



genetic support network of victoria

*empowering * connecting * supporting*

ANNUAL REPORT 2016



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**genetic
support
network
of victoria**

genetic support network of victoria

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WELCOME TO THE GENETIC SUPPORT NETWORK OF VICTORIA INC. (GSNV INC.) ANNUAL REPORT FOR THE PERIOD 1 JANUARY 2016 TO THE FINANCIAL YEAR ENDED 31 DECEMBER 2016.

As we reflect on the year in review, 2016, we will highlight some of our major achievements, challenges and outcomes; including an overview of governance and financials.

EMPOWERING THE GENETICS COMMUNITY

In 2017, the GSNV will celebrate 20 years of activity - educating, advocating and supporting 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 within a highly sophisticated and expanding genomics environment.

This annual report for the period 1 January – 31 December 2016 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.

In December 2016, the GSNV developed a new strategic plan to take us through the next period of growth and discovery in the genomics health sector. We recognise that only through leadership, collaboration and sustainable practice can we contribute to the development of access to the practical and pathways to the possible.

OUR VISION

Everyone can flourish!

OUR MISSION

We empower people to make decisions about their health and wellbeing.

We collaborate for equity and cultural change.

We provide an interface between people with genetic and rare conditions and health professionals to improve the general patient experience.

THE GSNV IN CONTEXT

The GSNV operates within a changing and challenging environment. Victoria continues to evolve as a world leader in genomic medicine and research, with the deliberate growth and placement of genetic hubs and significant resources being made available to support this leadership strategy.

In 2016, the GSNV has worked to become an integral force in educating and supporting people with genetic conditions, and those who support them. A review of our strategic approach positions us to continue to educate and support for empowerment and equity, and also to advocate for creating and supporting opportunity and possibility.

We continue to meet the information needs of the genetics community – a community that every year 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 genomic technology.

The GSNV will also continue our 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.

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THE YEAR IN REVIEW

IN THE 2016 FINANCIAL YEAR, OUR VISION AND MISSION HAVE LED TO OUTSTANDING ACHIEVEMENTS. HERE'S A SNAPSHOT...



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ABOUT THE GSNV

At all times the GSNV is guided by a set of core values, and the overarching principle that we serve the Victorian genetics and rare disease 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 collaborative 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

We support people with genetic conditions and those impacted by them by assisting them with access to the practical and pathways to the possible.

We do this by:

- Providing timely, accurate and balanced information
- Referring to support groups and other community services
- Referring to clinical services
- Facilitating 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
- A range of projects

WHO DO WE SERVE?

- People who are impacted by a genetic condition
- Support Groups and Stakeholders who represent or support 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 is a statewide service currently physically located within the Murdoch Childrens Research Institute (MCRI). We would like to acknowledge the valued support of Murdoch Childrens Research Institute and Victorian Clinical Genetics Services (VCGS).

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THE GSNV SUPPORTS VULNERABLE PEOPLE IN OUR COMMUNITY

Since its original set up, GSNV has been committed to improving the lived experience of Victorians impacted by a genetic and/or rare condition, and it 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. 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 member; 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 had long term representation on the HGSA Education, Ethics and Social Issues Committee (EESIC). We are members of the 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) and works in support of the GeniOZ project.

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 EuropeRare Connect
- 9

Genetic Alliance UK
- 10

Genetic Alliance US
- 11

ICHG – International Congress Human Genetics
- 12

SIGU – Italian Society of Human Genetics
- 13

GA-SA – Genetic Alliance South Africa
- 14

Dutch Genetic Alliance
- 15

International Genetic Alliance
- 16

GaRDN – Genetic and Rare Disease Network
- 17

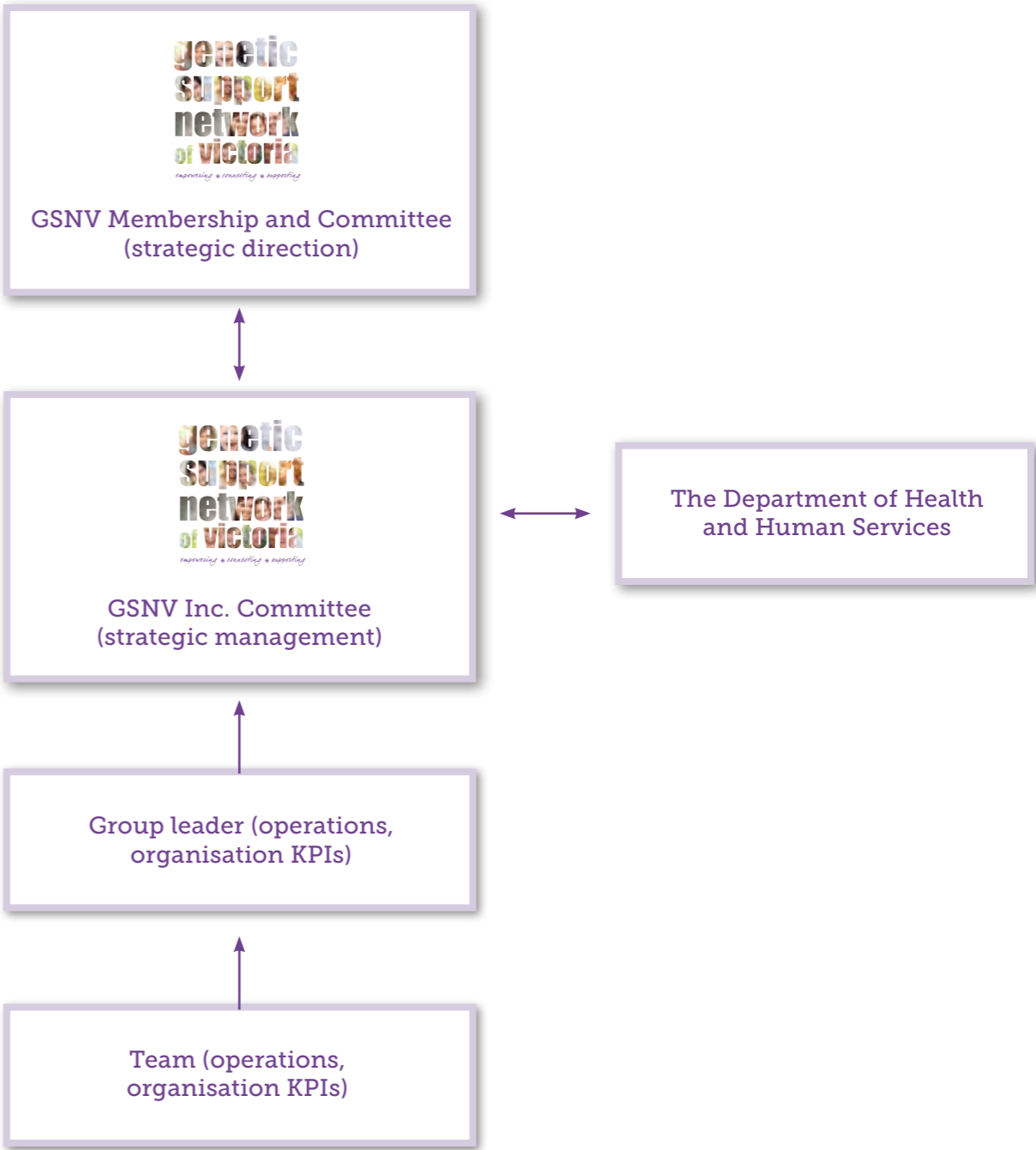
GAA – Genetic Alliance Australia
- 18

AGHA – Australian Genomics Health Alliance
- 19

MGHA – Melbourne Genomics Health Alliance

OUR PEOPLE

GSNV ORGANISATIONAL CHART



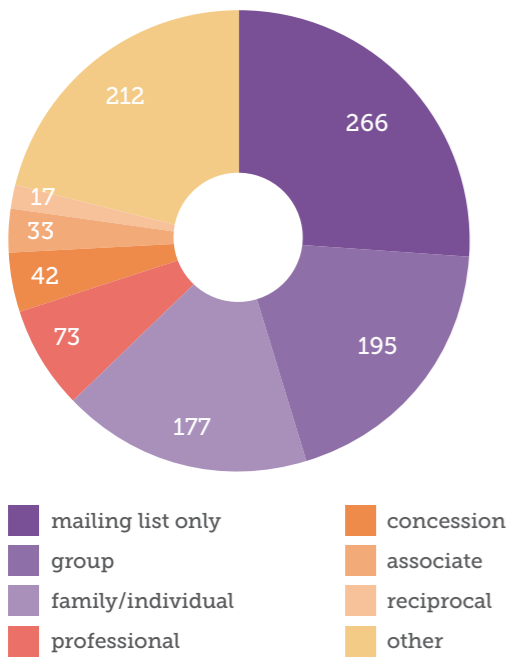
OUR MEMBERS

GSNV financial members make it possible for us to serve our most important stakeholder group – people with genetic conditions. Beyond paid members, we have a network and community that comprises the general, lay, genetics professionals and trainees, rare diseases communities, professional organisations/networks/alliances, biotech/pharmaceutical companies and researchers.

Our strength is our people; the feedback, ideas, and contribution we receive from them is vital, as it enhances and supports our work.

The GSNV would like to say a huge thank you to all members, and we look forward to a continuing positive and engaging relationship.

MEMBERS COMPOSITION 2016



1015 total members

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

GENETIC AND ALLIED HEALTH

- Clinical geneticists
- General practitioners
- Researchers
- Social workers
- Genetic counsellors
- Training health professionals
- Allied healthcare professionals

GOVERNMENT BODIES

- Victorian Department of Health and Human Services (DHHS)
- Public hospitals
- Local councils
- General practice divisions
- Funding bodies

COMMUNITY

- Individuals
- Families
- Carers
- Students
- Community groups
- Maternal and Child 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

Two thousand and sixteen proved to be a year of change at the GSNV at all levels; significant change for our Committee of Management, and within the organisation itself. I am so proud to lead a Committee that responded to these challenges in a positive and resourceful way.

Our year was one of achievement and activity, evaluation, and review, as we took the opportunity to conduct two important projects for the GSNV operations:

1. The Evaluation Project is detailed elsewhere in this report. We took a historical view of where our resources are allocated, how we are viewed by stakeholders, and what expectations are directed at the GSNV. We learnt some new things and had others validated.
2. The second project was a look to the future; a Transition Project to determine where the GSNV needs to be in the future, to remain relevant and active in a changing genetics and genomics environment. We evaluated our past and present activity, and examined what we need to be; how do we need to transition, and to what, to continue to serve our membership and the wider community.

The transition of the GSNV to become even more effective at leading by example, delivering performance excellence, and engaging and connecting people, organisations, and partners, is critical to the long term sustainability of the GSNV. The challenges of the external environment and the internal drivers are clear, non-negotiable and the genomics juggernaut is unstoppable. The GSNV recognises our responsibility to all Victorians to ‘keep up’ and remain relevant. The GSNV has a really strong base from which to build, established over nearly twenty years by people dedicated to serving people with genetic conditions. It’s time for the GSNV to take the next step. Change is upon us.

