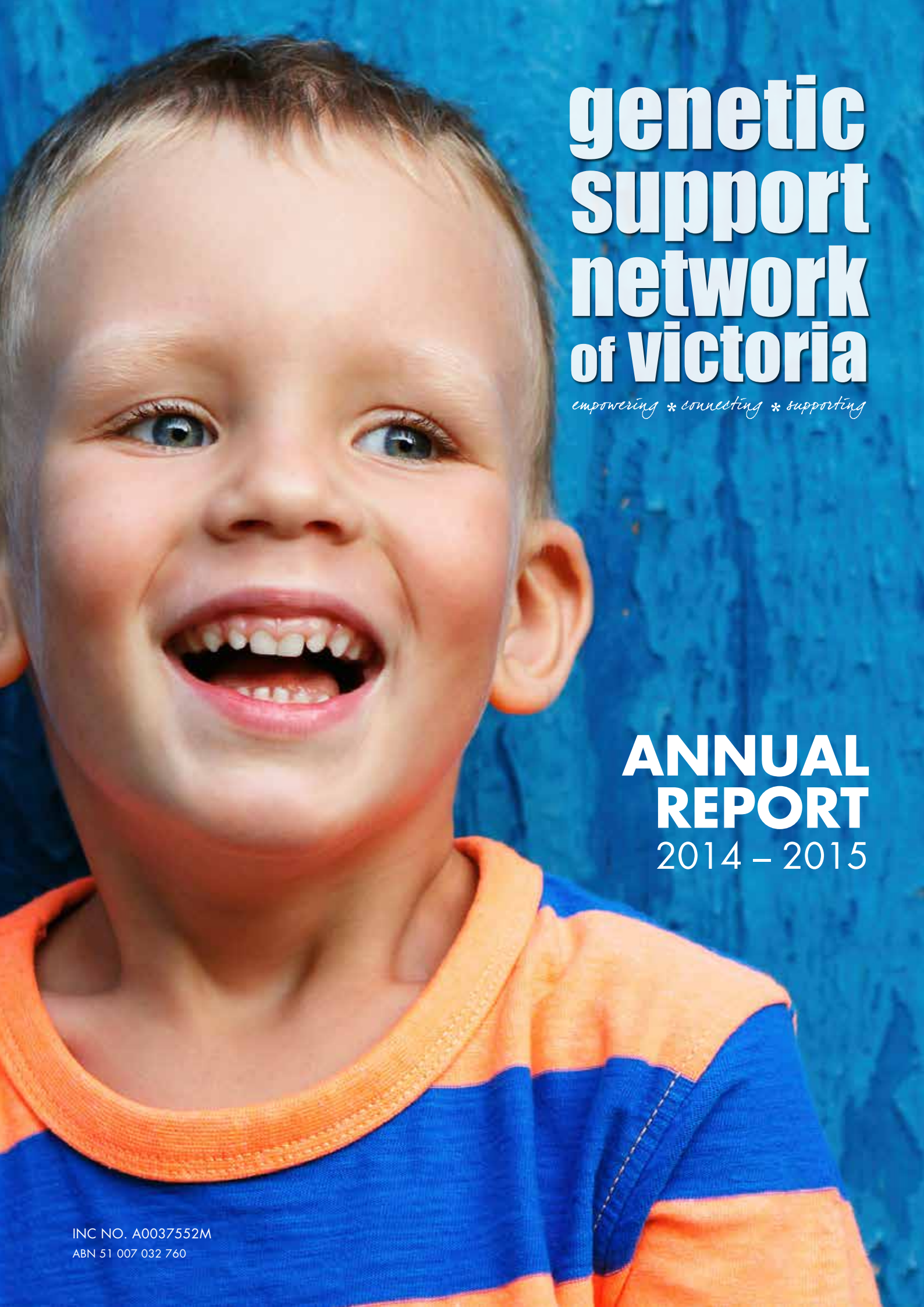


genetic support network of Victoria

*empowering * connecting * supporting*

ANNUAL REPORT
2014 – 2015

INC NO. A0037552M
ABN 51 007 032 760

genetic support network of victoria

genetic support network of victoria

Murdoch Childrens Research Institute
Flemington Road, Parkville VIC 3052 Australia
P (03) 8341 6315 **F** (03) 8341 6399
E info@gsnv.org.au **W** www.gsnv.org.au

CONTENTS

*empowering * connecting * supporting*

WELCOME TO THE GENETIC SUPPORT NETWORK OF VICTORIA INC. (GSNV INC.) ANNUAL REPORT FOR THE PERIOD 1 JULY 2014 TO THE FINANCIAL YEAR ENDED 31 DECEMBER 2015.

The GSNV made a strategic decision to move to a 'calendar' financial year in 2014 and therefore did not conduct an Annual General Meeting (AGM) in 2015. This is the first meeting conducted since the change to a January to December financial calendar.

As we reflect on the year in review 2014/2015 we will endeavour to highlight some of our major achievements, challenges and outcomes; including an overview of governance and financials.

EMPOWERING THE GENETICS COMMUNITY

For almost 17 years the GSNV has been actively engaging with the Victorian community and assisting those impacted by a genetic condition. From those early years of helping with the establishment of face to face support groups, we have built our capacity and developed a reputation as a strong network and a representative of the consumer voice.

Our work continues today helping the genetics community and engaging in psychosocial research within a highly sophisticated and expanding genomics environment. The future of the GSNV is tied strongly to the development of a leading genomics hub in Victoria and we seek to position ourselves to maximise the potential gained from this evolving environment.

This annual report for the period 1 July 2014 to 31 December 2015 is a reflection of the power of the genetics community through the commitment and care that our people and supporters provide every day in Victoria and in collaboration with other states and international leaders.

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 operates within a changing and challenging environment. Victoria and particularly the Parkville precinct is evolving as a world leader in genomics medicine and research and in 2014-15, the GSNV has strived to embrace the opportunities attributable to this. We have considered how we can strategically position ourselves and how we can continue to meet the information needs of the genetics community – a community that is increasingly knowledgeable about genetics, with greater expectations on what can be delivered in genetic health because of new technologies and increases in testing and diagnostic capability. The genetics community is seeking answers, particularly for those who remain undiagnosed and are looking to benefit from the use of genome technology.

The GSNV now operates within a physical landscape that includes major research institutes and clinical facilities in the Parkville/Melbourne precinct. The genomics and genetic expertise in Melbourne is leading toward a geographic centre of excellence and for the GSNV this represents greater validity, purpose and relevance for our services.

The concentration of clinical expertise further increases the focus on GSNV as having a vital role in 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 recognise our role is clearly to represent the people and to ensure continuous improvement in all areas of genetic services including service, advocacy, care and support.

*empowering * connecting * supporting*

THE YEAR IN REVIEW

IN THE 2014-15 FINANCIAL YEAR, OUR VISION AND MISSION HAVE LED TO OUTSTANDING ACHIEVEMENTS. HERE'S A SNAPSHOT...

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

THE GSNV SUPPORTS VULNERABLE PEOPLE IN OUR COMMUNITY

Since its original set up in the GSNV has been committed to improving the lived experience of Victorians impacted by a genetic and/or rare condition, and has focused on assisting with the processes of genetic testing, diagnosis, support and advocacy.

We are committed to reducing the feelings of isolation and vulnerability, which can result from living with or caring for someone with a genetic/rare condition and this underpins our work.

We aim to 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; help people in crisis; empower those motivated and working hard to represent a condition specific community; advocate and represent on behalf of our members; 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). We have representation on the HGSA Education, Ethics and Social Issues Committee (EESIC).

We are members of the Australasian Society of Genetic Counsellors (ASGC) Special Interest Group (SIG), the HGSA Victorian/Tasmania Branch and have in 2015/2016 sat on the HGSA Victorian/Tasmania Branch Local Organising Committee for the 2016 Tasmanian Annual Scientific Meeting (ASM).

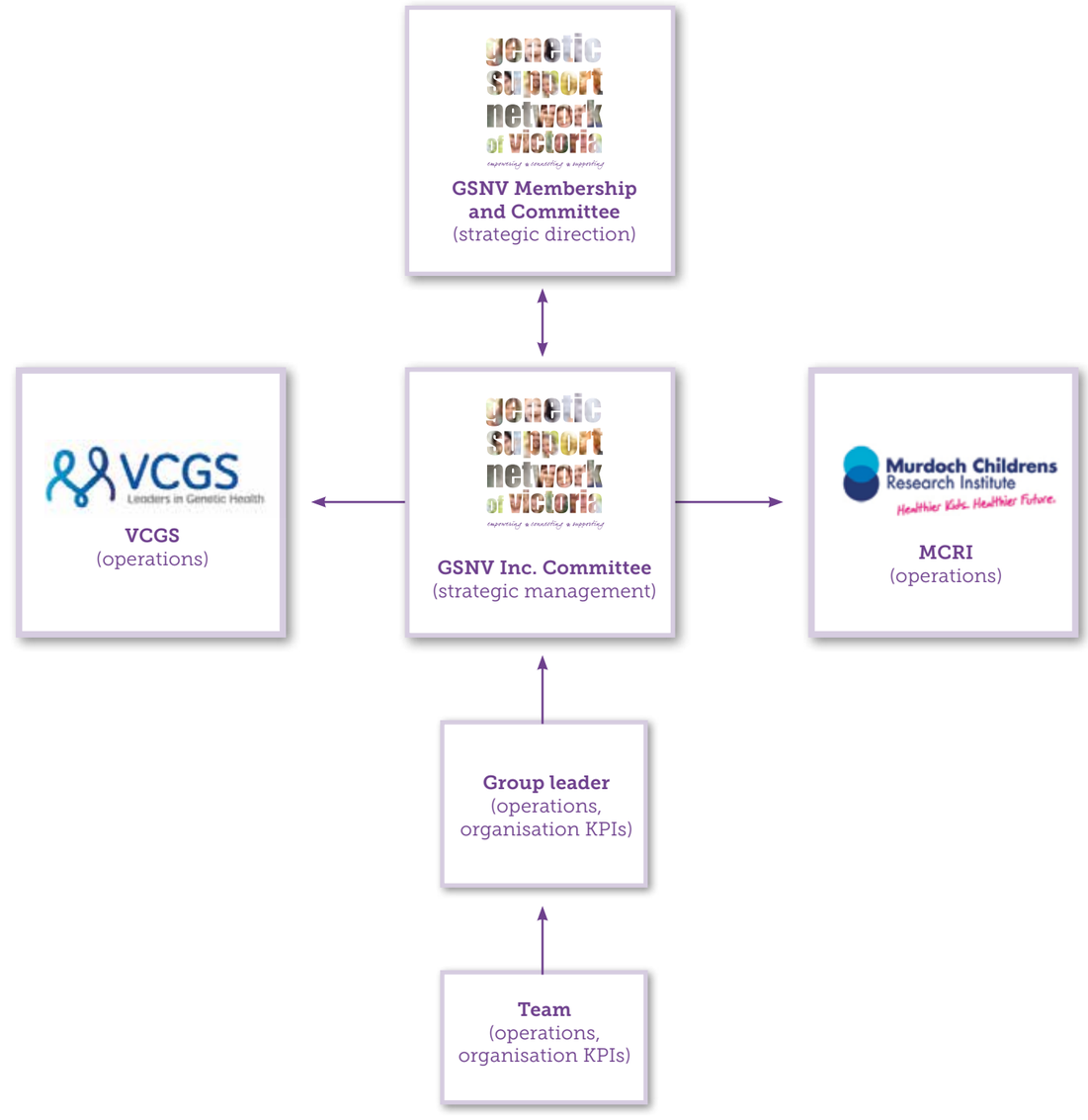
The GSNV also proudly sits on the Melbourne Genomics Health Alliance (MGHA) Consumer Advisory Group (CAG).

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

1. ASHG – American Society of Human Genetics
2. CAGC – Canadian Association of Genetic Counsellors
3. CCMG – Canadian College of Medical Geneticists
4. IFHGS – International Federation of Human Genetics Societies
5. ESHG – European Society of Human Genetics
6. NZOrd – New Zealand Organisation for Rare Disorders
7. HGSA – Human Genetics Society of Australasia
8. EuroDis – Rare Diseases Europe
9. Rare Connect
10. Genetic Alliance UK
11. Genetic Alliance US
12. ICHG – International Congress Human Genetics
13. SIGU – Italian Society of Human Genetics
14. GA-SA – Genetic Alliance South Africa
15. Dutch Genetic Alliance
16. International Genetic Alliance

OUR PEOPLE

GSNV ORGANISATIONAL CHART



The Royal Children's Hospital Melbourne

Murdoch Childrens Research Institute
Healthier Kids. Healthier Future.

