of the FSC this financial year has been to develop a set of operating guidelines for all FSC activities such as conducting our monthly meetings and grant applications (as detailed above). We focussed this year on setting in place the architecture that will allow a smooth transition for new FSC members as well as ensuring the FSC will be even more efficient and effective in 2015.

Thank you

I would like to personally thank all FSC members for their time and the dedication they have shown towards making this sub-committee a dynamic and successful addition to the GSNV Inc.

Rachel Pope-Couston

STATEMENT OF INCOME & EXPENSES

STATEMENT OF INCOME & EXPENSES FOR THE MONTH ENDED 30 JUNE 2014  
70680 GENETIC SUPPORT NETWORK (V)

	Actual 2014		Budget 2014	
	For this Period	Year to Date	Year to Date	Year to 31/12/2014
BROUGHT FORWARD BALANCE @ 1/6/2014		159,089.22		127,345.18
INCOME				
DHS GRANT	12,583.34	75,500.04	75,499.98	151,000.00
NHMRC CONSULTING	0.00	0.00	0.00	0.00
LORD MAYOR'S TRUST	0.00	0.00	0.00	0.00
MEMBERSHIP FEES	0.00	0.00	0.00	0.00
DONATIONS	30.00	739.00	0.00	0.00
CONFERENCE FEES	0.00	0.00	0.00	0.00
SUNDRY INCOME	40.00	595.00	1,000.02	2,000.00
Total Income	12,653.34	76,834.04	76,500.00	153,000.00
EXPENDITURE				
SALARIES & RELATED COSTS	5,448.71	54,036.10	75,499.97	151,000.00
COMPUTER HARDWARE	0.00	0.00	499.98	1,000.00
COMPUTER SOFTWARE & EXPENSES	27.27	27.27	499.98	1,000.00
ADVERTISING	416.00	416.00	0.00	0.00
FREIGHT & CARTAGE	0.00	0.00	0.00	0.00
POSTAL SERVICES	18.14	323.96	400.02	800.00
PRINTING, STATIONERY & PHOTOCOPYING	1,777.34	4,955.31	5,250.00	10,500.00
BOOKS & SUBSCRIPTIONS	0.00	85.46	649.98	1,300.00
TELEPHONE CALLS	40.06	259.67	289.98	580.00
SMALL GRANTS	0.00	0.00	0.00	0.00
SPECIAL FUNCTIONS – OTHER	99.00	545.09	1,500.00	3,000.00
STAFF TRAINING & CONFERENCES	136.36	620.86	1,000.02	2,000.00
TRAVEL	787.44	6,280.28	2,749.98	5,500.00
CORP SERVICES – IT/HR/FIN	0.00	0.00	7,500.00	15,000.00
OTHER ADMINISTRATIVE COSTS	97.38	1,201.65	6,100.02	12,200.00
Total Expenditure	8,847.70	68,751.65	101,939.93	203,880.00
OPERATING SURPLUS/(DEFICIT) CARRIED FORWARD @ 30/06/2014		167,171.61		76,465.18

## AUDIT STATEMENT



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Murdoch Childrens Research Institute  
Flemington Road, Parkville VIC 3052 Australia  
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W vcgs.org.au ABN 51 007 032 760

### Audit Statement

I confirm that the funds belonging to the Genetic Support Network of Victoria are held by the Victorian Clinical Genetics Service (VCGS). The VCGS also currently provides space & infrastructure for carrying out of the services of the GSNV.

The accounts of the VCGS are audited annually by KPMG who are our External Auditors. The accounts for 2013 have been audited by KPMG in accordance with this practice. In the Financial Year 2013/14 GSNV recorded a surplus of \$8,082.39. However, with the brought forward funding at the beginning of the year it was able to carry forward \$167,171.61 to the current 2014/15 Financial year.

Let me take this opportunity to wish you & your team another successful year ahead.

Best Wishes,

**Viren Abeyasinghe** MBA, FCPA, FCMA  
Chief Financial Officer  
Murdoch Childrens Research Institute/  
Victorian Clinical Genetics Service.



**genetic  
support  
network  
of victoria**

**genetic support** network of victoria

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