By implementing change through our new strategic plan, we will ensure:

- That people’s right to flourish is protected, encouraged, serviced, and celebrated; under our watch.
- That the GSNV becomes a peak body for consumer advocacy, education, and support in the genetics/rare diseases and genomics services sector.
- That GSNV funding is protected.
- That the GSNV remains relevant, consolidates, and gathers strength.
- That the GSNV continues to serve members and stakeholders, without compromise in quality.

An exciting year is ahead, and we are ready! Our Committee is strong, and I would like to thank them for their hard work, commitment, and support in 2016. There were many changes to our Committee at the AGM in May 2016, and I would like to also extend my thanks to the outgoing Committee members.

2016 also brought change within the organisation. Our incredibly passionate and capable Group Leader, Louisa Di Pietro stepped down from this position late in 2016, leaving a very big set of shoes to fill! The GSNV team all stepped up, and we have negotiated this period successfully to be even more committed, and connected to our vision. Thank you to all GSNV staff members for your resilience and determination over the past few months.

My personal thank you goes to Louisa for her guidance and support, and the thanks of all the Committee members past and present, for the difference that Louisa has made to many lives over many years. We are all looking forward to future engagement and involvement between Louisa and the GSNV.

Monica Ferrie
GSNV President



Reflections – from the Secretary

The annual report is a wonderful opportunity to stop and reflect on the amazing work done by the team since the last update. Quite a bit has happened since that time, so let's get stuck in!

I am writing this report for the first time as Secretary of the GSNV. I was previously working at the GSNV as Administrative Assistant, while studying a Master of Genetic Counselling. It was a fantastic opportunity then to learn about the needs of people living with genetics conditions, as well as the support and resources available. On finishing my studies I moved into a genetic counselling role, however, I was fortunate to be able to stay a part of the GSNV community as a committee member. This is really important to me, as I can stay informed about initiatives and resources available for the patients and families I see. I also hope I can give back to the GSNV, by providing information about new genetic technologies (such as whole exome sequencing) and research opportunities. I wish to thank the GSNV staff who supported me during this transition – Nancy, Keri and Emily. Also congratulations to Emily, who has recently finished her studies and moved into a genetic counselling role herself!

Navigating my way around the role of Secretary has also been made so much easier by the experience, wisdom, and patience of Monica Ferrie. Monica has made a huge contribution to the GSNV in the past year. She has stepped up and taken on the Group Leader role with great energy and enthusiasm. Under her guidance, the GSNV has continued to provide a much needed and well run service, as well as establishing plans for the future. Monica has been an integral part of the service review, and has been very busy working towards providing a more comprehensive and accessible service in the future. We cannot begin to thank you Monica for your dedication. Also a big thank you to Louisa Di Pietro for all your hard work in shaping the GSNV into the wonderful service it is today, and for your ongoing support into the future.

2016 also brought the review of the GSNV Strategic Plan for 2017-2020. The planning day was full of lively debate and thoughtful consideration by the committee about the future focus of the service. We have created a plan that we feel encapsulates the core values of the GSNV and strives to bring these into the rapidly changing new world of genetic testing and genetic support. With a focus on empowerment and collaboration, we hope to provide a service that strives to ensure everyone in the genetic support community has the opportunity to flourish!

Anna Jarmolowicz
Secretary

Reflections – from the Committee

After my first year on the GSNV Committee, I'm feeling confident that the GSNV's direction is continuing to move forward with a strong presence in the community sector, as well as renewed efforts in the dramatically advancing clinical genetics field.

The GSNV has seen many changes over the past year, including saying goodbye to Louisa Di Pietro as Group Leader. Louisa has been a pivotal part of the GSNV for a number of years, and whilst it was very sad to say goodbye, I look forward to a different working relationship with Louisa, who still has inspiring contributions to make in the GSNV's future.

I want to also acknowledge the tremendous efforts of Monica Ferrie leading the Committee, and stepping into a new role at the GSNV. Monica has done an amazing job of immersing herself in the clinical genetics world, making sure the GSNV has a clear presence and voice, and is abreast of the latest advances in the genetic field.

Overall it has been an exciting time to be a part of the GSNV's evolving network, and an honour to work with the other Committee members over the past year.

Catherine Beard
Committee
Genetic Counsellor, VCCC Familial Cancer Centre



WHO WE ARE

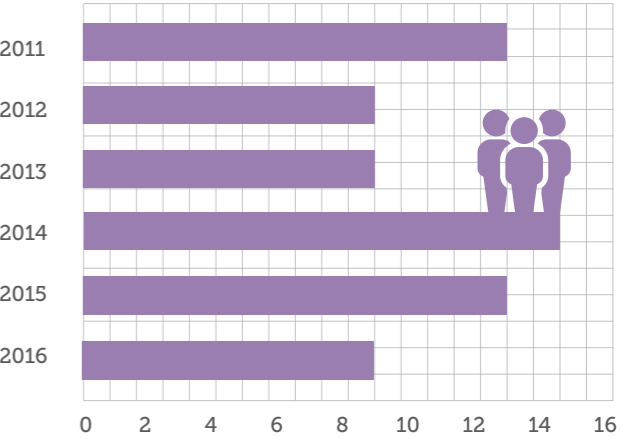
The members of the Executive and Committee who held a position at any time during or since the previous AGM are:

Monica Ferrie	President
Maree Maxfield	Vice President
Rachel Pope-Couston	Treasurer
Anna Jarmolowicz	Secretary
Abbie Kinniburgh	Committee
Christine Williams	Committee
Catherine Beard	Committee
Emily Higgs	Committee
Bill Ellerton	Committee

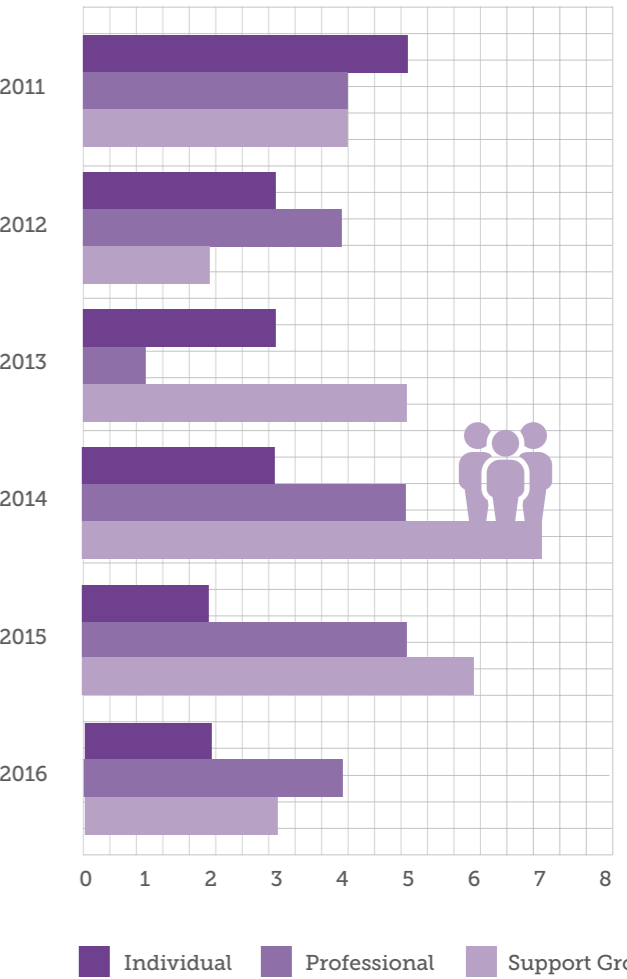
The Committee of the Genetic Support Network of Victoria (GSNV) Inc. operates through an Executive and General Committee with delegated authority and terms of reference (TOR) according to the Associations Reform Act 2012 Model Rules. In line with our essential role in the community and our status as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

COMMITTEE MEMBER GROWTH



COMMITTEE COMPOSITION 2010-2015



CORPORATE GOVERNANCE



THE 2016 PERIOD HAS BEEN DYNAMIC FOR THE GSNV

For most of the reporting period, the Committee of Management has consisted of nine 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). Each meeting, with 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
 - Strengthening support groups and stakeholders
 - Service review and accessibility
 - Stocktake of service and customer needs, and Committee performance
 - Professional development and volunteers
 - Advocacy: influence and reputation
- A Finance Sub-Committee has been operational since 2013-14, and continued to bring increased financial rigor and reporting, risk assessment and management as well as a focus on fundraising through grants.
- We would like to thank Bill Ellerton for his valuable contribution during 2016, Bill resigned his position as a member of the Committee at the end of 2016. We have a strong governance base and look forward to a productive and exciting year in 2017. I cannot wait to report our progress to you in 2018.

STAFF AS AT DECEMBER 2016

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.

Two changes in staffing occurred in 2016. The resignations of our Group Leader, Louisa Di Pietro, and that of our fantastic administrative assistant, Emily Allen. We wish both well in their endeavours and look forward to working with them again in the future.

Monica Ferrie acted in the role of Group Leader up until 31 December 2016. The Committee of Management confirmed this as a permanent part-time arrangement for 2017, in December 2016.

The GSNV operated with a maximum of 2.5 EFT (equivalent full time) with all staff members classified as part-time 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 that managed to positively impact operations. Our staff members often move on to pursue careers in 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 to the 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 and Victoria Rasmussen for your diligence, dedication, commitment and patience and we look forward to working toward great things as an effective team.

Through our commercial arrangement with the VCGS, 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. We say thank you to Martin Delatycki and his team, for their support and assistance in operational matters.

Professional development remains a key focus for all GSNV staff and committee, and in 2016, this focus has provided many positive outcomes.