THE UNIVERSITY OF MELBOURNE

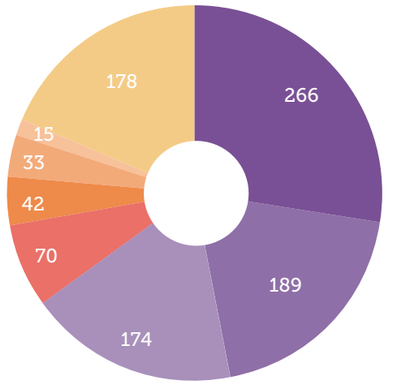
genetic support network of victoria

OUR MEMBERS

The GSNV financial members are our most important stakeholder group. Beyond paid members, we have a network and community that comprises the general, lay, professional/training genetics and rare diseases communities, professional organisations/networks, alliances and biotech/pharmaceutical companies and researchers.

Our strength is our people and the feedback, ideas and contribution to enhance and support our work we receive from them is vital. The GSNV would like to say thank you to all members and we look forward to a continuing positive and engaging relationship.

MEMBERS COMPOSITION 2015



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
- Training Health professionals
- Allied Health care professionals

GOVERNMENT BODIES

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

COMMUNITY

The GSNV currently has 984 members

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

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.

OUR COMMITTEE

PRESIDENT'S REPORT

Reflections – from the President

It has been a busy and active year and a half, and the GSNV team have been hard at it working to support the genetics community and to achieve outcomes that help us meet our strategic and organisational objectives. The team have done a wonderful job, while the Group Leader has been overseas spreading the word on how the GSNV model works, and increasing the profile of Australian innovation in Europe. It has been great to see Louisa have an opportunity to present at the European Society of Human Genetics (ESHG) and European Meeting on the Psychosocial Aspects of Genetics (EMPAG) Glasgow meeting, and as we speak, presenting in Barcelona at the same meetings.

Since our previous AGM we have introduced a new staff member, Victoria Rasmussen, who is coordinating the GSNV research evaluation study. This is a very important project and will help us establish some evidence to guide our future direction and service delivery. We have also said goodbye to a valued staff member, Anna Jarmolowicz, who has moved on to a genetic counselling role with VCGS. Thank you Anna for your wonderful work as part of the GSNV team.

Our events calendar has been very full but I think a particular highlight for me was the Rare Disease Day event for 2016. It was a great day! The team put together a fantastic event and the audience were addressed by prominent medical geneticist, Dr. Kym Boycott from Canada.

Dr. Boycott currently leads the Genome Canada and CIHR funded large-scale project 'Enhanced CARE for RARE Genetic Diseases in Canada', which is focused on improving the clinical care of patients and families by expanding and improving the diagnosis and treatment of rare diseases.

Dr. Boycott moves the international rare disease agenda forward through her role as the Chair of the Diagnostics Committee of the International Rare Disease Research Consortium. We were very privileged to have Dr. Boycott open our event and speak about her work.

I extend a big thank you to the GSNV committee who have worked hard (as volunteers do) and progressed important GSNV strategic and operations goals. Health has challenged some Committee members and we would like to thank them for pushing on and remaining committed to the GSNV. It has been a busy, active, eventful and challenging period but we are committed to achieving even greater things in the months ahead.

It is extraordinary how quickly the genetics environment is changing, and the lead Victoria is now taking in genomics. We have worked hard over the past year to ensure that we continue to have a strong governance environment and great working relationships with all stakeholders to continue to position us to support the GSNV organisation, community, and our overall vision and strategic direction.

Looking forward to reviewing our strategic plan and confirming our direction over the next exciting period of genetic support and genomic medicine in Victoria.

Kay Timmins
GSNV President

The Committee would like to thank the GSNV staff – Keri Pereira, Nancy Amin, Victoria Rasmussen, Emily Allen and Anna Jarmolowicz (former admin) and our Group Leader Louisa Di Pietro for their work and service, and share the following reflections:

Reflections – from the Secretary

It is hard to believe a year and a half has gone by. I feel it has been a year where we have learnt a lot through being challenged to step up and do business differently.

In a sector where science and genetic technology are developing at an exciting rate, the GNSV continues to strive to serve people who are impacted by a genetic or rare condition and those professionals who diagnose, treat, support and counsel them. This means working hard to be across all developments in the sector, liaising with clinicians, stakeholders, members, professionals (including training), researchers and support networks; always reviewing how what we are learning can positively impact.

With a successful Rare Diseases Day celebration, peer support training, support group events and numerous education and training sessions behind us, we see the remainder of 2016 as a time to reflect, consolidate and concentrate on achieving substantial outcomes and strategic goals.

We have been blessed over the past year to have the opportunity for our Group Leader to be immersed in the international genetics community through a placement in Italy. This has greatly increased the GSNV's links to international best practice, policies and procedures as well as access to global experts and their research, particularly in the rare diseases area. The GSNV is a flexible and forward thinking workplace and we have been pleased to develop a working arrangement for the Group Leader that has accommodated a stint abroad.

While having our Group Leader based overseas has challenged the Committee of Management, the GSNV staff have stepped up to support the Committee and continue to deliver great service. We have watched with pleasure as our staff have accepted the challenge of working with Louisa in a different time zone, dealing with slow international internet speeds and disrupted communications at times, and working with greater autonomy and initiative. GSNV staff have been excellent over the last year in dealing with questions and queries forwarded by the committee and accommodating Louisa's physical absence.

Monica Ferrie
Secretary

Reflections – from the Committee

What does GSNV mean to me? It means enabling.

GSNV enabled a group of mums at home with computers, scattered across the nation, to form a national support group that is flourishing.

We are AusDoCC. We advocate, unite and support Australians who are born without the major structure that connects the two hemispheres of the brain, the corpus callosum.

GSNV enables us to meet once a year, face to face to build our organisation.

GSNV enables us to ride the roller coaster that is underpinned by the DNA helix of our genetic destiny.

GSNV is always there for us, which enables us to be there for others. Heartfelt thanks to our chief enabler.

Maree Maxfield
Committee
Secretary, Australian Disorders of the Corpus Callosum (AusDoCC)

WHO WE ARE

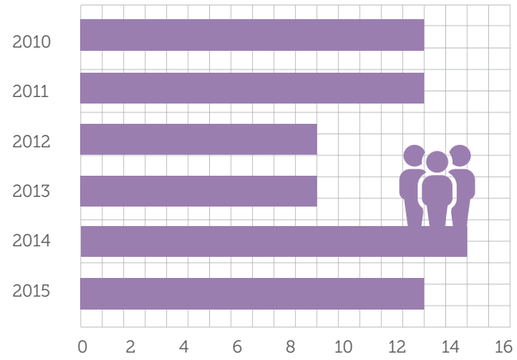
The members of the Executive and Committee who held a position at any time during or since the previous AGM are:

Kay Timmins	President
Shona Malberg	Vice President
Rachel Pope-Couston	Treasurer
Monica Ferrie	Secretary
Abbie Kinniburgh	Committee
Christine Williams	Committee
Doreen Floyd	Committee
Hanna Leslie	Committee
Katarina Radonic	Committee
Maree Maxfield	Committee
Marie Dunn	Committee
Moira Rayner	Committee
Jennifer Ralph James	Committee

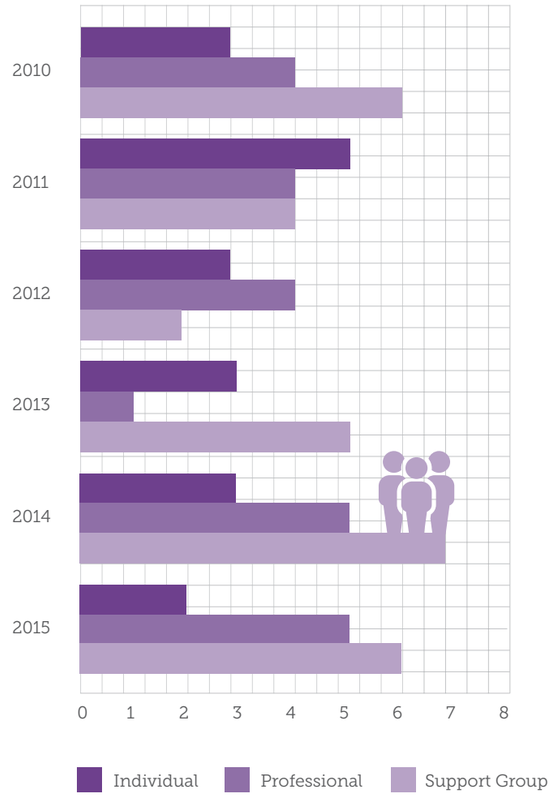
The Committee of the Genetic Support Network of Victoria (GSNV) Inc. operates through an Executive and general committee with delegated authority and terms of reference (TOR) according to the Associations Reform Act 2012 Model Rules. In line with our essential role in the community and our status as 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 organisational 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).

COMMITTEE MEMBER GROWTH



COMMITTEE COMPOSITION 2010-2015



GSNV COMMITTEE TERMS OF REFERENCE (TOR)

PURPOSE

The role of the Genetic Support Network of Victoria (GSNV) is to facilitate a fully supportive, ongoing and enduring relationship with Victorians who are having investigations into a genetic diagnosis, or who have had a genetic diagnosis. At the pre-conception, prenatal, neonatal, paediatric, adolescent and adult stages, patients/clients may have concerns and questions they wish to ask outside of the regular clinical genetics service.

We aim to maintain a network committed to promoting the interests and well-being of the genetic health consumer and through support for aligned support groups at the same time supporting and contributing to professional clinical and research environments of key organisations including Murdoch Children's Research Institute (MCRI) and Victorian Council Genetic Services (VCGS).

THE COMMITTEE OF MANAGEMENT

An effective managing body (the Committee) is central to the success of any organisation, setting its strategic tone, providing leadership to ensure the achievements of objectives and being innovative in adapting to changing environments. The managing body is a forum of critical inquiry into the overall health of the organisation, with a focus on risks, accountability and performance.

KEY FUNCTIONS OF THE COMMITTEE OF MANAGEMENT

Transparency and accountability

The Committee is responsible for ensuring the organisation is transparent and accountable. The Committee reports on its performance and management of its funds through the Annual Report. The Annual Report is both a reporting and a communications tool to funding bodies, members, stakeholders and the public.

Supporting Strategic, Financial and Operational performance

The Committee is also responsible for ensuring the environment is in place to support the delivery of strategic, financial and operational performance objectives for the GSNV. It cannot do this without information about how the organisation is performing. Good quality, timely information on how the organisation is performing enables the Committee to base its decisions on firm evidence.

KEY DOCUMENTS

The GSNV is an incorporated association operating under the *Associations Incorporation Reform Act* 2012 and the associated **Model Rules** established by Consumer Affairs Victoria.

The work of the GSNV is guided by the development of a **Strategic Plan** every three years, which is developed in consultation with GSNV senior staff, VCGS senior staff, key stakeholders and the Committee of Management.

The strategic plan is reviewed annually and the review provides the process that establishes the annual **Business Action Plan** (an operation plan) for the GSNV. This plan forms part of the operational report to the Committee of Management.

The Committee operates with a **Committee Planner**, which provides an integrated schedule of topics and reporting expectations for each year and is aligned to the Business Action Plan.

The planner is intended to provide an outline of key topics, which the Management Committee wishes to consider in depth. It is a framework for **Meeting Agendas** that address core Management Committee functions throughout the year. **Meeting Minutes** are kept and distributed for sign off for every formal meeting.

The planner is a guide to both the Management Committee as well as to the GSNV's senior staff, to enable effective and integrated governance and also to ensure that the Committee's key functions are attended to at appropriate points over the year.

Each meeting agenda is structured around a theme connected to key Committee functions. The Planner provides an outline of the actions and decisions for the Committee to fulfil its responsibilities. It sets out suggested discussion points and the reporting and advice the Committee can expect to receive from the staff to facilitate effective inquiry, critical thinking and balanced decision making.

All Committee members are provided with an **Annual Induction Kit**, which outlines roles and responsibilities and includes the annual undertaking of engagement to sign, declare and return to the GSNV.

New Committee members are provided with the **Terms of Reference** and Annual Induction Kit when joining the Committee of Management.

GSNV has a number of policies and procedures and it is expected that Committee members avail themselves of these policies: These include:

- Travel Reimbursement Policy
- Reimbursement Policy and Procedures
- Communications Policy
- Advocacy Strategy
- Target Board Strategic
- Business Continuity Plan
- Risk Management Plan
- HR policies – eg. bullying, harassment

MEETINGS

The Management Committee meets formally each month.

Meeting times and dates are reviewed and agreed annually.

Formal meetings include monthly and special meetings. Special or extraordinary meetings can be established in accordance with the process outlined in the Model Rules of Association.

Informal meetings can be held at any time between any Committee members, GSNV staff and other stakeholders.

Meeting decisions must be passed by a quorum at formal meetings to be valid decisions.

The GSNV Group Leader is present at Committee meetings by invitation.