EFT Allocation as at 31 December 2016			
NAME	POSITION	CLASSIFICATION	EFT
Monica Ferrie	Group Leader	PT	0.4
Keri Pereira	Genetic Support Coordinator	PT	0.5
Emily Allen	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 year have included:

Staff Member	Program
Keri Pereira, Victoria Rasmussen	HGSA 40th Annual Scientific Meeting (Hobart) The theme of the conference was “Integrating genomics into healthcare”, which explored the challenges of introducing genomics testing into the clinical arena.
Keri Pereira, Nancy Amin, Emily Allen	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.
Keri Pereira, Nancy Amin	Equal opportunity and advocacy workshop The course focused on advocating for people with disabilities and the impact of the human rights charter.
Keri Pereira, Nancy Amin	Melbourne Genomics Health Alliance: Demonstrating Success (Melbourne) Presentations from the members of the MGHA which focused on the outcomes of the project and the technologies used to successfully carry out this project.
Nancy Amin	Collective of Self Help Groups Workshop: Getting the word out - Promoting your group Presentations about strategies that can be employed to promote your support group, with brainstorming group work.
Nancy Amin	50 Years of Victorian Newborn Screening: Past, Present and Future Symposium One day symposium organised by VCGS to celebrate the anniversary of Victoria’s Newborn Screening (NBS) program. Invited local and interstate expert NBS speakers reflected on the history, current practice, and future directions. Families who have benefited from NBS also participated in the day and spoke about their experiences.
Keri Pereira, Nancy Amin, Emily Allen, Victoria Rasmussen	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, Nancy Amin, Emily Allen	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.
Louisa Di Pietro	European Society of Human Genetics (ESHG) and European Meeting on the Psychosocial Aspects of Genetics (EMPAG) International Meetings (Barcelona, Spain) Presentations focused on clinical genomic sequencing, sharing data, genetic counsellor training, patient centred care, genetic counselling practice in bilingual communities, and carrier screening.
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.
Victoria Rasmussen	REDCap Introductory Training Course

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

PROFESSIONAL DEVELOPMENT

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

Committee Member	Program
Catherine Beard	Variants In Practice (ViP) research workshop: ‘Polygenic Risk and Genetic Counselling’ The ViP research team presented the findings of their study thus far, including the psychosocial branch of the study, where women who had had a diagnosis of breast cancer were interviewed about their experience of receiving their polygenic breast cancer risk score. The next phase of the study is to disclose polygenic risk scores to further women involved in the study including women who have had breast cancer who have high or low polygenic risk scores, as well as women who have not had breast cancer who have a high or low polygenic risk score. The workshop was designed to help develop our communication skills regarding polygenic risk, as we will be part of the team providing these risk scores to participants. Adapting these new communication skills is pertinent as polygenic risk scores move into the clinical setting.
Catherine Beard	Victorian Statewide Genetic Counsellor meeting: ‘Fetal Exomes: How, When and Why’ This presentation demonstrated insight from three different perspectives regarding the implementation of conducting fetal exomes. The first was from the laboratory, who explained how the technology would work, and what can and can’t be achieved through testing. The second was from a clinical perspective, with case examples highlighting the usefulness of fetal exomes. The final perspective was from an ethical standpoint, and encouraged the audience to consider the ethical arguments for and against introducing the availability of fetal exomes. Overall, I learnt a lot from the presentation, which will help my own clinical practice when fetal exomes are available clinically.
Catherine Beard, Emily Higgs, Anna Jarmolowicz	HGSA Conference Many discussions were had around the potential introduction of the ‘laboratory genetic counsellor’ role, as well as the difficulty in achieving informed consent when using WES as a testing platform. The presentations and open discussions at this conference generated a lot of thought and consideration.
Catherine Beard, Emily Higgs	Familial Cancer Centre Seminar Series, The Royal Melbourne Hospital (RMH) Grand Rounds, Functional Genomics Seminars at Murdoch Childrens Research Institute (MCRI)
Maree Maxfield	Masters of Public Health (The University of Melbourne)
Emily Higgs	Genetic Testing and Insurance seminar A seminar presented to genetic health professionals at the Victorian Comprehensive Cancer Centre. Presented by an insurance broker who is at risk a genetic condition himself. Learned more about factors that influence insurance companies’ assessment of claims, ways to advocate for clients.
Anna Jarmolowicz	Grand Rounds at Royal Children’s Hospital
Anna Jarmolowicz	Consent and Privacy Issues in Health Research: Opt-Out or Opt-In Discussion of informed consent required to use patient tissue & data hosted by the Royal Melbourne Hospital Human Research Ethics Committee (HREC).
Anna Jarmolowicz	Australian Genomics Health Alliance Rare Disease: National Functional Genomics Workshop Leading functional genomics laboratories from around the country met to discuss their disease areas of interest and how this can be implemented into clinical practice.
Anna Jarmolowicz	Practical Genomics - A workshop for genomics in the clinic Comprehensive workshop presented by the Melbourne Genomics Health Alliance and member organisations. Provided an overview on the impact of genomic technologies, variant curation, informed consent and the impact of testing on patients and families as well as prompting ethical debate regarding the role of genetic counsellors in the future.
Rachel Pope-Couston	Joint meeting of the European Society of Human Genetics and the European Meeting on Psychosocial Aspects of Genetics, Barcelona, Spain. This was a 5 day conference bringing together genetic professionals and experts from across Europe and the world. I presented a poster based on a family I had worked with regarding a cancer syndrome, and I presented alongside Louisa Di Pietro who was showcasing some of the great information coming out of the GSNV Evaluation research project. The conference was an incredible opportunity to learn about not only the underlying genetics of conditions but also the psychosocial impact on individuals, families, the community, and on practicing genetic professionals as well. I left the conference with a deeper understanding of many aspects of my role as a genetic counsellor and how I can better assist people impacted by genetic conditions.

INTERNATIONAL MEETINGS

European Society of Human Genetics (ESHG) and European Meeting on the Psychosocial Aspects of Genetics (EMPAG) International Meetings (Barcelona, Spain)

The European Human Genetics Conference is a forum for all professionals in human and medical genetics to review advances and develop research collaborations.

The conference has become one of the premier events in the field of human genetics with over 3,000 delegates, more than 215 oral presentations, 18 workshops, 8 educational sessions, and over 150 exhibiting companies. The ESHG conference is where the latest developments in human genetics are discussed, and where professionals from all parts of human genetics meet. 2016 marked another joint congress with the European Meeting on Psychosocial Aspects of Genetics.

The European Society of Human Genetics promotes research in basic and applied human and medical genetics and facilitates contact between all persons who share these aims. As a seasoned EMPAG/ESHG delegate I am pleased to report that this year was an outstanding meeting and the organisation, ambiance and professionalism in Barcelona notable. Withstanding the practical elements of the conference that were all rated highly, the conference content this year was very much about genomics and the use of new technologies and how they may impact clinical and research outcomes. In the EMPAG conference, which is where I spent most of my time, it was terrific to see the work of the Melbourne Genomics Health Alliance (MGHA) flagship projects profiled on a number of occasions, and received very favourably by the international audience. The work of the MGHA is highly regarded and has placed Victoria (and indeed Australia) squarely in the top ranked genomics hubs around the world.

EMPAG is always interesting as the sessions get to the very heart of genetics and genomics medicine, and the issues we need to be thinking about from a reflective practice and patient point of view. On the first day of the conference (Sat May 21) a joint ESHG and EMPAG session explored genetic privacy and data sharing and got right down to the ethical issues around privacy. 'The hitchhikers guide to data sharing' was an interesting session looking at the era of ubiquitous genetic information for research, clinical care and personal curiosity, but referenced all that against the need to protect the genetic privacy of the data originators. The presentation provided an interesting 'technical map' outlining potential threats to genetic privacy. The presenter took a different approach to the standard privacy versus utility argument and hypothesised trust-enabling techniques designed to create solid partnerships between researchers and participants.



Louisa Di Pietro (GSNV Group Leader) presenting her poster at European Meeting on Psychosocial Aspects of Genetics (EMPAG), Barcelona

On day two (Sun May 22) of EMPAG, the educational session 'Direct to Consumer (DTC) Testing: Empowering patients, caring for consumers' looked at the impact of DTC on genetic healthcare delivery and the shift toward a more empowered patient, taking charge of their own health. The presentation explored the challenges of DTC testing and the mixed reactions and 'concerns' expressed by medical professionals and researchers around DTC testing.

The underlying theme of this presentation was that, it is the public's right to access personal genetic information, when it is delivered in a responsible manner, and this can be supported by health professionals. Key principles of DTC testing delivered by private companies, is that if there is respect for consumer rights and privacy, if reports are written at an appropriate and comprehensible level, if there is educational content to support consumers, using valid test methods, and there is compliance with local regulation and policy, DTC testing and research can support the consumer's ability to comprehend their personal genetic information without evidence of psychological harm.

I think the general opinion of the room was that we still have a long way to go in providing this in the private domain and in general. Physicians are currently not prepared to deal with DTC test results presented by their patients. This lack of preparedness may have a negative impact on the patient - clinician dynamic. I think an evidence-based assessment of this is important and although DTC testing companies are working hard to engage and educate health professionals on DTC testing, it's still a rocky road of acceptance.

The following session on patient 'empowerment' and DTC testing looked at the use of this buzzword to support the idea that consumers should have direct access to genetic risk information without the involvement of health professionals. The session explored three theories on the concept of empowerment in the DTC space (such as empowerment as an expression of individual autonomy) and looked at how we can avoid unduly 'simplistic and binary assessments' of whether or not a technological practice such as DTC testing empowers or disempowers consumers.

The joint ESHG/EMPAG Symposia on 'The Future Lies in Uncertainty' provided excellent discussion on the clinical significance of findings through new genetic technologies, and that often the significance is unknown until much more evidence (bioinformatic or clinical) can be gathered. The phenotype to genotype approach represents a quantitative as well as a qualitative leap in clinical practice. Overall, this has important implications on how consent is obtained, for example with whole genome sequencing.

“The conference has become one of the premier events in the field of human genetics with over 3,000 delegates, more than 215 oral presentations, 18 workshops, 8 educational sessions, and over 150 exhibiting companies.

With new technologies gathering lots more data (often with unknown clinical significance) we can no longer expect that patients are able to consider 'all the possible outcomes' from such testing during the consent process. The session focused therefore on the issue of how much we can tolerate 'uncertainty' in the clinic. The following session followed on with a discussion on personal genomic testing and how individuals understand and respond to genetic risk information for a range of conditions. The main topics addressed public understanding and attitudes regarding testing options, the challenges in communicating test results the psychological effects of genetic risk, and health behaviour changes following testing.

The EMPAG session on day three (Mon May 23) looked at incidental findings and consent, and continued the discussion on genetic testing and incidental findings but focused more specifically on broad consent, consent models and 'pre and post' disclosure attitudes toward the return of test results. In this symposium Ivan Macciocca of VCGS, gave an excellent presentation on the Melbourne Genomics Health Alliance 'shared' clinical exome sequencing consent form which was developed for clinical services and testing laboratories working with the ten member organisations of the Alliance (e.g. VCGS, MCRI, Melbourne Health etc.).

The aim was to establish a simple and standardised consent form that could be used across multiple health facilities and ultimately enable data sharing in an ethical manner. The third presentation in this symposium was excellent and aptly considered professional and family ethics in the era

of unsolicited findings. The session was entitled 'who is my family's keeper' and moved the audience to think seriously about the ethical dilemma of protecting patients' privacy and potentially providing lifesaving information to relatives. During this session the presenter successfully got me thinking about 'who is responsible for conveying genetic risk information to family members?' The current debate on incidental findings draws some attention to the patient as a kind of moral agent co-responsible for their children's or siblings health.

The conclusion drawn in my mind is that there indeed a moral basis for the claim that both patients and health professionals have a duty to warn relatives that are at risk of hereditary diseases, particular where early intervention or clinical intervention is needed. New strategies for genetic disclosure and shared responsibility are indeed needed moving into the genomics future.

Family and patient communication was an important theme throughout EMPAG/ESHG but also very prominent in the satellite conference for the Transnational Alliance for Genetic Counseling (TAGC). Here I had the wonderful opportunity of co-facilitating a TAGC session on 'Facilitating family communication about genetic risk development and the use of novel interventions to assist that process'. The session was conducted by myself, Allison Metcalfe (UK), Diana Scotcher (UK) and Margaret Sahhar (VCGS, Melbourne). We worked toward developing a review of the current state of interventions in promoting family communication. Small group exercises were developed to assist participants to develop techniques to support family communication in their own professional practice.

“EMPAG is always interesting as the sessions get to the very heart of genetics and genomics medicine, and the issues we need to be thinking about from a reflective practice and patient point of view.

The session was concluded with a facilitated discussion of the benefits and the challenges of introducing family communication techniques into genetic counselling curricula, with reference to cultural implications. The basis for the session was founded on the fact that there is sufficient evidence suggesting that many families experience difficulty in talking about an inherited genetic condition that affects one or more of them.

There has been work in the UK, developing interventions in the in psychiatric setting, that has shown promising results in multi-family discussion group settings that could be applied to other scenarios such as talking about genetic risk. Often parents want more support from health professionals about managing inherited genetic risk within the family, and advice about talking to their children. There is however few appropriate opportunities to do so, and genetic counsellors (GC) are uncertain about how involved they should be in helping families to communicate risk information.

Current practice focuses predominantly on the support of the individual affected or at risk, often to the exclusion of the wider family unit. Alison Metcalfe and team in the UK, have conducted the design of an intervention that will assist parents and children in talking about, and coping with the genetic risk affecting their family. Following some initial research and

consultation with senior family therapists, GC leaders, patient group representatives and researchers, it was agreed that a multi-family discussion group (MFDG) intervention might be the most suitable mode in the genetics context. Their work has been outstanding and at this stage they are now working on the further evaluation and testing of the effectiveness and economic viability of their intervention, before it is integrated into genetic counselling practice.

Excluding the stunning location, the tapas, the weather and excellent collegial company in Barcelona, the ESHG/ EMPAG/TAGC conferences were again a highlight for me and perhaps provided a fantastic finale to a year of conferencing, workshopping, networking and learning.

Louisa Di Pietro

experience in the genetic health and support community. The GSNV Volunteer Program.

We currently have 31 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.

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

Some examples of volunteer-support group matches in 2016 are:

Volunteer organisation	Volunteer match	Activity
PEERS (Pediatric Evaluation of Emotions, Relationships and Socialization) study - Royal Children's Hospital	2	Assistance with the research project by visiting schools and administering cognitive measures and data entry.
Klippel-Feil Support Group	1	Assistance with web-page design
The Aussie Hands Foundation	1	Assistance with marketing and social media
Scleroderma Victoria	3	Assistance with mail-outs, designing flyers, and fundraising stalls
Alpha-1 Association of Australia	1	Assistance with fundraising and grants
Australia Alopecia Areata Foundation	2	Assistance with trivia night
Syndromes Without A Name	2	Assistance with Undiagnosed Children's Awareness Day

THE GSNV HAS RECEIVED ENCOURAGING FEEDBACK FROM BOTH PARTIES AND IS CONTINUING TO RECRUIT MORE PARTICIPANTS



I feel privileged to work with AusDoCC and I thank them and the GSNV for affording me this opportunity.

My name is Elicia and I am currently studying a Bachelor of Science, majoring in Genetics. I'm really interested in the ways that recent enhancements in genetic knowledge and technology are being applied to the healthcare arena. I hope to one day enter this fascinating field, so volunteering with the GSNV seemed like the perfect opportunity to gain firsthand insight into various genetic disorders and the support networks that exist to assist people living with these conditions.

I'm currently volunteering with Australian Disorders of the Corpus Callosum (AusDoCC), a support group that assists individuals with complete or partial absence of the corpus callosum (the band of nerve fibres that connects the two brain hemispheres) and their families.

Being a rare condition with different variations, symptoms and clinical presentations, the support group is vital for individuals and their families to meet, make friends and work together towards promoting the disorder so that better diagnoses and treatments can be made. In my position I have been involved with the development of a member database, working on a symptoms list so others can better understand the condition, and assisting with a volunteer grant application.

I will also be helping with preparations for the next conference to be held in May 2017. It is so inspiring to see how passionate and hardworking this group is. I feel privileged to work with AusDoCC and I thank them and the GSNV for affording me this opportunity.

It is so inspiring to see how passionate and hardworking this group is.

– Elicia Pettiroso

To have someone like Emily there ready to help out is a great benefit to us.

– Scleroderma Victoria



Scleroderma Victoria would like to say thank you for connecting us with our volunteer Emily Aulich.

Emily has worked at both our fundraising stalls at the Monash Medical Centre this year, 25 June and 28 November.

Emily arrived early, helped with the setting up, served customers all day, and then stayed to the end to pack up as well. We really appreciate her hard work, enthusiasm and energy, she is just a lovely girl!

We are a very small not-for-profit group. All of the people who help out at the Monash stall are elderly and have Scleroderma, so all have mobility issues. To have someone like Emily there ready to help out is a great benefit to us.

The stall last Monday raised \$621.00 for our small organisation, much of this is due to Emily's bubbly personality behind the stall. She even had to show someone how to wear a scarf, the person ended up buying the scarf!

Emily is going to stay in touch with us and hopefully she is able to assist us again next year.

We are very grateful to your organisation for offering us volunteers. I will contact you again next time we need help."

Congratulations Emily, great work!



STAKEHOLDERS

The GSNV team has increased our focus across all statewide genetic services and the Department of Health and Human Services (DHHS) Victoria to create open, transparent and effective relationships that are mutually beneficial in the achievement of our vision.

We work very hard to secure sufficient and sustainable funding to meet our commercial obligations, and to expand our work from year to year. This is a particular challenge in the coming years, as our work is diverse, and the demand on our services has expanded exponentially. We have taken a project approach to explore funding through diverse channels.

MESSAGE FROM THE GROUP LEADER

Once again I was privileged to lead the GSNV over a productive, reflective, and as is always the case, a challenging year. In 2016, the GSNV was managed under an unusual arrangement whereby I was in Italy, with one eye on the GSNV in Melbourne and one eye on the broader genetics and genomics environments of Europe. This was not without challenge, but in the end, provided an opportunity for us to see, do and approach things a little differently.

The strength of our GSNV Committee, President, Secretary, and staff was duly challenged in my physical absence, but all rose to the occasion, and in so many ways improved their capacity. Many a task and responsibility was taken on in support of my year abroad, and I thank them for their vision and support to me, while doing so. I was so impressed with the initiatives and drive over my year away, and was absolutely assured that the GSNV is a team that works together.

Living and working abroad was a terrific opportunity for me personally. After eight years at the helm of the GSNV, this gave me some space to be reinvigorated and inspired, to learn and to be fulfilled by what I do. While living in Italy, the Italian Society of Human Genetics adopted me as one of their own and sent frequent invitations to attend National meetings. In all, I contributed to three national workshops, four regional workshops and six professional meetings scattered across, Milan, Rome, Bologna and Rimini. Each one drew on my experience and expertise developed over my eight years at the GSNV. I was honoured and privileged to do this at the international level.

Each and every session I participated in was a 'special time' and awarded me with an abundance of new knowledge and many new connections. The European Society of Human Genetics (HGS) and the European Meeting on the Psychosocial Aspects of Genetics (EMPAG) in Barcelona were also incredibly insightful, and as expected, were the absolute highlights of my professional development calendar for 2016.

With a huge focus on genomics and new technologies, many of the sessions looked at whole genome sequencing (WGS) and the benefits of using the technology for the diagnosis of rare and complex conditions. There was much attention awarded to the problem of what to do with incidental findings (which is an ethical issue in WGS) and best practice in consent procedures. It was wonderful to see the Melbourne Genomics Health Alliance feature prominently at EMPAG and to see this Australian initiative become the envy of our European counterparts.



XV111 Italian National Meeting on Clinical Genetics, hosted by the Italian Institute of Genetics Medicine (Istituto di Medicina Genomica) Focus – best practice in clinical diagnosis and addressing the diagnostic odyssey experienced by families and their clinicians.

HERE ARE SOME OF OUR HIGHLIGHTS FROM A PRODUCTIVE YEAR – JUST A SMALL SAMPLE OF THE AMAZING WORK OUR PEOPLE DO EVERY DAY.



GENETIC AND RARE DISEASE SUPPORT

Leading up to Rare Disease Day (RDD) in 2016 the GSNV conducted a number of activities designed to raise awareness amongst professionals and the general community, about Genetic and Rare Disease and the lived experience of the Genetic and Rare Disease community. I am particularly proud of the 'Show us Your Support' – The Face of the Genetic and Rare Disease Community – which followed on from the success of our 2015 Facing Forward campaign.

Our 'Show Us Your Support' campaign called on support groups to show us who they are by way of photo contributions. Our aim was to highlight and celebrate the vital work conducted by support groups in Victoria and across the country. Building a visual tapestry of the faces behind hard working support groups, and those people who dedicate enormous energy in support of others, is a creative and inclusive way of focusing well deserved attention on the sometimes invisible warriors supporting the genetic and rare disease community in Australia.



RARE DISEASE DAY 2016

Dr. Kym Boycott, a Neurogeneticist at the Children's Hospital of Eastern Ontario, Canada (CHEO), and Investigator at the CHEO Research Institute opened our RDD event and gave a snapshot of her research interests into bridging clinical medicine science. Kym is focused very much on the use of next generation sequencing approaches to routine diagnostics and patient care. Her attendance complimented the GSNV focus in 2016, which was very much on genomics and developments in Australia to integrate genomic technologies as a health intervention, into broader health.



2016 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.
Am J Med Genet A. 2016 Aug;170(8):2052-9.
Louisa Di Pietro - co-author

"Survey of healthcare experiences of Australian adults living with rare diseases"
Orphanet Journal of Rare Diseases. 2016. 11 (1): pp. 30-30.
Louisa Di Pietro - co-author

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.

"Very glad I came [to the training], helped with new ideas and reaffirmed what we practice. Great workshop"
- support group leader

GENOMIC HEALTH CARE FOR VICTORIA AND AUSTRALIA

The Department of Health and Human Services conducted a major call out for community feedback into its Genomic Healthcare for Victoria, Discussion Paper. The Discussion Paper invited community input, and discussion to inform a new statewide genetics/genomics framework, and that subsequent service-planning reflects all important community opinions and views. The GSNV encouraged its community to read the Discussion Paper, and provide their input and response to the priority areas and actions explored.

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. Louisa is 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, Louisa 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.

In 2016 the GSNV 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. All stages of the study have been completed and analysed, and we hope to publish the findings in a suitable peer reviewed journal. A poster presentation of the preliminary findings of this research was presented in Barcelona Spain at the Psychosocial Aspects of Genetics (EMPAG) meeting, and at the Human Genetics Society of Australasia (HGSA) Annual Scientific meeting, Hobart.