MEMBERSHIP

Committee of Management membership seeks to represent the skills, knowledge and experience required to fulfil the purpose and functions.

New members are targeted and invited to join the Committee to match an identified gap in skills, knowledge or experience. The Committee reviews the current skills and identifies any gaps as part of the annual strategic review.

Executive positions are voted in annually at the Annual General Meeting (AGM) in accordance with requirements outlined in the Model Rules of Association.

New general Committees members are also voted in at this time. No membership is accepted outside the AGM.

Committee members are advised that absence from three consecutive meetings without a formal apology will be considered a resignation from the Committee. Apologies must be in writing to the Secretary prior to the meeting. The Secretary must advise the President in the case of an apology.

CONFLICTS OF INTEREST

All meetings require the declaration of any conflicts of interest and are dealt with as required by the Model Rules of Association.

CODE OF CONDUCT

General Conduct

The Committee of Management expects its members and the GSNV staff to conduct themselves in a professional manner at all times. Members must adhere to the bullying, harassment and other relevant policies of the GSNV.

The Committee of Management and GSNV staff expects that everyone will perform their duties conscientiously, respectfully, honestly, and in accordance with the best interests of the GSNV.

Committee Members must not use their positions or the knowledge gained as a result of their positions for private or personal advantage. Conflicts of interest will be declared.

Committee Members must not accept entertainment, gifts, or personal favours that could, in any way, influence, or appear to influence decisions in favour of any person or organization with whom or with which the GSNV has, or is likely to have, business dealings. Similarly, Committee Members must not accept any other preferential treatment under these circumstances because their positions might be inclined to, or be perceived to, place them under obligation to return the preferential treatment.

Any and all claims for reimbursement from Committee members must be advised and approved by the GSNV Group Leader and provided to the GSNV administration delegated.

Committee Members may not receive payment or compensation of any kind, except as authorised under GSNV's business and payroll policies. Any breach of this rule will result in immediate termination and prosecution to the fullest extent of the law. Any claim for reimbursement must be agreed prior to the cost being expended.

Committee reimbursements for costs related to travel to and from Committee meetings, events and special purpose gatherings are capped at $200.00 per claim. All travel claims must follow the travel reimbursement policy and all other claims will only be obliged with the provision of original receipts and proof of payment.

Committee recommendations for spending GSNV funds or incurring any reimbursable personal expenses, must to ensure that good value is received for every recommendation and follow prescribed procedure.

Accurate and reliable records of many kinds are necessary to meet GSNV's legal and financial obligations and to manage the business of GSNV. GSNV's records must reflect in an

accurate and timely manner all business transactions and be made available to the Committee in detail on request and in general as part of monthly financial reporting. Committee Members must not make or engage in any false record or communication of any kind, whether internal or external.

Access to personal information will be limited to those with a legitimate reason for seeking that information. The Committee will use only personal information for the purposes for which it was originally obtained. Obtain the consent of the person concerned before externally disclosing any personal information, unless legal process or contractual obligation provides otherwise.

Dealing with external people and organisations

Committee Members must take care to separate their personal roles from their Committee position when communicating on matters not involving GSNV business.

Committee Members must not use GSNV identification, stationery, supplies, and equipment for personal or political matters.

When communicating publicly on matters that involve GSNV, Committee Members must not presume to speak for the GSNV on any topic, unless they are certain that the views they express are those of the GSNV, and it is the GSNV's desire that such views be publicly disseminated.

OBLIGATIONS AND COMPLIANCE

All members of the GSNV Committee of Management will:

- Comply with the Committee's Terms of Reference
- Comply with all requirements of the Model Rules of Association
- Declare conflicts of interest at the commencement of each meeting

CORPORATE GOVERNANCE

The 2014-15 period has been dynamic for the GSNV. In September 2014, the GSNV conducted a special meeting to change our financial year end date from financial year to calendar year. This motion was carried and calendar year reporting is now in place. Our vigilance during the transition, which necessitated an extended reporting period (July 1, 2014 to December 31st 2015) has been 'top of mind' for the Committee and featured during our monthly meetings.

In December 2014, the Committee of Management reviewed the GSNV existing Strategic Plan as part of our ongoing governance to determine its relevance for the quickly evolving genetics environment. As a result, a number of sub-committees were formed to strengthen our advocacy role in the sector.

Over the reporting period, the Committee of Management was consolidated at 13 members ensuring a spread of representation, expertise and resources. It remained a key goal of the Committee to ensure diverse representation including individuals living with and affected by genetic/rare 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 generally met on the third Thursday of each month at the Murdoch Children's Research Institute (MCRI). This was revised for a short period (with general meetings being held every second month) when the sub-committee structure was functioning to provide scheduled time for areas of key focus. Each meeting, 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.

Over the reporting period, these have included:

- Marketing, fundraising, engagement and image
- Financial management
- Strengthen support groups and stakeholders
- Service review and accessibility
- Stocktake of service and customer needs and Committee performance
- Youth
- Professional development and volunteers
- Advocacy: influence and reputation

A Finance Sub-Committee has been operational since 2013-14, and continues to bring 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 2016. Cannot wait to report our progress to you in 2017.

STAFF AS AT DECEMBER 2015

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

In the period between July 2014 to December 2015, the GSNV has continued to experience staff and workforce planning change.

Goodbye to Anna Jarmolowicz and welcome Emily Allen.

In February 2015 the GSNV sadly said goodbye to our wonderful administrative assistant, Anna Jarmolowicz. With no task to big or too small, Anna provided assistance and leadership on GSNV operations and was wonderfully proactive in doing so. Anna supported many areas during the Group Leaders stint abroad and assisted the committee in keeping on track with administrative obligations. We thank Anna for her amazing efforts, contribution, skills and ease during her time with us. Although it is always hard to say goodbye we fully support Anna's career progression and wish her all the very best in her genetic counselling role with the VCGS.

We welcome to our team Emily Allen (second year Master of Genetic Counselling) who will continue on from Anna and be our go to person for all administration tasks. Emily has already launched into the role and identified a number of areas for review and 'tidying up'. In the interest of running a green office and remaining efficient, Emily is looking closely at our electronic filing system and data storage. We look forward to greater efficiencies under the watchful eye of Emily and support her dual role as our trusty administrative assistant and as a training genetic counsellor.

The GSNV currently operates with a maximum of 2.5 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 Counselling Course (University of Melbourne) resulting in a

high staff turnover, 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 all GSNV current staff for their initiative and dedication and indeed for their support to the Group Leader. Thank you to Keri Pereira, Nancy Amin, Emily Allen and Victoria Rasmussen for your diligence, dedication, commitment and patience (with the Group Leader's Wi-Fi and internet issues overseas) and we look forward to working toward great things as an effective team.

In remaining co-located within the VCGS/MCRI environment, the GSNV adopts the MCRI corporate services policies and procedures, and delegates authority to MCRI to facilitate corporate services on our behalf. We thank all corporate services areas, but particularly HR, IT, Payroll and finance for their support of our team and for helping us to run efficient and smooth operations. Again we say thank you to David Amor 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 committee and in 2014-15, this focus has provided many positive outcomes.

EFT Allocation as at 31 December 2015

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	0.4
Nancy Amin	Administrative Assistant and Communications	CS	0.4
Victoria Rasmussen	Research Assistant	PT	0.2
		TOTAL	2.3
		TOTAL Budgeted EFT	2.5

PROFESSIONAL DEVELOPMENT

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

Staff Member	Program
Louisa Di Pietro	**DNA of Management, Update Training**
Anna Jarmolowicz	**Peer Support Training – Chronic Illness Alliance (CIA)** The course introduced the CIA's new 'Best Practice Framework', which covers all aspects of managing a chronic illness peer support program from getting started through to program evaluation, as well as their new Peer Leaders Online Training (PLOT), a step by step guide to setting up and running a peer support group.
Keri Pereira, Anna Jarmolowicz	**HGSA 39th Annual Scientific Meeting (Perth)** The theme of the conference was *Rare Diseases and Indigenous Genetics*, which explored genetic counselling services for women with Intellectual disabilities, and the development of legislation and policies supporting people with rare diseases. The GSNV presented a poster at the meeting.
Keri Pereira	**6th National Paediatric Bioethics Conference (Melbourne)** The theme of the conference was *Things that keep us awake at night*. The GSNV was interested in presentations including *Sequencing a child's genome – what should we do with the 'grey' findings?* and *Moral distress, ethical uncertainty and mere tragedy*.
Keri Pereira, Nancy Amin	**7th National Paediatric Bioethics Conference (Melbourne)** The theme of the conference was *Child health ethics: In the hospital and beyond*. The GSNV was interested in presentations including advocacy for children from asylum seeker backgrounds, caring for children with extreme medical needs, and issues regarding access to disability support services.
Nancy Amin	**Using Social Media & Other Platforms: Risk Or Opportunities? (Health Issues Centre)** The course focused on the purpose of having an online presence for support groups, the audience that we want to target, platforms, and the risks of using social media and ways of dealing with the most common concerns.
Nancy Amin	**Next Gen Sequencing Workshop (Melbourne)** The course covered the background behind next generation sequencing, the technology and how it is used to identify familial disease, and how to determine if a variant causes disease.
Nancy Amin	**Fragile X Update and Family Day (Melbourne)** Presentations by Fragile X researchers and clinicians focused on newly characterised symptoms in carriers, and new treatments and therapies.
Nancy Amin	**Leukodystrophy – Past, Present and Future (Melbourne)** Presentations by neurologists, researchers and a parent, introduced the medical background behind what causes the symptoms in children with Leukodystrophy, a family story about how finding the gene change in their child lead to a treatment, and an update on advances in stem cell and gene therapies.
Keri Pereira, Anna Jarmolowicz, Nancy Amin	**Grand Rounds** 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	**VCGS Friday Functional Genomics Seminars & Clinical Laboratory Interface Meetings** 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.

11th Meeting of the European Human Genetics Societies Sunday, June 7, 2015, Glasgow – Scottish Exhibition and Conference Centre Seminar Suite

Staff Member	Program
Louisa Di Pietro	**European Society of Human Genetics (ESHG) and European Meeting on the Psychosocial Aspects of Genetics (EMPAG) International Meetings (Glasgow, Scotland)** Presentations focused on the epigenetic basis of disease, gene editing technologies, and current issues in genethics, psychosocial issues in cancer genetics, reproductive decision making, ethical issues in genomic testing, and family dynamics and communication. The GSNV presented a poster at the meeting.
Anna Jarmolowicz	**Rare Voices Australia Rare Disease Summit (Melbourne)** The summit showcased current initiatives to progress a National Plan for Rare Diseases, to drive coordinated and collaborative action among stakeholders to address important, common needs and concerns of people living with a rare disease.
Emily Allen	**FSHD Information Night (Melbourne)** Presentations by experts specialising in Facioscapulohumeral muscular dystrophy (FSHD) spoke about infantile FSHD and the different types of assessments that are used, the genes involved in FSHD, symptoms and how it differs from other forms of muscular dystrophy, and recommendations for care of those affected.
Louisa Di Pietro	**Health Care Guidelines on Rare Diseases. Quality Assessment (Rome, Italy)** Provided training on the use of the AGREE II quality assessment tool, developed under the Rare Best Practices EU project and designed to assist in the identification of best practice guidelines in rare diseases clinical treatment. AGREE II has become the gold standard tool and is now adopted by many EU countries.
Louisa Di Pietro	**XV111 National Congress on Clinical Genetics (Rome, Italy)** This national congress represents an opportunity for clinical genetics and health professional to present important research findings and discuss complex and interesting case studies, particularly in the undiagnosed category. The focus of this conference was on genetic disorders of connective tissue (e.g. Marfan Syndrome), childhood syndromes and dysmorphia. The GSNV presented a poster at the meeting.
Louisa Di Pietro	**Dysmorphology Club (Bologna & Rimini, Italy)** A quarterly meeting discussing clinical perspectives, case studies and reflective practice. This is an important initiative in the rare diseases and undiagnosed area of clinical genetics.
Louisa Di Pietro	**5th Alpha-1 Global Patient Congress (Barga, Italy)** This meeting drew 200 people from 26 countries to hear renowned Alpha-1 scientists, clinicians, experts, industry partners, patients, caregivers and family speak on the status of the latest research, the regulatory framework for licensing therapies for rare disease in Europe, the status of patient registries and ways to strengthen the Alpha-1 message globally. The Congress was the first organized by Alpha-1 Global, a program of the Alpha-1 Foundation dedicated to building a collaborative network of organizations for patients and their families worldwide. It was launched as a result of the Barcelona Congress held in 2013, where attendees decided to create a way to encourage communications between Alphas around the world. Since then, a website has been launched as a global platform to support the community with digital tools and content.