GSNV EVALUATION PROJECT PRELIMINARY FINDINGS

The findings from service user groups suggested that participants were largely satisfied with the GSNV describing the tailored and flexible help they received from GSNV staff. The Network was often the sole source of support for participants at crucial times. Overall, participants expressed admiration for the Network, which they felt had in many instances gone beyond their expectations when fulfilling support requests. Some participants were dissatisfied with what they felt was unequal representation/support across conditions and one participant had experienced an unfulfilled request.

Health professionals and previous staff members' major criticisms of the GSNV pertained to the inconsistent awareness of the Network within the community/across health services and overall lack of visibility. Professionals also described a lack of knowledge regarding GSNV services and resources, resulting in infrequent referrals to the Network. Professionals and previous staff shared the strong belief that the Network needs to increase its presence, clarify their role and inform professionals of the impact of support. We have begun to address these concerns by promoting the GSNV to the wider community e.g. We described our services in a recent edition of the Vicdoc AMA magazine.

THE GSNV RESPONSE TO THE GENOMIC HEALTHCARE DISCUSSION PAPER

The Genetic Support Network of Victoria's response to the Genomic Healthcare for Victoria discussion paper was prefaced on a consideration of the current health care system and that within that system, significant change, planning, assessment and problem solving is needed in order to successfully integrate genomics into general healthcare and medicine. Our response was articulated from the perspective of both 'genetics' and

'genomics', recognising that each may elucidate differing responses and priority areas for different groups, including health professionals, the general community, patients/families with a genetic condition, and the rare diseases community.

Overall we see that with the introduction of genomics into health care, particularly with a focus on mainstreaming, there could be potential for risk, by way of people being given misleading/incorrect information from direct-to-consumer providers, as well as mainstream health professionals having limited knowledge/understanding of what the test can/can't do, and its interpretation. We maintain that a Victorian strategy looking to mainstream genomics into healthcare, should focus on what's working, what needs changing, and doing no harm in the process.

Increased patient benefits and outcomes are linked closely to key relationships between clinical services and research. Robust, strong and shared relationships between research and clinical services will lead to the delivery of research translation in a timely manner, and deliver greater benefits for patients quicker. From the lab to the clinic and the clinic to the lab, this is a relationship that is central to an enduring strategy that will deliver novel and successful therapies, interventions and solutions.

A multidisciplinary approach to the integration of genomics into health care and the delivery of best practice genetics services is central to any assessment of future models. Collaborations and joint care planning are intrinsic to delivering the very best outcomes possible, considering the 'whole' patient and their ever-changing needs throughout all life and health stages. In many ways, genomics necessitates a 'team effort' and a fundamental agreement that there are many players in the team, with the patient at the centre.

Genomic and genetic medicine of the future entails very important considerations around the collection of reliable and high standard patient data, the curation and management of that data and excellent systems management of the data. Genomics has ushered in an even greater responsibility to properly consent and protect patient data, but before that, the system needs to be very serious about the functions and strategies employed across the whole of life of that data.

In addition to representing our members and community on a Victorian genomics services strategy, in 2016 we continued to be engaged with the Melbourne Genomics Health Alliance, and the Australian Genomics Health Alliance. We also provided a strong consumer voice and contribution to the consultation process regarding a National Health Genomics Policy Framework. With AHMAC ministers and the Federal Government looking seriously at how we can ethically integrate Genomics into the health system. The GSNV remains committed to ensuring that consumer consultation is highly valued, consumers are engaged and are an important component of any implementation plans.



MANAGING CHANGE AND THE FUTURE

As our wonderful GSNV President, Monica Ferrie, has said, ‘the universe has an interesting sense of humour and finds ways to challenge us all, right when we think we are on track’. Very true!

The GSNV has been challenged with my indefinite leave and absence as Group Leader. This has been subsequent to my return from Italy. I will return to the GSNV in a new capacity but have very reluctantly resigned as the Group Leader. Monica Ferrie will continue to act in my capacity and progress the work of the GSNV. I’m so grateful for that.

All thanks to you

None of our triumphs in 2016 would have been possible without the support of the GSNV community and the very people who live with a genetic and/or rare disease in Victoria. I extend a heartfelt thanks to everyone who joined, donated, volunteered, supported and worked for the GSNV in 2016 – because we couldn’t have done any of this without you.

In reflection of 2016 and its happenings, and projecting into a great future, I sign off by saying ‘I am so much looking forward to being part of the next stage of the GSNV’s long, important history’.

Louisa Di Pietro

[Signature]



REVITALISING OUR STRATEGIC DIRECTION

In 2016 the GSNV commenced a major review of our current activities and operations and what our strategic priorities are. We embraced that challenge and began a systematic review of our work, our focus and our purpose to ensure that we continue to serve our members effectively and with outcomes.

Our review has been underpinned by an evaluation project designed to develop a clear understanding of our current activities and operations and the development of a strategic framework for genetics and genomics in Victoria, which will provide direction and priority over the coming period. In 2016 we reviewed our current operations with an eye to the requirements of the future as outlined in the strategy. We intend to play a valuable role in contributing to the Victorian vision where the health of all Victorians is optimised through clinically appropriate integration of genomics into health care. In 2017 the review will be finalised by a member survey, seeking input from our all important community and stakeholders.

SNAPSHOT



empowering * connecting * supporting

An evaluation of the Genetic Support Network of Victoria: A mixed methods participatory approach

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4. The Peter MacCallum Cancer Institute, Melbourne, Australia
5. The Genetic Support Network of Victoria Committee, Melbourne, Australia
6. The Department of Health and Human Services, Victoria, Australia

Background

The Genetic Support Network of Victoria (GSNV) provides an essential service to families affected by genetic and/or rare disease and professionals working in related fields. On average, over 250 individuals utilise GSNV services per year. Recent advances in genomic technology and the rapid expansion in community awareness of genetics have impacted the way individuals engage with their health and support services.¹⁻³ Evaluation research investigating the perspectives of service users and stakeholders is needed to elucidate what form genetics support services should take to be most relevant and viable to individuals in need of specialised support.

Aims

1.To determine the extent to which the GSNV is meeting the needs and expectations of its service users and stakeholders and fulfilling its wider strategic objectives.

2.To explore individuals' attitudes and beliefs about the GSNV, including satisfaction with services provided and ideas for organisational growth and change.

Methods

Approval to recruit participants for this study was granted by the Human Research Ethics Committee (HREC) from the Royal Children's Hospital (Melbourne, Victoria).

Study design: This mixed methods evaluation study investigates participants' experiences and perspectives in three key stages:

Stage 1: Database analysis

Stage 2: Focus groups

Stage 3: Survey

Preliminary results from stage 2 are presented.

Population: Australian adult residents who were proficient in English and had utilised GSNV services within the past three years.

Recruitment: A purposive sampling method was used to recruit eligible participants from the enquiry database into stage 2. Individuals were invited to participate in one of five focus groups with participants from a similar background:

- Group 1: Individuals affected by genetic and/or rare disease and parents/caregivers
- Group 2: Support group organisers, students and volunteers
- Group 3: Past GSNV staff/genetic counsellors (GC)
- Group 4: Genetic health professionals
- Group 5: Stakeholder representatives

Preliminary findings from groups 1-4 are presented.

Data analysis: Adopting a phenomenological approach, focus group discussions were transcribed and coded using thematic analysis to identify major themes. All participants were assigned pseudonyms.

Preliminary Results: Stage 2

Participants

A total of 139 invitations were distributed with 11 returned following relocation. A further 52 individuals actively declined and 37 did not respond. Overall, 30 individuals expressed interest in participation, however, 10 were unavailable at the scheduled time (e.g., work commitments, travel time) resulting in a 16% retention rate (n=20).

Characteristic	Participants (n = 20)
Participant group, n(%)	
Affected individual	4 (20%)
Parent	1 (5%)
Support group organiser	3 (15%)
Student/volunteer	2 (10%)
Previous staff/GC	5 (25%)
Genetics health professional	5 (25%)
Gender, n(%)	
Female	17 (85%)
Male	3 (15%)
Age, mean(range)	40 (24-66)

Qualitative analysis: Major themes

Sociocultural context: Living with a genetic/rare condition in the genomics era

Participants described the impact of technological advances and cultural shifts toward the increased value of psychosocial factors/support on affected individuals' experiences in clinical settings and wider society.

"I think one of the things that may have influenced that is just the prevalence of genetic, the ongoing or the newly sort of new ability to diagnose children at such a young age that they're basically now managing to find a cost effective way of doing testing." (Lauren, support group/parent)

"So I think that the geneticists and the clinicians realise that they need somewhere for these patients to go, they can't just give them a genetic diagnosis and then send them off into the wild on their own which is effectively what's happening now. So for them to encourage support groups makes their job easier as well." (Claire, support group/parent)

Perceived impact of the GSNV: Attitudes and beliefs about the Network

Overall, affected individuals and family members were satisfied with GSNV services and reported an enhanced sense of empowerment associated with GSNV-facilitated connections with supports, health professionals and clinical services and through the provision of information.

Figure 1. Major themes related to the perceived impact of GSNV services on participants

Participants expressed gratitude toward the Network for its role in providing immediate assistance at diagnosis, or in crisis, and for being a source of on-going support:

"In terms of what I asked for like to connect with a support group, they emailed me pretty much an hour later and got me in touch with the support group on Facebook that I use. Basically before that I had nothing, I had no support, no advice, no anything. So it's helped me a lot. And it's still helping me." (Mae, parent)

"GSNV are a bit like our fairy godmother, they really are. They are the people we ring when we say look we've got to have such and such a plan." (Amelie, support group/parent)

Challenges and unmet needs of individuals affected by genetic/rare conditions and their family members

Affected individuals and family members were challenged by experiences of isolation and stigmatisation, lack of understanding, the need to develop new skill sets to manage health and/or facilitate a support group and many experienced a fundamental loss of control.

"I mean we're all new, we come into it new, we know nothing and we've got one life and suddenly we have a whole other life... Suddenly I was controlled by Centrelink, the train system, the education system, the legal system, the welfare system, everybody controls me now, I don't control myself." (Amelie, support group/parent)

Ideas for GSNV growth and organisational re-form

Affected individuals and family members' ideas for GSNV development were related to barriers that they experienced in accessing appropriate healthcare and support including, lack of recognition for condition across institutional settings and the impact of limited knowledge/misinformation on health-related decisions.

"That would be really cool if [GSNV] could network with the health professionals... I go to see a doctor, I tell my doctor my condition they say oh you mean familial polyps? No, I mean hyperplastic. Oh... Ohh I've never heard of that. I know, no one has!" (Ellen, affected individual)

Both affected individuals and health professionals envisioned the GSNV as having a greater role in quality control of information, support groups and other resources. This change was expected to help individuals identify credible and useful sources and feel less overwhelmed.

"And that's a big problem... you don't know if [research findings are] real or they're..." (Jamie, affected individual)

"You know that there's still that thing of, you know, um, it would be great to have a-a resource you know, to know that a resource had gone through some level of scrutiny before we hand it out to our patients." (Hunter, health professional)

Discussion/Conclusion

Overall, affected individuals and family members were satisfied with the GSNV's ability to address their needs and perceived the Network as a source of appropriate and useful assistance. Findings provided additional insight into the impact of advances in genomic and communication technologies on participants' experiences and needs. The challenges faced by participants across interpersonal and institutional settings aligned with supports offered by the GSNV, such as the provision of information to assist individuals to navigate clinical services. Affected individuals and health professionals shared the desire to see the GSNV take on a greater role in quality control of information and support resources. The unknown quality of existing supports was reported to underlie some health professionals' hesitancy to provide referrals. Affected individuals also identified areas for GSNV growth in relation to systemic challenges that they experienced in accessing healthcare and Government funded assistance. In particular, participants expressed the desire to see the GSNV drive cultural change toward the increased recognition of genetic and rare conditions. The expansion of their advocacy role was expected to reduce experiences of isolation and stigma and assist participants in gaining equal access to healthcare, school supports and Government assistance.

Conclusion Overall, the findings suggest that the GSNV is adapting its services to remain relevant and viable within a rapidly developing healthcare context. The completed results will be employed to further enhance the utility of the Network and will provide crucial data to inform the development of genetic support structures.

Acknowledgments

GSNV Staff Members & Volunteers

Louisa Di Pietro Keri Pereira Nancy Amin
Tiffany O'Brien Ellen Pieper

Supervisory Board

The project design was guided by a supervisory board consisting of representatives from key stakeholder groups: The Department of Health and Human Services (Victoria), Victorian Clinical Genetics Services (VCGS), Victorian genetics health professionals and individuals affected by genetic disease. Supervisory board members are acknowledged above as co-authors.

REFERENCES

1. Barker, K. (2008). Electronic support groups, patient-consumers, and medicalization: The case of contested illness. *Journal of Health and Social Behaviour*, 49, 20-36.
2. Grob, R. (2006). Parenting in the genomic age: The 'cursed blessing' of newborn screening. *New Genetics and Society*, 25, 159-170.
3. Timmermans, S. & Buchsinder, M. (2010). Patients-in-waiting: Living between sickness and health in the genomics era. *Journal of Health and Social Behaviour*, 51, 408-423.

FOR MORE INFORMATION ON THESE AND OTHER STUDIES PLEASE VISIT WWW.GSNV.EDU.AU

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

GP Roadshow

Two thousand and sixteen 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 was aimed educating GPs on women’s issues, including genetic related issues, which may present themselves at their clinics. Two roadshows were held in 2016 and one in 2017. These were very successful with great feedback from the GPs that attended. We hope to continue our collaboration with HEAL in the future.

COMMUNITY

Pre-Pregnancy Planning Presentations

This year the GSNV continued to participate in the education of couples prior to pregnancy through the Pre-Pregnancy Planning seminar. These education sessions are run by the Epworth Freemasons Hospital and were held three times over 2016 with between 80-100 attendees at all sessions.

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.

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. In 2016 we collaborated with Cystic Fibrosis Victoria to bring a fresh perspective to our training.

Training is free for GSNV financial members, and upon completion attendees receive a certificate as well as a peer support contract with the Genetic Support Network of Victoria.

STUDENTS

Australian Catholic University



The GSNV was invited to present a guest lecture to the ACU Biomedical Science students. In 2016 the GSNV presented on the impact of genetics in society, and possible career paths in genetics. We discussed with students, 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.

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.

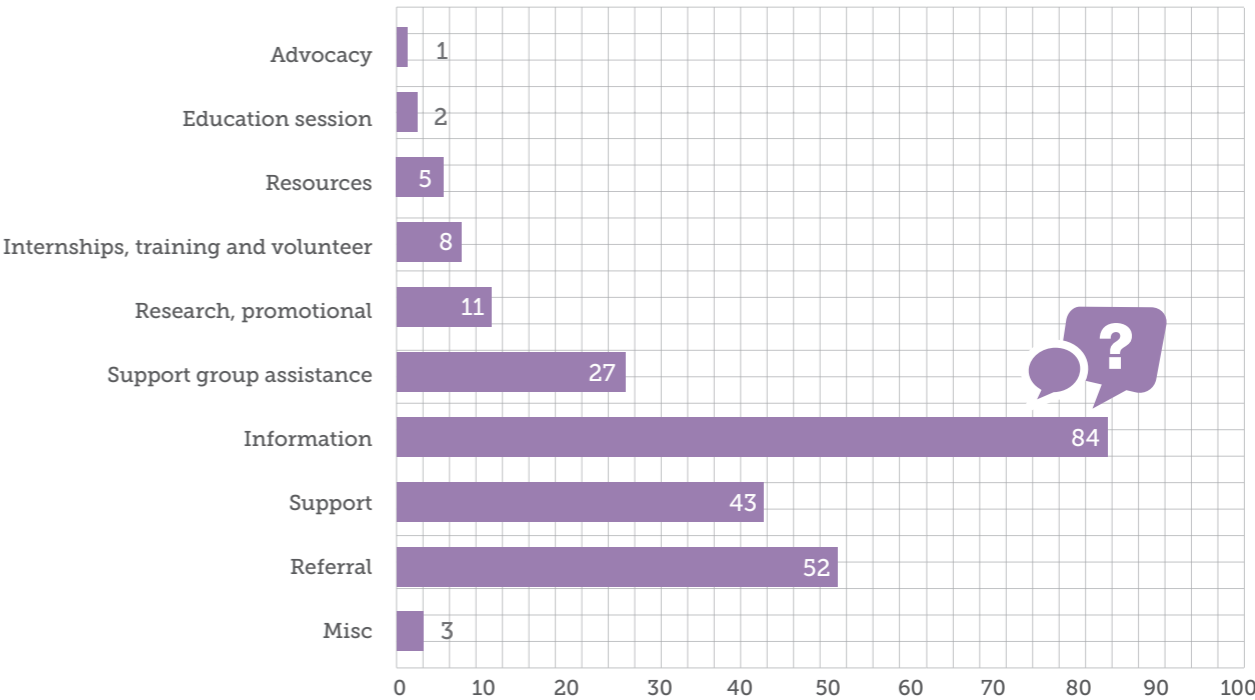
Providing support for support groups is a significant part of our role. We can aid support groups in the following ways:

- 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:

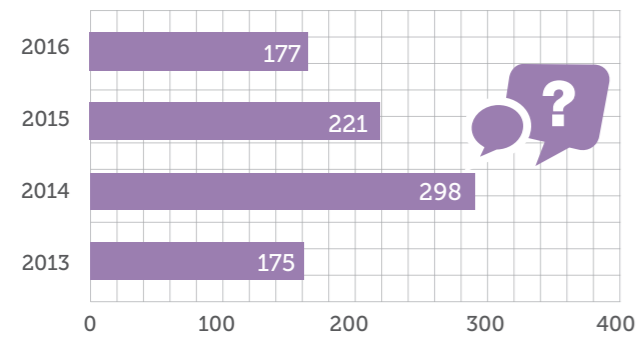
- Nel’s place, support group setup
- Neuromuscular Support Group, seminar
- Usher Kids, information day
- Vascular Anomalies, family day
- Porphyria Association, AGM
- Australian X & Y Spectrum support, family information day
- Syndromes without a name, undiagnosed children’s awareness day

TYPES OF ENQUIRIES RECEIVED

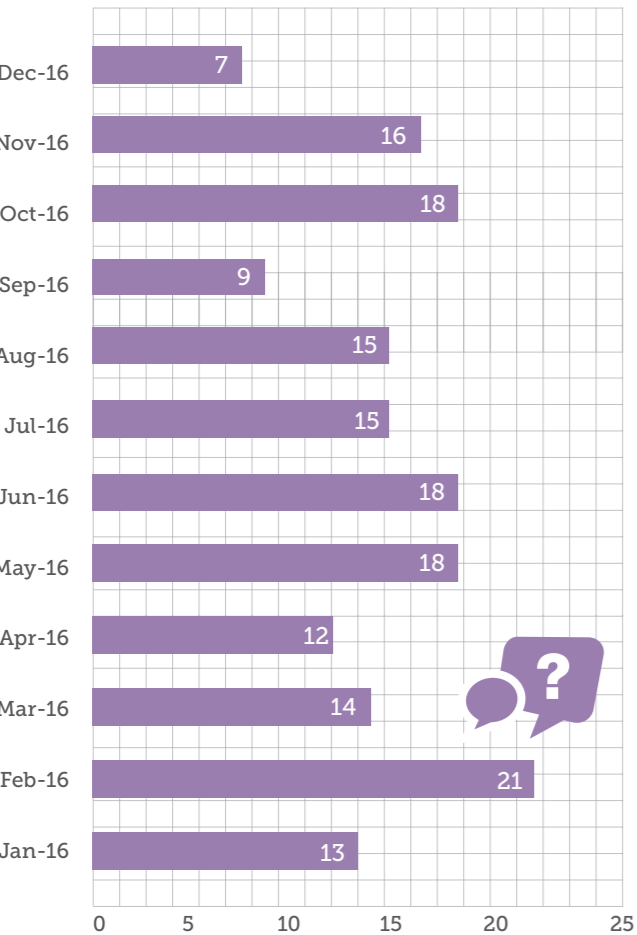


INFORMATION REQUESTS

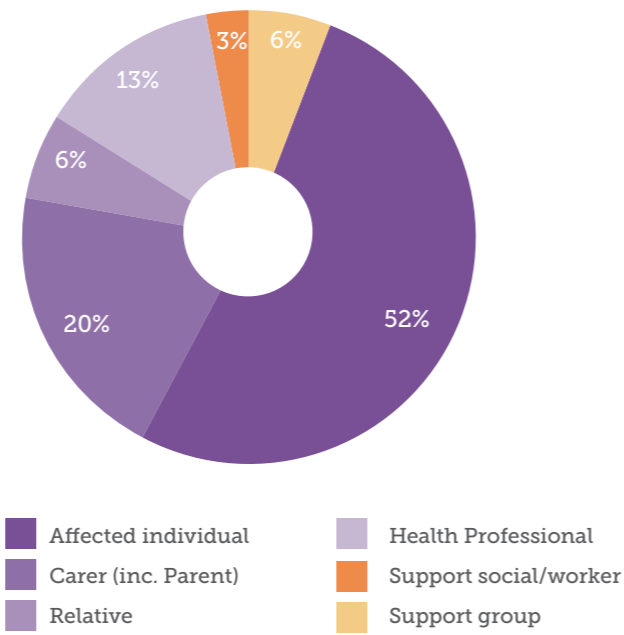
ANNUAL ENQUIRIES



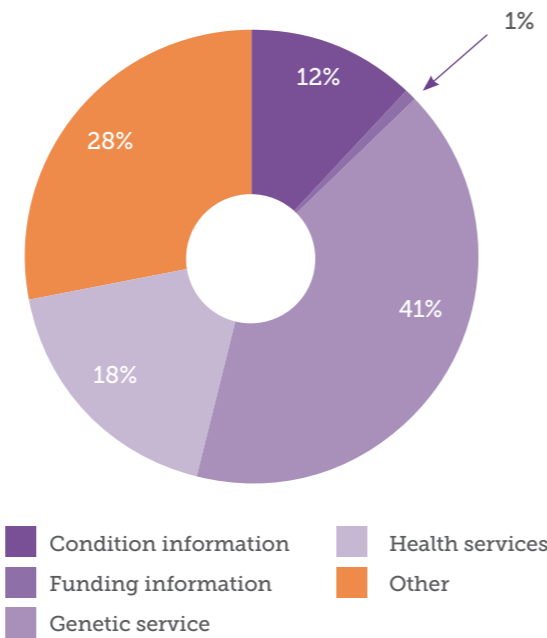
2016 ENQUIRIES BY MONTH



WHO IS SEEKING SUPPORT?