Committee Member	Program
Abbie Kinniburgh	**GSNV Peer Support Training**
Abbie Kinniburgh	**Leadership Training – 'I CAN' Network**
Moira Rayner	**Walter and Eliza Hall, Human Ethics Review Committee**
Rachel Pope-Cousten	**Melbourne Genomics Alliance 2 Day Workshop** The workshop covered information about the Melbourne, Australian, and international Alliances. Understanding genetic testing platforms, interpreting complex genomics data accurately and accessing international data.
Maree Maxfield	**Masters of Public Health (The University of Melbourne)**
Maree Maxfield, Abbie Kinniburgh	**Autism CRC (Brisbane)** The Cooperative Research Centre for Living with Autism (Autism CRC) is the world's first national, cooperative research effort focused on living with autism, attracting world-renowned researchers to meet the challenge; taking a 'whole-of-life' approach to autism, from early diagnosis, to education and support throughout adult life.

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

PRACTICAL GENOMICS WORKSHOP

Genomics is the new frontier of genetic testing and diagnosis. New technologies, known as next generation sequencing (NGS) are allowing the de-coding of whole exomes and genomes. One way to think of the new technology is to start by imagining your genetic information as one giant recipe book. Each recipe is the genetic code for how an individual component of a person develops and functions. Just like with recipes, there can be enormous variation between the recipes that each person, or family, uses, even for the same dish. However, some changes in the recipes can mean that they don't turn out quite as expected or can't be made sense of at all. These changes can be as small as a single letter, like a spelling mistake, or they can be like a missing word or paragraph.

In the past, if someone was thought to have a genetic condition, we would have to be very certain of which gene was responsible before we could use a genetic test to look for these changes. Sort of like needing to know which recipe you are looking for in order to turn to that page and read it. NGS allows the whole book to be read from start to finish and all the variations catalogued. Of course, with that much data to interpret, there are also huge challenges including how to 'read' all that information and how to work out what is variation that is expected but what is variation that can impact on the way a person functions, to the point of them being diagnosed with a genetic condition?

Being a genetic counsellor practicing as this new frontier arrives is incredibly exciting; we know how important it is to many families to understand the cause of the condition their loved-one may have. However, it is also incredibly challenging as it brings with it uncertainty for me and also sometimes for families and individuals that we see. We do not yet understand all of the human variation in the world. Sometimes the testing reveals unexpected information due to the level of genetic detail being seen. Part of my role is to help people navigate the uncertainty or consequences of an unexpected finding.

The Practical Genomics Workshop was hosted by the Melbourne Genomics Health Alliance and was an opportunity for genetic health professionals to get some hands on experience interpreting NGS information and in working with individuals and families with this technology. The Melbourne Genomics Health Alliance is a partnership between The Royal Melbourne Hospital, The Royal Children's Hospital, The University of Melbourne, The Walter and Eliza Hall Institute, the Murdoch Childrens Research Institute, the CSIRO, the Australian Genome Research Facility, the Peter MacCallum Cancer Centre, Austin Health and Monash Health.

This Alliance is using their resources to offer NGS to more people than ever before in Australia. At every stage, research is being undertaken to better understand the experiences of people directly impacted by genetic conditions and their families.

The Melbourne Genomics Health Alliance is also working closely with the Global Alliance for Health and Genomics and the Australian Genomics Health Alliance, putting Victoria on the national and international stage for genomic research.

A major goal for me as a genetic counsellor and member of the GSNV Inc. executive is to continue to build relationships with the health-professionals and organisations involved in this genomic research, particularly the research into the impact of testing on individuals and families. There are many opportunities on the horizon for the GSNV Inc. Team Leader, committee, and membership to help shape the direction of genomics and ensure that is used to maximum benefit with the least negative impact possible. It is an important time to be working in this field and I am grateful for the opportunity to represent the GSNV Inc. as I continue on this journey.

Rachel Pope-Couston

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 22 volunteers and 11 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 Mitochondrial Disease Foundation	2	Assistance with mito information day and assistance with website
Heartkids	1	Assistance with Gala
Psoriasis Australia	1	Assistance with office administration tasks
Scleroderma Victoria	1	Assistance with mail-out
Children's Tumour Foundation	1	Assistance with camp
Syndromes Without a Name	1	Assistance with mail-out
Australian Alopecia Areata Foundation	1	Assistance with thank you certificates
UsherKids Australia	1	Assistance with website

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

STAKEHOLDERS

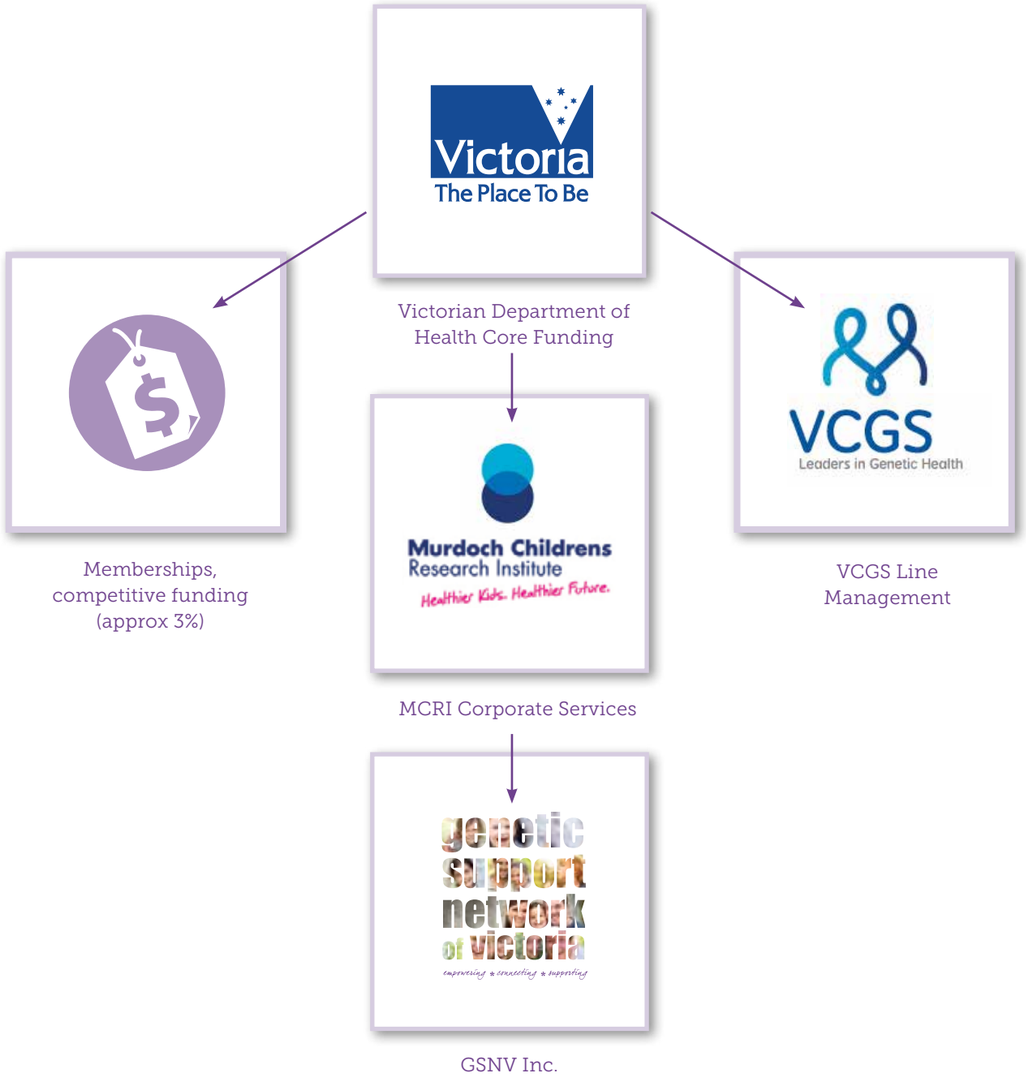
The GSNV team has continued to work closely with the Murdoch Childrens Research Institute (MCRI) and the Victorian Clinical Genetics Services (VCGS) and the Department of Health (DOH) Victoria to create open, transparent and effective corporate relationships that are mutually beneficial.

As major stakeholder groups, each plays and essential role in the sustainability of the organisation 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 from 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 18 months the GSNV has continued to support Victorians impacted by a genetic and or rare condition and engaged with our interstate counterparts to ensure all Australians are getting the support they need.

With demand for our services on the rise and our operating environment changing, 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 hard at work in Victoria for nearly twenty years and we have a long history of 'people helping people'. This year we have concentrated our efforts on rare diseases and genomics, ethical medical genetics, volunteering and governance strengthening. All efforts in these areas, will inform our future direction and help us to improve what we do day to day.

As we review the period since the previous AGM it is important that we continue to focus on building our strengths, engaging our community and stakeholders and empowering vulnerable people impacted by a genetic condition.

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 evidence based approach
- Advocating for change and improvement
- Mobilising people
- Inclusive and engaging
- Working collaboratively with national and international partners

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 work focusing on our governance and operations structures. These will have influence on strategic and operational matters in the coming months.

Terms of Reference and Conflict of Interest

In seeking to improve our governance structure and strengthen the capacity of committee the GSNV has reviewed the committee terms of reference (TOR) and articulated new terms. These have provided clearer guidelines to our committee and assisted in establish best practice governance. In addition, the GSNV has considered the recommendation of the *Reform Act* 2012 and articulated a conflict of interest policy, which guides committee on declarations of conflict.

In maintaining dual roles and managing a number of different interests, it plausible that from time to time committee members may have a conflict to declare. This initiative is deemed important in maintaining an ethical approach to governance and upholding transparency across all committee activities.

INVESTING IN THE SUPPORT OF PEOPLE SUPPORTING EACH OTHER

The GSNV is committed to the empowerment of support groups and support group leaders 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 invests in our most important stakeholder group and contributes to the improvement and delivery of support group outcomes.

In February this year the GSNV financially supported the visit of Karni Liddell, Paralympian (living with a neuromuscular condition) as guest speaker at the second Neuromuscular Support Group for Young Adults meeting, Melbourne.

The GSNV has been instrumental in setting up this new support group for young people and has a permanent online presence for the group on our website. This session was made possible through a GSNV small grant, and a further subsidisation of the event to bring Karni Liddell to Melbourne as the keynote speaker. The GSNV also offered significant in-kind and administrative support. This project and support of the Neuromuscular Support Group for Young People is on target with GSNV's mission to empower and support the genetics community and play a leading role in developing and facilitating support and information services. This event also facilitated closer ties with other genetic services and hubs in Victoria as the project was co-developed with a leading genetic counsellor, specialising in neuromuscular disorders and working at Melbourne Health. Invitations and information on the event was widespread and targeted genetic health professionals throughout Victoria.

Every year the GSNV offers Small Grants to members for projects that assist the important 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, events, and the 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 2016 Small Grant recipients in December of this year.

2015 SMALL GRANT RECIPIENTS

Organisation/Group	Description of project
Lymphoedema Association of Victoria	Seeking funding/support for postage to send out the newsletter each quarter
Australian Pompe's Association	Funding to cover printing, stationary, mailing costs to add Pompe's to NBS register
Australia Alopecia Areata Foundation Inc.	Purchasing of 500 window stickers to distribute to hairdressing boutiques and salons
Aspergers Victoria	Safe Talk Workshop upskilling volunteers, depression and anxiety peer support
SIDS and Kids Limited	Administrative and financial support to run Difficult Decisions support group
UsherKids Australia	Videos for website and promotional flyers
CHARGE Syndrome Association of Australia	Cover costs of support group meetings
Hearts In Mind	Home office expenses for two facilitators
People With Multiple Sclerosis Vic Inc.	Provide taxi vouchers and petrol vouchers to assist people who may experience difficulty in attending because of the costs associated with special transport as well as accommodation costs

GSNV PEER SUPPORT TRAINING FOR LEADERS – MARCH 2016

The GSNV ran a Peer Support Training session for leaders to complement our Genetic Support Awareness Week (GSAW) and strengthen our position as an information provider. The session was co-developed and facilitated by a trained professional from Cystic Fibrosis Victoria (CFV), who has extensive experience in delivering training in a similar environment and with similar audiences. The identification of CFV as a suitable training provider comes from my involvement in the CFV carrier screening community awareness program, which I have been a current board member of for over two years.

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. At every opportunity, we work hard 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 (EMPAG) meetings in Glasgow, Scotland and Barcelona and the Italian Society of Human genetics (SIGU) meeting in Rimini. All three 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 Catherine Beard, I presented research in Glasgow and Rimini that looked specifically at the experience of carriers identified through a population reproductive genetic carrier screening panel in Australia.

Every year the GSNV supervises one or more Master of Genetic Counselling thesis research projects and has direct input into the research outcomes. In maintaining a strong and reputable position as a peak body and representative voice, GSNV involvement in research outcomes is very important. Journal publication and the presentation of our research at national and international meetings are the main goals of our research involvement. It is always a pleasure to be the presenting author of outstanding projects.