TYPE OF INFORMATION REQUESTED



COMMUNICATIONS

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

- A bi-annual Newsletter
- Monthly 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.

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:

"Thank-you another very packed Newsletter. I always enjoy them & get a lot from the info included"

Yvonne Waite –Parent To Parent Program Co-ordinator

"Visually, the formatting of the entire magazine looks wonderful and the content of the entire publication excellent."

Ally Hensley-The Sisters For Love MRKH Foundation

"This was a very interesting and informative read. Well done everyone. "

Jan Hodgson – Senior Lecturer Department of Paediatrics Melbourne Medical School Royal Children's Hospital

"Thanks for bits and pieces. It is always jampacked full of good stuff. I have shared it with our group."

Maree Maxfield – AusDoCC

"Thanks very much. Great article [about MGHA] and it was great to see Louisa talking about Melbourne Genomics in her editorial too."

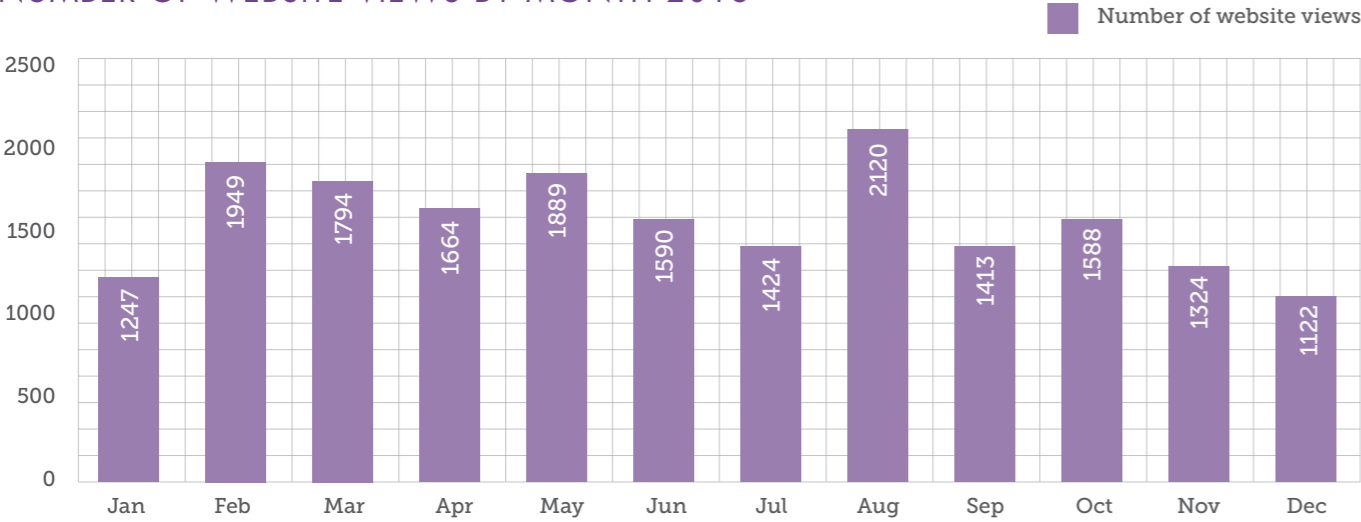
Elly Lynch- Clinical Project Manager Melbourne Genomics Health Alliance

WEBSITE, DATABASE AND SOCIAL MEDIA

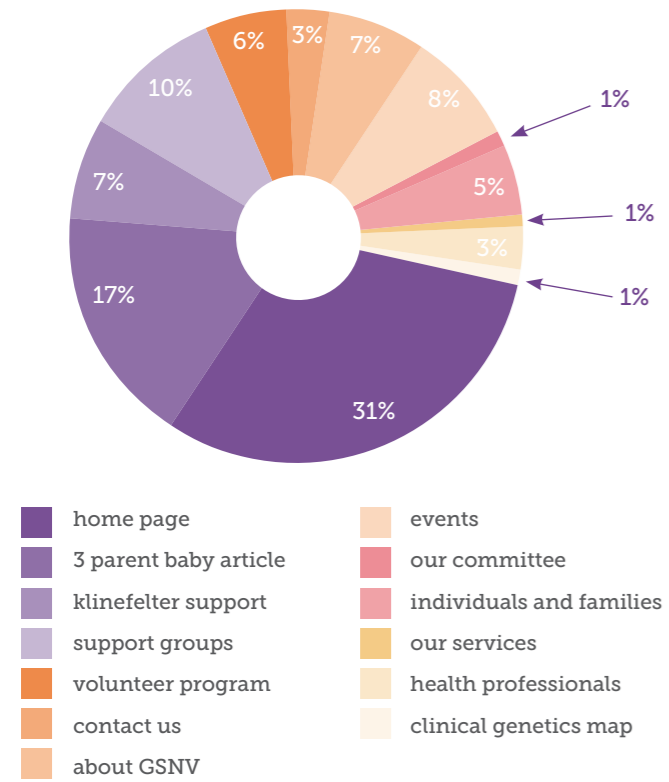
Social media remains an important tool in communicating to our members. In 2016 the GSNV used social media campaigns and blogging to highlight our important events and information. Social media has been identified as an acceptable form of communication both formally and informally and is incorporated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

The GSNV seeks to follow trend 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.

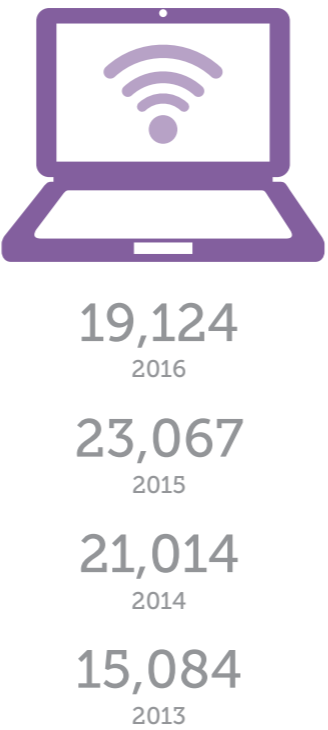
NUMBER OF WEBSITE VIEWS BY MONTH 2016



WEBSITE PAGE VIEWS

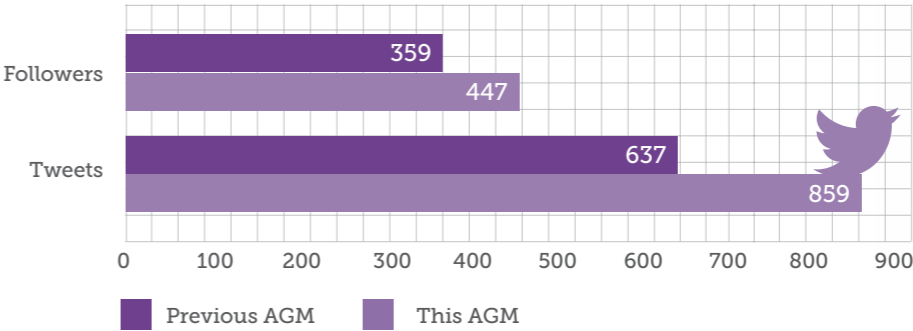


NUMBER OF WEBSITE VIEWS

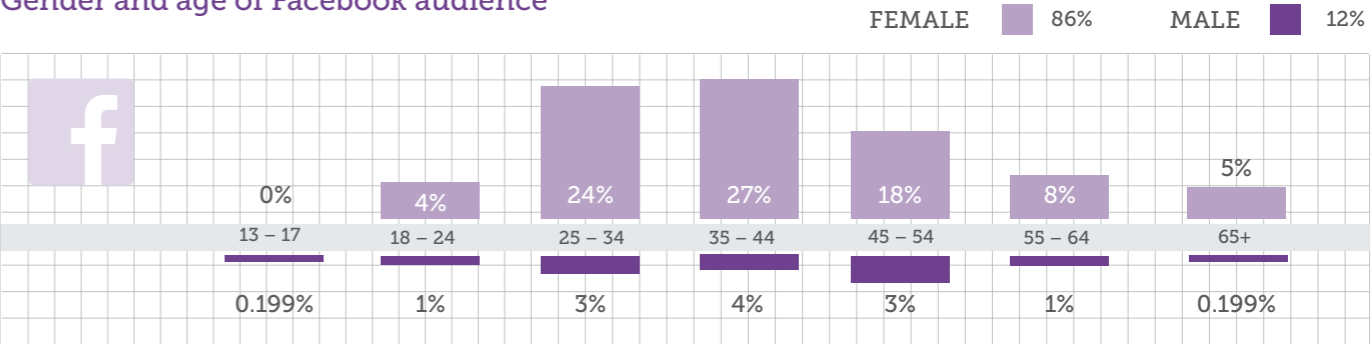


WHO ARE WE REACHING ON FACEBOOK AND TWITTER?