This year the GSNV has embarked on major independent research project designed to evaluate our services and generate important finding on what the future of our services may look like. This independent project is an important step that will assist us to develop an evidence-based model to present to funding agencies in the future. The GSNV is keen to explore a number of models that may best meet our needs in the future and further improve how we support and deliver to the genetics community.

Significant headway has been made with the study since project planning began in June 2015. The project's low risk ethics application has been peer reviewed and has successfully passed all requirements under the HREC ethics review process.

Stage 1 of the study has been completed with the GSNV enquiry data (2010-2015) having been cleaned and coded for analysis. Stages 2 & 3 of the study – focus groups and quantitative surveys are currently in progress.

A poster presentation of the preliminary findings of this research has been presented in Barcelona Spain at the Psychosocial Aspects of Genetics (EMPAG) meeting. A presentation of the findings is all intended for the Human Genetics Society of Australasia (HGSA) Annual Scientific meeting, Hobart. We hope to also publish the findings in a suitable peer reviewed journal.

RECENT PUBLICATIONS

"I'm Healthy, It's Not Going To Be Me": Exploring Experiences of Carriers Identified Through a Population Reproductive Genetic Carrier Screening Panel in Australia.
American Journal of Medical Genetics
Louisa Di Pietro – co-author

"Survey of healthcare experiences of Australian adults living with rare diseases"
Orphanet Journal of Rare Diseases.
Louisa Di Pietro – co-author

SNAPSHOT

genetic support network of victoria

*empowering * connecting * supporting*

An evaluation of The Genetic Support Network of Victoria: A mixed methods study based on stakeholder participation

Louisa Di Pietro,[1] Victoria Rasmussen,[1] Keri Pereira,[1] David Amor,[2] Justine Elliott,[2] Rosemary McKenzie,[3] Laura Forrest,[4] Monica Ferrie,[5] Nancy Amin,[1] Paul Fennessy[6] and on behalf of the Genetic Support Network of Victoria.

1. Genetic Support Network of Victoria, Murdoch Childrens Research Institute, Melbourne, Australia
2. Victorian Clinical Genetics Service, Murdoch Childrens Research Institute, Melbourne, Australia
3. Centre for Health Policy, The University of Melbourne, Australia
4. Peter MacCallum Cancer Centre, Melbourne, Australia
5. The Genetic Support Network of Victoria Committee, Melbourne, Australia
6. The Department of Health Victoria, Australia

INTRODUCTION:

Individuals affected by genetic and rare conditions have unique support and health care needs. Supportive services seeking to provide appropriate and accessible help must firstly understand the needs of these individuals and how they interact within a rapidly developing genetics health environment. This study aims to determine the information and support needs of individuals who had previously accessed the Genetics Support Network of Victoria (GSNV) and to explore their beliefs and attitudes toward the network, including level of satisfaction with services provided. Taking a participatory evaluation approach, the views of stakeholders and GSNV staff members were additionally explored to uncover avenues for organisational improvement.

MATERIALS AND METHODS:

Participants were GSNV service users who had accessed the service within the past 12-months (e.g., individuals affected by genetic and rare disease, professionals working in clinical, research and education settings and support group organisers), representatives from key stakeholder groups and GSNV staff members (n=40). Five focus groups were conducted with participants grouped together based on the aforementioned participant categories. Adopting a phenomenological approach, the focus group discussions were transcribed verbatim and coded using thematic analysis to identify major themes.

RESULTS:

Preliminary data will be presented revealing important themes identified across the group discussions related to participants' experiences seeking support from the GSNV, beliefs and attitudes towards the network, unmet information and support needs and ideas for organisational growth and change.

CONCLUSION:

This research will provide important insight into the experiences of members of the genetics health community, including their help-seeking behaviours. The findings may enhance the appropriateness and utility of GSNV services and will provide crucial data to inform the development of genetic support structures elsewhere in Australia.

GSNV EVALUATION STUDY

In July 2015, the GSNV commenced an extensive internal evaluation in collaboration with important stakeholders. The overarching aims of the evaluation study are; Firstly, to determine the extent to which the GSNV is meeting the needs and expectations of its service users and stakeholders and fulfilling wider strategic objectives and; Secondly, to explore individuals' attitudes toward and beliefs about the GSNV, including satisfaction with services provided and ideas for organisational growth and change. Ethical approval for the study was granted by The Human Research Ethics Committee (HREC) at The Royal Children's Hospital.

The evaluation study will collect data on GSNV service users, stakeholders and staff members' experiences and perspectives across three key stages: 1. Analysis of the GSNV enquiry database; 2. Focus groups; and 3. Quantitative survey. The project design was guided by a supervisory board consisting of academics with expertise in evaluation and qualitative research and representatives from the following key stakeholder groups: the Department of Health Victoria, Victorian Clinical Genetics Services (VCGS), Victorian genetics health professionals and individuals affected by genetic disease. The supervisory board, chaired by Keri Pereira (senior GSNV staff member), continues to oversee the study and make important decisions about data collection and analysis on a bi-monthly basis.

Analysis of service user enquiries (Stage 1) has provided insight into the personal context of individuals using GSNV services and facilitated the identification of specific service user subgroups' (e.g., individuals affected by disease, health professionals and family members) information and support needs. Enquiry data spanning 2010-2015 has revealed that individuals affected by genetic and/or rare disease and their family members were the most frequent users of the network (46%), followed by health professionals (25%) and support group organisers (21%). Over forty percent of all enquiries made by affected individuals and family members involved seeking information regarding genetic/health services and health conditions. Support group and/or peer support connections (35%), clinical and health service referrals (15%) and requests for specific GSNV services (e.g., advocacy letters) (9%) were among the most frequently recorded types of assistance requested by this group.

Focus group (Stage 2) and survey (Stage 3) methodology will be employed to gain an in-depth understanding of participants' information and support needs and expectations of specialised genetics support services in the community. Recruitment for the five focus groups segregated by participant group (e.g., health professionals, family members, etc.) began March 2016 with the first three groups expected to run in April and May 2016. Once the focus groups have been completed, the final stage of the study will commence and an anonymous quantitative survey will be distributed to service users drawn from the GSNV database. The preliminary research findings will be presented by Louisa Di Pietro at the European Human Genetics Conference, Barcelona in May. We hope to disseminate the aggregate findings in full at a national conference later this year and through publication in a peer-reviewed journal.

Victoria Rasmussen

RECOGNISING OUR COMMUNITY AND CELEBRATING THE FACES OF GENETIC AND RARE CONDITIONS

Facing Forward Photo Book

The GSNV has proudly produced an enduring portrait of the wonderful children, adolescents and adult that make up our genetics and rare disease community. In celebration of the GSNV Genetic Support Awareness Week (GSAW) and Rare Disease Day 2015, this book is a permanent memory of our successful 'Facing Forward Campaign' and those who contributed so generously in providing their picture and words to us. We have made the book available for purchase. This project was generously supported by an education grant from Sanofi.

SANOFI

'Show Us Your Support'

GSNV Genetic Support Awareness Week 'Show Us Your Support' Social media campaign 2016 followed on from the Facing Forward campaign idea and aimed to give the public a visual insight into the genetic and rare diseases support groups around Australia. An additional goal of the campaign was to raise awareness of the importance of support services in genetic and rare disease health. Over Genetic Support Awareness Week, the GSNV used social media as a wide reaching platform to showcase these images.

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, genomics and the needs of consumers. We look forward to reviewing our advocacy strategy in the latter part of 2016 and delivering effective outcomes.

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 “arm’s length” representation when needed
- Empowering GSNV member groups to lobby and approach local, State and Federal government on issues that affect them
- Raising issues and providing feedback on a range of topics
- Focusing on access and equity in genetics/genomics health care

What is the GSNV doing in Victoria around the issues of access and equity?

The GSNV has recently considered access and equity as core issues to our genetics and rare diseases community. In accordance with the strategic objectives, vision and mission of the GSNV, we have established a working group/sub-committee which will seek to identify common themes and issues in relation to ‘access and equity’ and what these two crucial areas may mean for the genetic and rare diseases community. This group will seek to identify critical areas where access is of concern to the genetics and rare diseases community and critical areas where equity is of concern. In each case this working group will seek to develop an action, response and or strategy that addresses the issue and provides advocacy.

The GSNV works with many families, children and adults impacted by a genetic and rare disease. We learn about their stories and ongoing journeys, articulating what the ‘burden of care and cost’ really means. In having conducted five consecutive years of Rare Disease Day activities, and in collecting feedback around this day, overwhelmingly, access (to services, health professionals, information and research) and equity (affordable services) are reported as the top priorities for Victorians impacted by a genetic and rare disease year after year. Our subcommittee will work towards achieving some outcomes on behalf of our members and community in the months to come.

In recent times the international media has taken particular interest in new genetic technologies or genome wide association studies (GWAS). Recent news articles include studies that have identified genetic modifiers of Huntingtons Disease, pancreatic cancer, leukaemia and schizophrenia risk.

GWAS are particularly useful in finding genetic variations that contribute to common and, complex diseases, such as asthma, cancer, diabetes, heart disease and mental illnesses.

A genome-wide association study is an approach that involves rapidly scanning markers across the complete sets of DNA, or genomes, of many people to find genetic variations associated with a particular disease. The impact on medical care from genome-wide association studies could potentially be substantial. This research is also leading the way for an era of personalised medicine, in which the current ‘one size-fits-all’ approach to medical care will give way to more personalised strategies. Currently cancer genetics is making headway in personalised medicine.

With future improvements in the cost, efficacy and efficiency of genome-wide studies and other new genetic technologies, genetic services will have the tools to provide patients with individualised information about their risks of developing certain diseases, their prognosis and age of onset. In consideration of population health and health prevention strategies, the information provided by GWAS will enable health professionals to tailor prevention programs to an individual’s unique genetic makeup.

NEL’S PLACE

The GSNV has recently become involved with a new initiative and becoming a driver in the space around BRCA 1 & 2 support in Victoria. Nel’s Place is a support group for individuals affected by the BRCA (breast and ovarian cancer) gene changes.

Lisa Penman who carries a BRCA2 gene change is establishing the group. Lisa identified that there was no support group for individuals who are BRCA positive in Victoria and very little in Australia overall. She has also established that there is a lack of services available in the public health system. Nel’s Place is being developed to provide emotional support and information to individuals with BRCA mutations and the GSNV is on board in collaboration with the Cancer Council of Victoria to address the information and support gaps of this community.

Nel’s Place is a support group for individuals diagnosed with BRCA (breast and ovarian cancer) genetic mutations, and was developed to offer support to individuals with BRCA 1 or 2 mutations who are at risk of developing breast or ovarian cancer but have not yet had a cancer diagnosis.

Nel’s Place’s short term goals are to:

- Provide support and information to those diagnosed with a BRCA mutation.
- Provide advice to those with BRCA mutations about screening and preventative treatment options.
- Provide information about the latest research into BRCA mutations.
- Create a safe and comfortable environment to enable people to voice their opinion without judgement.
- Assist those diagnosed with negotiating the medical system to achieve a positive outcome.

In the long term, Nel’s Place hopes to:

- Further research into familial cancer.
- Create a repository of information regarding BRCA mutations for those affected, medical professionals and the general public.
- Create an expert advisory panel of medical professionals for those diagnosed with a mutation.

Nel’s place is the passion of Lisa Penman, 43 years old of Melbourne, Australia. Lisa was diagnosed with a BRCA2 gene mutation in 2013. As a result of her diagnosis, Lisa has had preventative surgery to minimise her risk of developing breast and ovarian cancer. This has involved a full hysterectomy and prophylactic bilateral mastectomy.

Lisa and her siblings lost her mother to ovarian cancer in 2014 and Nel’s place is named in her honour. Nel was a teacher and musician with a strong history of familial cancer. She was also an identical twin and she and her sister participated in a number of genetic studies associated with cancer and other illnesses in order to provide assistance to the medical community and the general public.

Over the last 18 months 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 Education, Ethics and Social Issues Committee (ESIC)
- Consumer Health Forum
- Cystic Fibrosis Victoria Carrier Screening Reference Group
- Melbourne Genomics Alliance Consumer Advisory Group
- Victorian Genetics Advisory Committee
- 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

OUR FUTURE – STRATEGY 2016 AND BEYOND

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.

We have continued working towards the goals we set in 2015, 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 June 2016. 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. Our financial position is always a concern and our efforts in the short to medium term will concentrate on improving our position and expanding our capacity.

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:

- Strengthen our financial position
- Developing effective partnerships with partner agencies, governments and stakeholders to build our network
- Investing in our staff and committee
- Enhancing member engagement and membership growth
- Improving volunteer engagement
- Increasing our capacity to deliver services in all genetics hubs in Victoria
- Incorporated rare diseases into our philosophy and key messages
- Reaffirming the importance of the GSNV as a consumer representative and peak body

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 our 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 18 months. There have been significant contributions from the Executive and the Finance Sub-Committee.

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

Our successes have included:

- Further streamlining of governance processes
- An opportunity to reflect, review and improve current practice
- Reflect on our advocacy strategy
- Improve committee terms of reference
- Capacity analysis of staff and Committee
- Focus on our financial situation
- Review and add to our risk management strategy
- Developed a grants 'hit-list' and made applications
- Develop independent research
- Participation on the Victorian Genomics Advisory Committee

The GSNV is a wonderful place to work

The GSNV prides itself on providing flexible working arrangements and providing staff with opportunities to develop, improve and hone their skills. Over the past 12 months I have had the privilege of conducting my work with the GSNV while living in beautiful Rimini, Italia. It has been an amazing experience and it would not have happened without the support, vision and flexibility of the GSNV committee, team and MCRI corporate services. I am truly grateful to have had this opportunity to work in a remote location that is truly beautiful and inspiring.

In 2016 and beyond 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

Excerpt – An Expat Life

After arriving in Italy April 7 I hit the ground running and was attending the 5th Alpha-1 Global Patient Congress and the 2nd Biennial International Research Conference on Alpha-1 Antitrypsin in Barga, Lucca – Italy by April 9. The congress was superb and represented the very best of international cooperation, coordination and rare disease approach. I applaud the Alpha-1 Global Foundation for a very successful congress and for bringing together patient groups, the scientific/research community and clinicians in an open and cooperative environment. All stakeholders were given an opportunity to participate and contribute to an agenda that was interesting and engaging. It was apparent from the very beginning of the congress that the Alpha-1 international community is strong, active, supportive and very well led.

I was very privileged to be part of the Alpha-1 Australian contingency and I thank the Dr. Charlie Strange of the Medical University of South Carolina (MUSC) and the Alpha-1 Global Foundation for their support with getting me to beautiful Barga, Lucca. Steven Knowles of the Alpha-1 Association of Australia (AAA) was greatly missed but was absolutely with us in spirit. As a member of the Alpha-1 Global Steering Committee, Steven was integral to the organisation, and development of the congress. I also thank Steven for his vision in getting a few extra Australians to Italy (including me as a GSNV and Murdoch representative) and in facilitating an opportunity for key collaborators to meet and spend some time discussing future work.

The AAA was strongly represented by Jennifer Nankervis and John Arkinstall from Australia and it was wonderful to see the relationships they have developed with the international Alpha-1 community and the respect they receive from their peers. My experience at the congress was truly inspiring and in many ways, has set the scene for further opportunity, research and collaborations while I pursue my sabbatical (work) in Italy. I look forward to working with the Alpha-1 Australian community and progressing important projects with them.

Following the success of the Alpha-1 Global congress I had my feelers out and was keen to take up any immediate opportunities for learning and networking here in Italy. Upon invitation from some learned colleagues in the region of Emilia Romagna (Rimini) I was fortunate to attend the Regione Emilia-Romagna XXVIII Convegno – Anomolie Congenite Del Sistema Nervos Centrale, a one day workshop facilitated by the Gruppo Di Studio Sulle Malformazioni Congenite (I.M.E.R). It included discussions on dysmorphology, syndrome diagnosis, rare conditions of the central nervous system and case histories of disorders of the corpus callosum. With my very best Italian language understanding in progress, I was delighted to participate in the workshop and gain further understanding of the rare disease clinical and research environment here in Italy. With a long standing congenital malformations and anomalies of the central nervous system registry very active in this region, I was interested to read the I.M.E.R Annual Report and definitive data on such rare diseases in Italy.

Two congresses in two weeks has constituted a wonderful start to my working life in Italy and I look forward to following up on the networking conducted at both.

SNAPSHOT

Genetic Support Network of Victoria

9th Floor, South Building, Murdoch Childrens Research Institute
Flemington Rd, Parkville, VIC 3052
Ph: (03) 8341 6315 Fax: (03) 8341 6212
Email: info@gsnv.org.au Web: www.gsnv.org.au

To Whom It May Concern,

I write on behalf of the Genetic Support Network of Victoria (GSNV) in support of XXXX application for financial assistance, with regards to school aid for her daughter, XXX. XXX has a diagnosis of 22q11 deletion syndrome also known as Velocardiofacial syndrome (VCFS).

The GSNV is an active not-for-profit organisation committed to ensuring the well-being of people affected by genetic conditions. Although many genetic conditions are rare, such as 22q11 deletion syndrome, it does not mean that they do not carry a significant burden on the families living with the diagnosis. 22q11 deletion syndrome has a number of features including: cardiac abnormalities cleft palate and delayed growth. Another feature of 22q11 syndrome is that children may be affected with developmental delay, including delayed speech development and learning disabilities and as such, would require special aid for education. We understand that XXX's cardiologist, speech therapist, as well as the geneticist in charge of XXX's case are also writing letters in support of XXXX's application, which outlines XXX's specific medical concerns and educational needs. In addition to the cardiologist and geneticists letters, the family have provided diagnostic information, which greatly demonstrate, her need for further education support through school.

We have been in touch with XXXX's family a number of times and cannot stress enough the importance that families with rare genetic conditions do not miss out on the opportunities and assistance that other families with more recognised special needs receive. All too often these families are indirectly discriminated against because their condition is not recognised in the wider community. Individuals and families affected by rare diseases often live within the shadow of financial hardship while also coping with difficult condition specific issues, such as learning disabilities. As a government organisation, the GSNV deems that you have responsibility and a duty of care to ensure that families who legitimately require assistance receive it, regardless of whether their condition is rare.

We are very happy to be contacted if you wish to discuss this letter further.

With Kind Regards,

Keri Pereira

Keri Pereira, Genetic Support and Education Co-ordinator
On behalf of the
Genetic Support Network Victoria
E-mail: keri.pereira@vcgs.org.au
Phone: (03) 8341 6315

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) GP Roadshow

Two thousand and fifteen has brought a very exciting opportunity for the GSNV in terms of its collaboration with HEAL.

In previous years we have worked with HEAL to provide education seminars for regional health professionals. Unfortunately funding for this project was cancelled in 2015 however through the support of a NAB grant HEAL and GSNV as well as a number of other organisations have put together a General Practitioner Roadshow.

This event is aimed educating GPs on women's issues, including genetic related issues, which may present themselves at their clinics. The roadshows will start in the second half of 2016 and will include simulated scenarios, information session held over the course of one day.

Maternal and child health nurse seminars

In 2014 the GSNV was invited to present at the Maternal and Child Health professional development session.

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.

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 four times over 2014/2015.

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 2016.

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.

"Thanks for organising a wonderful course with Eilis Hughes. I received the laminated certificate today"

– Peer Support training participant

THE UNIVERSITY OF MELBOURNE

STUDENTS

University of Melbourne – Societal Issues and Personal Genomics

The GSNV participated in the University of Melbourne Societal Issues and Personal Genomics course in late 2014. With a focus on personal genomics the course explored the history of genomics, eugenics, what is personal genomics, what do consumers what to know about their genomes, direct to consumer testing and the future of health services with employment of sophisticated genetic technologies.

"Just a brief note to thank you for once again for participating in the Patient Perspective Sessions this month at Melbourne University.The feedback from the students already has been very positive and we appreciate the fact that you were willing to share your personal story with the medical students.

The students spend a lot of time in the first year of their course learning and practising the principles of good communication. These sessions allowed the students to hear your personal account of interacting with health professionals and the community in general.

I am hoping that you may be willing to come along next year when we run similar sessions again.Once again thank you for your time and enthusiasm joining us in this program."

- Patient Perspective Sessions course co-ordinator

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.

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.

Monash University – Biomedical sciences

The GSNV was again invited to present a guest lecture to the Monash University Biomedical Science students. In 2014 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."

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 organisation. These information pack support our presentations to various health professional and community groups.

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.

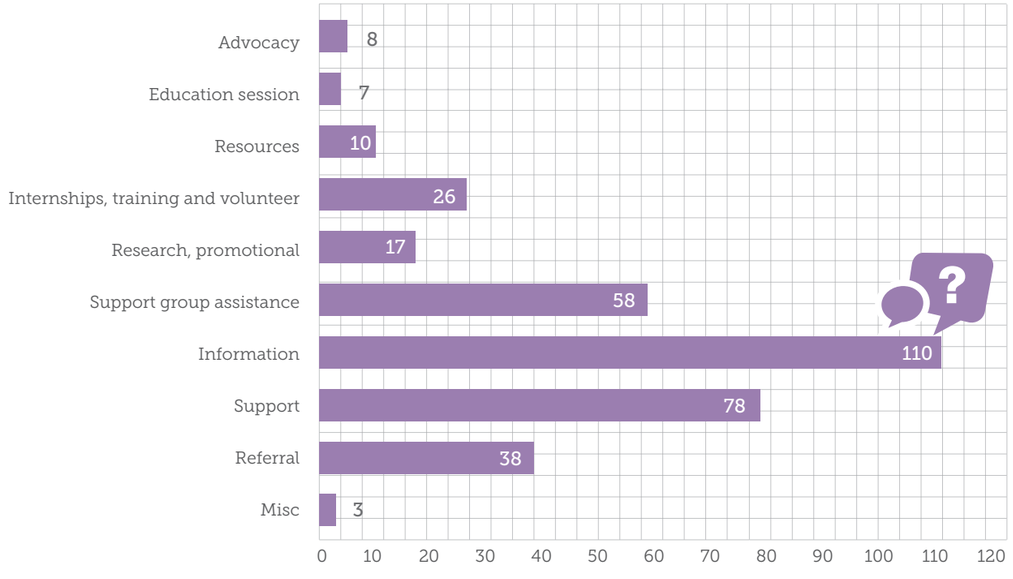
Providing support for support groups is a significant part of our role. We can aid support groups in the following ways:

- 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:

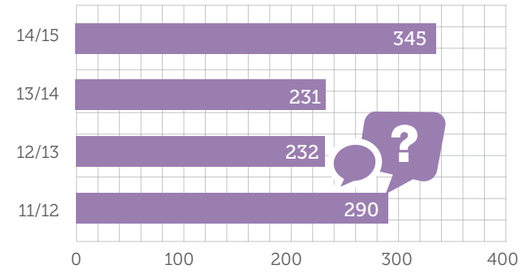
- Neuromuscular support group webinar – September 2014
- Syndromes Without A Name, Ambiguous Loss workshop – September 2014
- AusDoCC parent information day – October 2014
- Mitochondrial Disease Association – November 2014
- Rare Diseases Day – February 2015
- Australian X & Y spectrum support – March 2015
- Ichthyosis family information day – May 2015
- Wangaratta Support Group Meeting – Oct 2015
- UsherKids – support group setup
- CDH1- support group setup 2015
- Nel's place – support group setup 2016
- Neuromuscular Support Group Conference 2016

TYPES OF ENQUIRIES RECEIVED

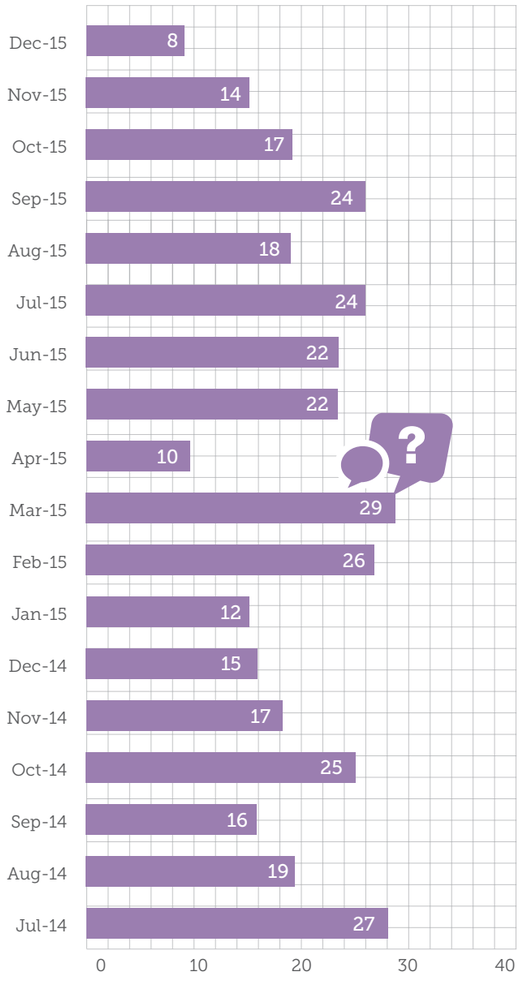


INFORMATION REQUESTS

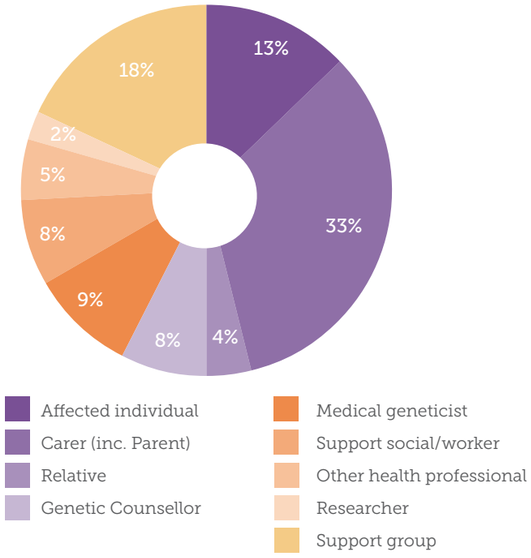
ANNUAL ENQUIRIES



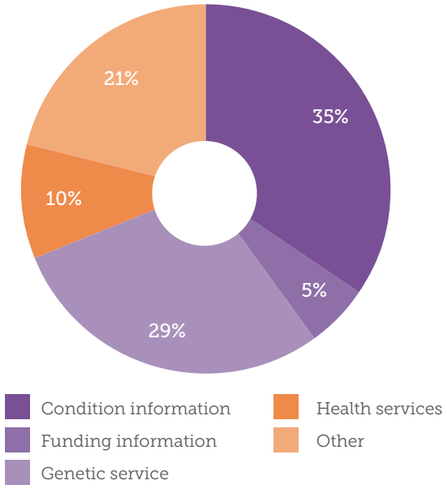
2014-2015 ENQUIRIES BY MONTH



WHO IS SEEKING SUPPORT?



TYPE OF INFORMATION REQUESTED



SNAPSHOT

ICHTHYOSIS MEET

By Carly Findlay, from her blog carlyfindlay.blogspot.com.au

Carly Findlay partnered with the Genetic Support Network of Victoria (GSNV) to host the first official Australian Ichthyosis Meet – held in Melbourne on Saturday 9 May 2015, as a part of Ichthyosis Awareness Month. This event gave adults, children and families affected by Ichthyosis the chance to meet others with this rare condition.

We had around 70 attend – and around 20 of those were affected by Ichthyosis.

The day featured three sessions: an informal meet and greet, a trip to the zoo, and a dinner for those who want to socialise further.

The first session was a panel discussion featuring a panel of medical specialists: Professor Ingrid Winship (genetics), Dr David Orchard (dermatology) and Nurse Practitioner Emma King (dermatology).They talked a lot about the research that's been done around Ichthyosis – locally and overseas.

After morning tea, we did a 45 minute activity, providing peer support in small groups and then with the larger group. This was just wonderful. All suggestions were valued, and when members of the small groups read out responses to the room, applause was given. I was so proud. Our facilitators did a great job if making everyone feel comfortable enough to share their experiences.

To end, we talked about what attendees would like to see after the Australian Ichthyosis Meet. What is the direction of the Australian Ichthyosis Community?

It was unanimous that people want regular meets – perhaps a national meet every second year, but regional and informal activities are encouraged. We can form a committee to develop the community into a charity.

And then it was off to the zoo!

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:

"Another terrific Newsletter – thanks – it provides so much info and great to know all that's happening"

Yvonne – Parent To Parent Program Co-ordinator, Gippsland

"Thank you all for a very informative news update. I have printed off the MedIDs.com because I think it is very useful. Congratulations on the great work you are all doing."

Anna – Australian Gorlin Syndrome Mutual Support Group

"Thank you and the team at GSNV for preparing the Bits & Pieces newsletters. They are such a great source of information and raising awareness of various fundraising events – I'd never heard of Super Boss Day before, but will be dobbing my boss in for sure!!"

Louise – HAE Australasia

"I received the Summer edition of the newsletter this morning, great read!"

Sarah – Project Coordinator, Programs & Support Services, Cystic Fibrosis Victoria

Thanks for your great service – including your wonderful, newsy newsletter.

Lindy – Genetic Counsellor

"That looks fabulous! You do a beaut job with the layout. Thanks for making me a part of the GSNV Newsletter."

Evelyn Bugel – Psychologist

We are going Green!

NEWSLETTER COMMUNICATIONS

96% of our communications have been electronically delivered via e-mail and 4% 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** and request to receive future newsletter editions by email.

PRINT AND E FORMAT

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

- A bi-annual 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.

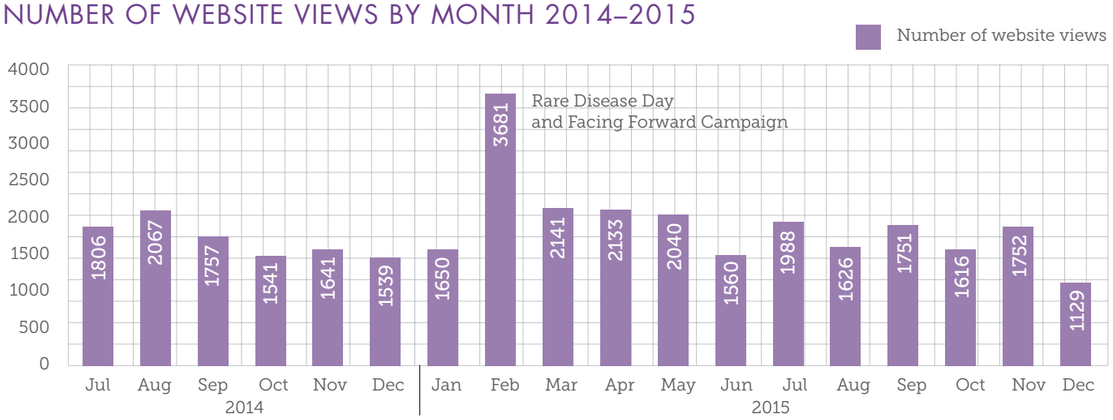
Our print communications represent a significant expenditure in our budget given that the GSNV has opted for a highly professional production. We are considering cost recovery and better efficiencies for the future and will look towards strategies in 2015 to reduce our expenditure.

WEBSITE, DATABASE AND SOCIAL MEDIA

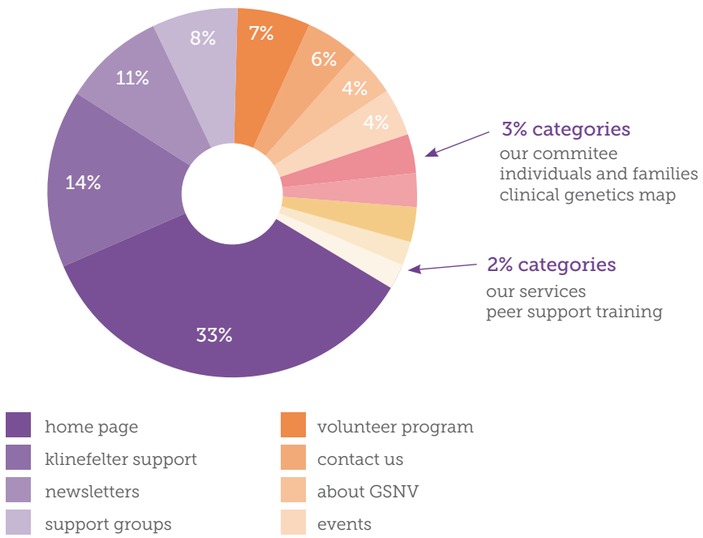
Social media remains and important tool in communicating to our members. In 2016 the GSNV will use social media campaigns and blogging to highlight our important events and information. Social media has been identified as an acceptable form of communication both formally and informally and is incorporated 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 management.

NUMBER OF WEBSITE VIEWS BY MONTH 2014–2015



WEBSITE PAGE VIEWS



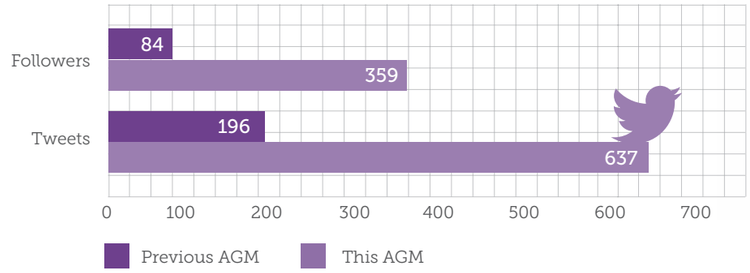
NUMBER OF WEBSITE VIEWS

33,378
1 July '14- 30 Dec '15

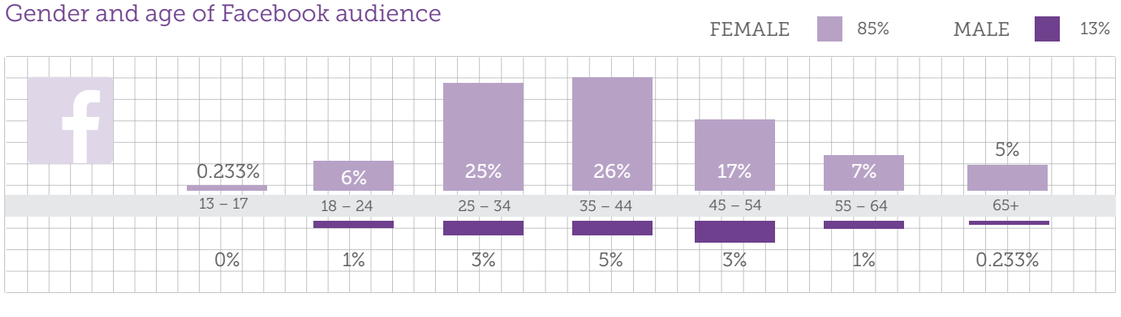
21,462
1 Jan '12- 30 June '13

WHO ARE WE REACHING ON FACEBOOK AND TWITTER?

Twitter reach

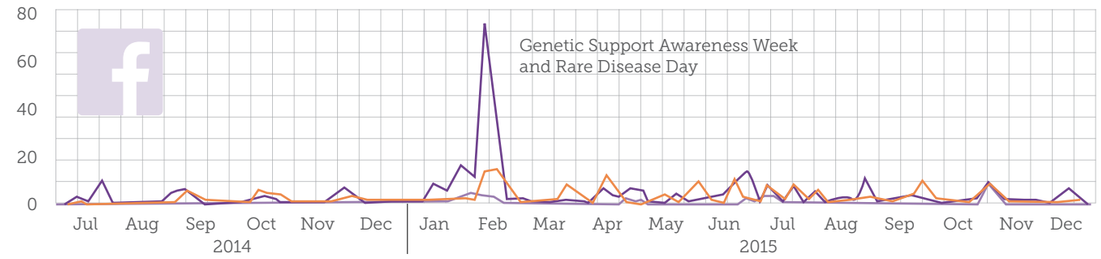


Gender and age of Facebook audience



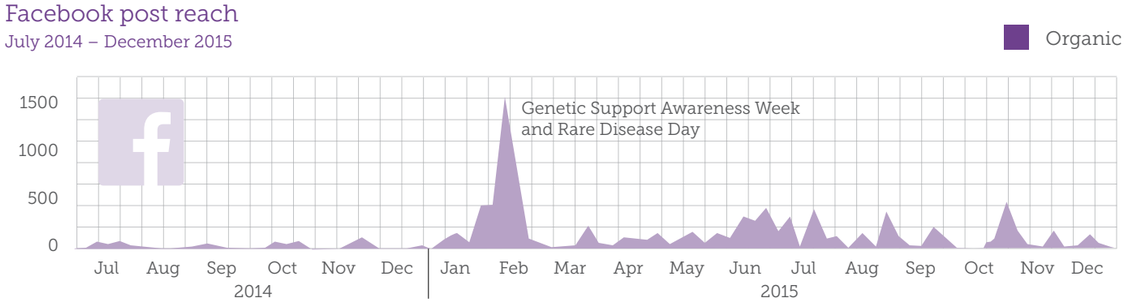
Facebook - Reactions, comments and shares

July 2014 – December 2015



Facebook post reach

July 2014 – December 2015



SNAPSHOT

RARE DISEASE DAY 2015 AND GENETIC SUPPORT AWARENESS WEEK

In the lead up to Rare Disease Day 2015, the GSNV hosted a week long awareness event entitled *Genetic Support Awareness Week (GSAW)*. From the 23 – 27 of February, GSAW aimed at raising awareness about living with a genetic and rare condition and to support people impacted by genetic and rare conditions.

During the countdown to GSAW, the GSNV highlighted achievements in the field of genetics and shared stories from the community. The finale of GSAW was the GSNV Rare Diseases Day celebration on the 27 February 2015. We hosted a presentation morning, which included talks from:

- Kathryn North who gave the key note address
- Dr. Sue White (Melbourne Genomics Health Alliance) who spoke about the experience of doing exome sequencing in the Melbourne Genomics Childhood syndromes flagship
- Heather Renton (Syndromes Without A Name Support Group) who gave a family perspective of living with a rare disease
- Panel discussion and questions from the audience, facilitated by Louisa Di Pietro – Panel: Dr. David Amor, Dr. James Pitt, Ivan Macciocca, Dr. Sue White

The event was followed by a morning tea and opportunity to mingle, with guests – raising their hands in support of Rare Disease Day.

A very successful part of our GSAW program was the GSNV "Facing Forward" campaign, which aimed at giving the genetics and general community a visual insight into the lives of people impacted by genetic and rare conditions and to raise the awareness of the importance of support services in genetic health.

The GSNV invited its members and community to send in their visual interpretations of living with a genetic and rare condition with a short message. We were very pleased to showcase these on the GSNV website and social media. The contributions were stunning and the campaign a great success.

MICHELLE'S STORY:

Having a child with a rare disease is a journey into the unknown. Michelle Karam was lost when the doctors told her they suspected something was wrong with her little girl AJ. After bringing her daughter in for a formal blood test, she was told they thought it might be Maple Syrup Urine Disease, a recessive metabolic disorder leading up to a build-up of amino acids and their toxic by-products in the blood and urine.

"At this point I was completely lost! I had never ever heard of MSUD let alone how my little lady (that we tried many, many years to have) ended up with it. Finally we were taken in for the blood test. My little AJ was two weeks old with nurses jabbing needles in her tiny little arm, and I was trying to understand what was going on (my father had passed away 8 weeks earlier) and the weight of it all got too much to hold. Looking around at all these strangers running around frightened me and I realised that we were in a very serious position.

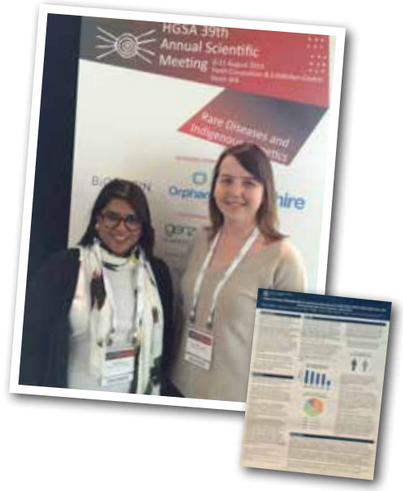
After the nurses got the blood from AJ, I was sent to wait in a cubical for her results. I started praying to my Dad to protect AJ and really hoping that my world was not going to be turned upside down. A few hours later a clan of doctors and dieticians walked in, my heart dropped!

The clan took me into a private room and explained what AJ has and how she got this disease. I was still in a spin as to what this disease was and how it would affect my family. I wanted to know how long she would live for. To my relief they felt my pain and so thoughtfully answered "we're here to help her live a long normal life." As time has passed I understand a hell of a lot more about this very rare disease and how to control her levels thanks to the most amazing team God has put on this earth. Without the support and efficiency of the metabolic department I would have lost my baby girl, but instead I have a very healthy and happy 11 month old."

SNAPSHOT

HGSA 39TH ANNUAL SCIENTIFIC MEETING

By Anna Jarmolowicz



The theme of the Human Genetics Society of Australasia (HGSA) 39th annual scientific meeting was Rare Diseases and Indigenous Genetics, topics which are incredibly important within the genetic community, though often face difficulties with support and management of conditions.

The Genetic Support Coordinator, Keri and I attended the conference held at the Perth Convention and Exhibition Centre, overlooking the Swan River.

Prior to the conference, there was a meeting on Saturday of the Australian Society of Genetic Counsellors Special Interest Group. International guest speaker, Brenda Finucane from the Geisinger Autism & Developmental Medicine Institute in Pennsylvania, America gave an interesting and insightful talk into providing genetic counselling services for women with intellectual disabilities. She spoke about what women may find helpful and how genetic counsellors can better meet these patient's needs. We then heard from Annette Stokes and Christine Jeffries-Stokes about the amazing success of the Western Desert Kidney Health Project. It was great to learn about the project and also be able to take away key messages about how to provide appropriate services for Indigenous Australians.

Notable speakers at the conference included Stephen Groft, an internationally respected figure within the Rare Disease community. He spoke about the growth and development of legislation supporting those with Rare Disease both in the United States and worldwide, and highlighted the import role that patient advocacy groups play in influencing research and also driving policy development. The take away message of his talk was that change will only occur with a global collaborative effort from international Rare Disease communities which include patients, researchers, health professionals and industry.

The GSNV also sees the value in international collaboration and is supporting our Group Leader, Louisa, while she works from Italy this year. It has been a fantastic opportunity for her to meet with and begin discussions about support for Rare Disease with European groups.

The scientific poster session was held on Sunday night and we were pleased to see the finished poster of the recent survey undertaken in collaboration with the Office of Population Health Genomics Western Australia, Genetic Support Network of Victoria, Rare Voice Australia, Genetic Alliance Australia and the Genetic and Rare Disease Network. This research provides key insights into the challenges adults living with rare disease face and will be a strong piece of evidence to provide government to aid future policy making.

Being originally from Perth, I was also able to show my colleagues some of the great sites the city has to offer. We were able to explore Perth city and also the lovely port town of Fremantle. A major highlight of the conference was the Gala dinner at Fraser's in Kings Park. It was a fascinating and enjoyable weekend.

The next HGSA annual scientific meeting will be held Hobart, Tasmania, Australia on 6-9 August 2016. The theme for the conference is 'Integrating Genomics into Healthcare'. The GSNV is actively involved in the local HGSA Victoria/ Tasmania conference organising committee and is working hard to ensure a strong scientific but also support orientated programme. We are also hoping to present original research, led by our research assistant, Victoria. We look forward to seeing you there!

SNAPSHOT

MASTER OF GENETIC COUNSELLING COMMUNITY PLACEMENT

By Elaine Stackpoole

The GSNV offers a diverse range of services for individuals and families affected by genetic conditions and the health professionals involved in their care.

From providing information and support to facilitating access to services, connecting individuals/families with peers and/or support groups, assisting support groups, organising education and training sessions, offering volunteer opportunities and advocating for individuals/families, the GSNV covers a lot of ground.

During my placement I have had the opportunity to become involved with many of these aspects and have learnt about various genetic conditions in the process. More importantly, I have seen these genetic conditions from a different perspective and have observed how the GSNV empowers, supports and connects individuals and families affected by a genetic condition.

The highlight of my placement was attending the Cystic Fibrosis Victoria (CFV) community conference. As a Master of Genetic Counselling student, I have spent a significant amount of time learning about the genetics of cystic fibrosis, but the conference gave me an opportunity to meet people with a personal connection to this condition. I was fortunate enough to talk to parents, siblings, other relatives and individuals living with cystic fibrosis and hear their stories.

The CFV conference really emphasised the power of community and how valuable it is for people to connect with others who understand and can relate to what they're going through. Seeing adults living with cystic fibrosis achieving personally and professionally also offered hope to others whose lives have been touched by cystic fibrosis.

This placement has shown me how important it is to utilise the services of support organisations such as the GSNV and how beneficial peer/support group connection can be for individuals and families affected by genetic conditions. I am thankful that I had the opportunity to observe what goes on behind the scenes at the GSNV and will definitely remember this experience going in to the future.

By Emily Allen

The GSNV has been a great organisation to complete my community placement.

While here I have had the opportunity to observe the running of a support group, attend a number of meetings and information sessions at the hospital and around the city, and have been working on developing a research project. Through my experiences here I have gained an understanding of the importance of providing support to people who have a genetic condition, as well as the ways in which educating people about genetics and genetic conditions can be beneficial.

One of the highlights of my time with the GSNV was an afternoon spent at CF Victoria. It was a wonderful opportunity to learn about how a support group runs and what sort of roles that they take in the community. The team at CF Victoria were very welcoming and were eager to tell me about the great work that they are doing.

One of the projects I have taken on during my time at the GSNV has been to help develop a needs assessment survey for the Klinefelter syndrome community. Through this experience I have gained an understanding of how the research process works, and have had the opportunity to work collaboratively with a variety of professionals during the development stage of this project. I am excited to continue working with the GSNV beyond the time of my placement here to see this project through to completion.

Through my experiences at the GSNV I learned the importance of providing support, education and other resources, as well as how beneficial it can be to connect people together, both on a personal and professional level.

Emily and Elaine

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 do what we do. We thank Prof David Amor for his support of the GSNV staff as operations line manager and providing that all important sounding board to the Group Leader. We wish David well as he moves on to a role with the RCH and welcome Martin Delatycki as VCGS Medical Director. We look forward to working with Martin on mutually beneficial projects.

Goodbye David Amor

Thank you and goodbye to David Amor, Medical Director VCGS, who has provided guidance and leadership to the GSNV staff for a number of years. David has assisted the team by supporting operations and management on a day to day basis, and in this way, helping us to meet our strategic priorities. David has shared the GSNV vision and assisted a collaborative working arrangement with the VCGS/MCRI who remain important stakeholders. Our colocation within the VCGS/MCRI precinct has been an important strength over the years.

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 1 JULY 2014 TO THE FINANCIAL YEAR ENDED 31 DECEMBER 2015.

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

As GSNV Inc. has an infrastructure agreement with the VCGS, accounts for GSNV are audited by KPMG who are the External Auditors for VCGS. They report that the GSNV Inc. concluded the 2014/2015 Financial Year with a surplus of $43,953.77. This surplus combined with the DoH funding means that the GSNV Inc. carries forward a total of $196,953.77 to the current 2016 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

The Financial Sub-Committee (FSC) for 2014/2015 included myself as Treasurer, our President, Kay, and general committee members; Shona, Doreen and Katarina.

MEMBERSHIP STRUCTURE AND FEE SCHEDULE REVIEW

As outlined in the 2013/2014 Annual Financial Report, one of the main goals for the FSC in 2015 was to conduct a review of the membership structure and fee schedule of GSNV Inc.

We would like to reassure members that there will be no changes in service delivery or fees for individual members. We are seeking endorsement in the form of a vote from the GSNV Inc. membership at the current AGM.

The initial step in the evaluation process involved researching the membership structures and fee schedules of other organisations that provided services comparable to the GSNV Inc. One of the points that became clear was that other organisations have membership structures which clearly distinguish between memberships for people directly impacted by a condition, and memberships for professionals such as health-care professionals working in the field.

The FSC recognised the importance of differentiating these membership types for a number of reasons. One of the most important reasons is to keep memberships as affordable as possible for individuals directly impacted by genetic conditions in the face of rising costs of conducting business. Creating a new category for professional members will allow the GSNV Inc. to implement a membership fee which reflects the number of services we provide to professionals while retaining the same low fee for individuals. Professionals, such as genetic counsellors, often receive financial support from their employer to cover membership fees. We have also attempted to make it more attractive to employers to support professional memberships by implementing a fee structure which allows an employer to pay one fee to cover multiple memberships (outlined in Table 1).

A further category has been proposed for large sponsorships. Many not-for-profit organisations have successfully built sponsorship arrangements with a charity, professional body or corporate entity. We think that this category of membership could prove to be of great benefit to the GSNV Inc. going forward, allowing for sponsorships of special events such as Rare Disease Week. These sponsorship arrangements will be carefully evaluated to ensure that the other party is a good fit for the GSNV Inc. vision and mission. Sponsorship arrangements will be undertaken with transparency and with full accountability in place for any sponsorships or partnerships the GSNV Inc. may form.

TABLE 1: Existing and proposed membership structure and fee schedule

GSNV INC. NOW		GSNV INC. PROPOSED	
$22	Individuals	**$22**	Individuals
$11	Student/ Concession	**$11**	Student/Concession
$22	Families & Groups including Professionals and Service Providers	**$60**	Health professionals e.g. genetic counsellors
		$120	Organisation with 2-10 employees
		$220	Organisation with >10 employees (excluding reciprocal memberships)
No other options		**Sponsorships** Case by case basis. Would involve listing on sponsors page of website, printing on newsletter etc. and other privileges depending on arrangement	

CHANGE TO FINANCIAL REPORTING DATES

One of the important changes implemented by the FSC in 2014 was to move our financial reporting from Financial Year to Calendar Year meaning that our end of financial year date changed 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 enabled 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 this 2014/2015 Finance Report will encompass financial reporting from 1 July 2014 to 31 December 2015. At the next AGM in 2017, the finance report will be for the year 2016 only.

Thank you

I would like to thank the members of the FSC and general committee for the time and consideration they have invested in the activities undertaken by the FSC in the 2014/2015 financial year.

Rachel Pope-Couston

genetic support network of victoria

genetic support network of victoria

Murdoch Childrens Research Institute
Flemington Road, Parkville VIC 3052 Australia
P (03) 8341 6315 **F** (03) 8341 6399
E info@gsnv.org.au **W** www.gsnv.org.au

Visit us on Facebook and Twitter