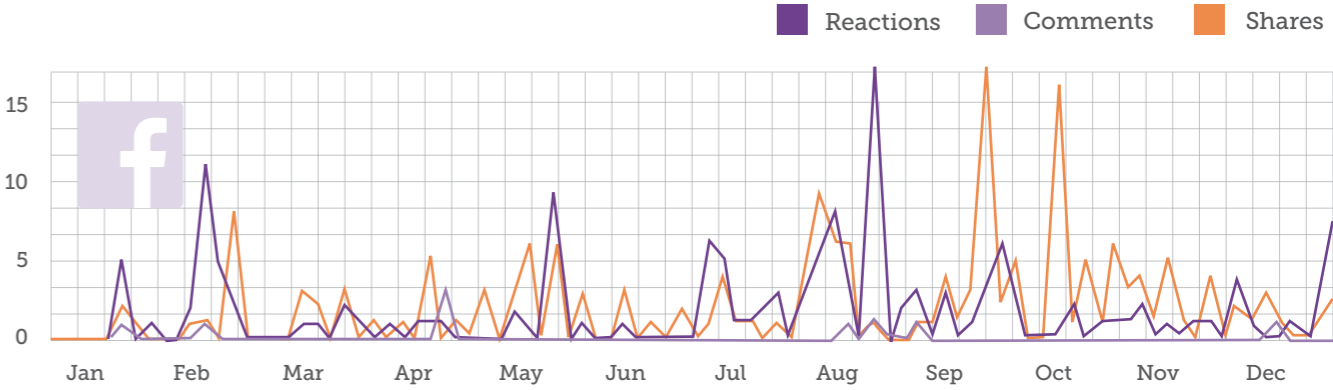
Twitter reach



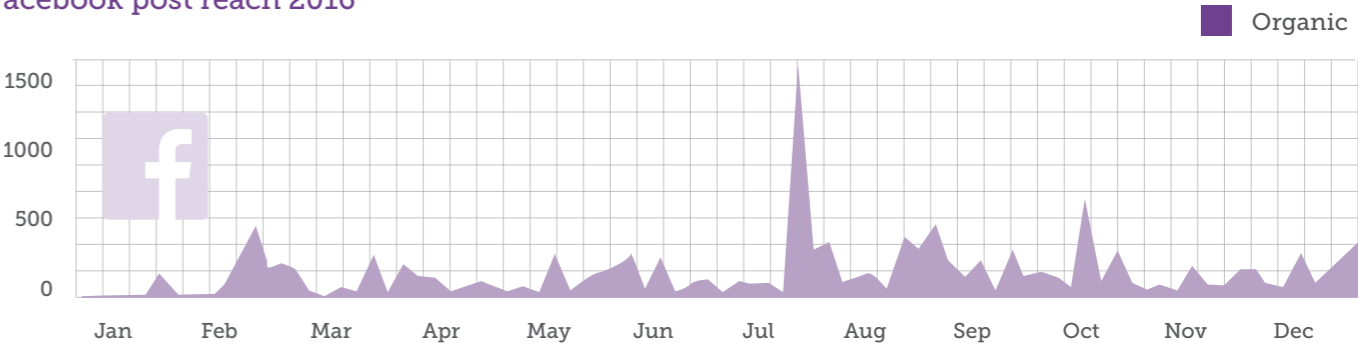
Gender and age of Facebook audience



Facebook - Reactions, comments and shares 2016



Facebook post reach 2016





USHERKIDS INFORMATION DAY By Emily Allen

On September 17, UsherKids Australia held an information day at the Royal Children's Hospital, to mark Usher Syndrome Awareness day. The group held a half day information session followed by a light lunch, which the GSNV attended. Usher syndrome is a genetic condition which can affect vision, hearing, and balance.

The Keynote speaker for the session was Mark Dunning, the chairman for the Usher Syndrome Coalition in the USA, and father to a teenage daughter with Usher syndrome. Mark gave a background to his family's experience with Usher syndrome, as well as information about the condition itself.

Mark talked about the importance of community when faced with a condition like Usher syndrome. He addressed how a strong community, when working together, can come together to support those affected by a common condition, and find ways to progress the research and treatments available to those affected. Mark discussed the important role that Usher families have played in progressing the research done in the area. He gave in depth examples of the ways in which Usher syndrome research has progressed over the years and how it is changing the lives of kids diagnosed today with Usher syndrome. Mark's presentation left the audience with a strong sense of hope for the future of those diagnosed with Usher syndrome.

The next speaker was Jonathan Goerlach, a paratriathlete diagnosed with Usher syndrome type 2. The focus of Jonathan's speech was on the importance of community. He spoke about the difference in the amount of support that is currently available to those with Usher syndrome, because of the community that they have built, as compared to what was available when he was a child growing up with Usher syndrome. He talked about the isolation that he experienced before being joined in with others who were similarly affected, and the positive impact that that support has had on his life. Jonathan talked about the importance of sport throughout his life and how he has incorporated it into his life with Usher syndrome.

Next, Kate Morell, a graphic designer and writer diagnosed with Usher syndrome, shared her story with the audience. Kate presented a beautifully written piece about her experience

growing up with Usher syndrome, and its effect on her sense of self. She spoke about how being connected with the Usher syndrome community had provided her with a sense of belonging and helped to turn her life around. Kate's moving and deeply personal reflection provided the audience with an insight into her life with Usher syndrome and again emphasised the important impact that a support community can have on individuals.

The final two speakers spoke from a more medical and scientific standpoint. First, Lisa Kearns, a research orthoptist and genetic counsellor provided an update on the research in the area of genetic vision loss. She talked about ways in which the vision loss portion of Usher syndrome, cause by retinitis pigmentosa, can be treated even though it cannot yet be cured. Lisa touched on the value genetic information can have for a family with a rare disease.

Finally, Dr Rachel Burt spoke about a study currently being undertaken with the Molecular hearing laboratory and the Melbourne Genomics Health Alliance called the Deafness Flagship Project which aims to search for a genetic cause of hearing loss in every infant identified through the Victorian Infant Hearing Screening Program. She spoke about the different causes of deafness and hearing loss, and the different ways that these can be tested for, with an emphasis on the value of having a genetic diagnosis identified for hearing loss.

This information day presented a variety of different speakers, who had each come from different backgrounds and were speaking for different reasons. Despite these differences, every speaker touched on the importance of support for those living with a rare condition, which highlighted the importance of the work of UsherKids Australia and other support groups.

For more information on the UsherKids Australia information day, you can visit their website at www.usherkidsaustralia.com.

SNAPSHOT

HGSA WRAP-UP

By Keri Pereira and Emily Allen

This year's annual Human Genetics Society of Australasia (HGSA) conference was held in Hobart in August. Although it was the middle of winter, Hobart brought some wonderful chilly but sunny days for the conference attendees. The theme for this year conference was the very apt 'integrating genomics into healthcare'.

The conference started off with the Special Interest Groups (SIG) meetings. The GSNV attended the genetic counselling SIG where Assistant Professor Allison Werner-Lin presented her research looking at prenatal genomic testing. Allison's background in family counselling and psychotherapy gave her a strong client centred approach to her analysis of the data. She shared with the conference her insights into the effect of variants of unknown significance and the pregnancy and neonatal experience of both mothers and fathers. These insights included the benefits of talking about emotional content openly and directly in counselling sessions, and the importance of partners attending prenatal appointments. She kept the audience engaged with her interesting findings and charismatic presentation style.

From there, the genetic counsellors presented their interesting cases, with presentation ranging from, communication of genetics to people with an intellectual disability, to difficulty accessing services in regional areas. Following this there was a panel discussion looking at genomic genetic counselling and the role of the genetic counsellor. This caused a lively debate about how the role of the genetic counsellor might diversify with the introduction of genomic testing to the clinical arena.

The rest of the weekend involved a combination of SIG meetings and general sessions. The genetic counselling SIG on the second day of the conference involved presentations ranging from discussions of the use of mindfulness in genetic counselling practice, to the implementation of new genetic technologies into clinics.

During the afternoon of day two, the Sutherland Lecture was given by Professor Ravi Savarirayan on the subject of the world of medicine in the time of genomics. Ravi engaged the audience with a history of genetics in medicine, shown through the lens of his own work with families with short stature and bone disorders. Ravi's presentation was both entertaining and engaging.

Day three of the conference began with a session themed on the social and consumer impact of genomics. This session concluded with a beautiful address from Margaret Sahhar, reflecting back on her career and the development of the Master of Genetic Counselling program at the University of Melbourne. This year marked Margaret's retirement from her roles at the Royal Children's Hospital and University of Melbourne, and we wish her all the best in her retirement.

On the final day of the conference, Professor Agnes Bankier give the HGSA Oration, a fascinating reflection back on her career with the Victorian Clinical Genetics Service (VCGS). Agnes was once the director of the VCGS, and now sits of the Human Research Ethics Committee at the Royal Children's Hospital.

This year's HGSA conference presented a number of interesting topics all relating in some way to the overarching theme of genomics in healthcare, and provided insight into the future direction of genetics and genomics in both the healthcare setting, as well as in society as a whole.



SNAPSHOT

RARE DISEASE DAY 2016 – THE PATIENT VOICE

By Keri Pereira

Rare Disease Day (RDD) was held on 29 February 2016 (a rare day!). RDD aims to raise awareness of the impact of living with a rare disease on individuals and families.

This day also aims to highlight some of the challenges and achievements that happen along their journey. It's an international day of celebration which recognises the experiences, triumphs, difficulties and the human faces of the rare diseases community worldwide. This year's slogan was "Join us in making the voice of rare diseases heard", which emphasised the patient voice and the importance of recognising the patient's experience. At the GSNV we use this day to bring together researchers, clinicians, and the community; and share information between these groups. This year we held a seminar with presentations from health professionals, researchers and support group leaders.

The GSNV was very lucky to have international researcher Dr. Kym Boycott presenting the work she has been conducting on rare diseases. Kym is a medical geneticist at the Children's Hospital of Eastern Ontario (CHEO) and clinician scientist at the CHEO Research Institute. She was Lead Investigator of the Genome Canada and CIHR funded 'Finding of Rare Disease Genes in Canada' (FORGE Canada) project, which investigated the molecular aetiology of rare paediatric diseases. She 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.

As her work suggests, Kym is highly interested in ensuring research into rare disease continues. As the Chair of the Diagnostics Committee of the International Rare Disease Research Consortium Kym stressed the importance of international collaboration as when working together, we are

more likely to be successful in gaining a better understanding of rare diseases.

Professor Kathryn North, the Director of MCRI and Associate Professor Tiong Tan, Clinical Geneticist at VCGS both gave fantastic presentations on the institute's perspective on rare disease and the clinical and research initiatives that are under way. The MCRI is taking a leading role in progressing research and clinical management for people with rare disease through projects such as the Melbourne Genomic Health Alliance (MGHA) and the Australian Genomics Health Alliance.

We were extremely happy to have the founders of UsherKids Australia presenting as they drove home the theme of "the patient voice". They spoke eloquently about their children's diagnosis of Usher syndrome and their journey to starting the newly formed support group. The relationships that they have built with clinicians and researchers has seen them become part of the Deafness flagship with the MGHA, which is a big step forward for children diagnosed with hearing loss.

The overarching theme which came out of the day was the importance of collaboration between clinicians, research and the community, not only nationally but internationally as well. The sharing of knowledge and resources will only benefit the rare disease and medical community.

The GSNV would like to thank MCRI, the presenters, and all that attended the informative day. We would also like to acknowledge that Kym's visit to Australia was supported by the Royal College of Pathologists of Australasia and the GSNV would like to thank them for allowing Kym to speak at this event.



SNAPSHOT

ADOPTION AND GENETICS:
ADOPTEE AND GENETIC HEALTH PROFESSIONAL PERSPECTIVES

By Rhiana Spinoso and Tiffany O'Brien



My name is Rhiana Spinoso. I completed a BSc degree with a major in genetics and a diploma in modern languages in 2015. I am currently enrolled as student in the MSc Genetic Counselling degree at The University of Melbourne. I volunteer with and am a current member of the committee of management of Syndromes Without A Name (SWAN).

My name is Tiffany O'Brien. I have completed a BSc in Biomedicine and a GradDip in Genetics. I am currently working at Monash Familial Cancer Centre as an intake assistant. I have recently completed my first year as a student in the MSc Genetic Counselling program through the University of Melbourne.



Between 1945 and 1975, over 45,000 adoptions occurred in Victoria alone. During this time, closed adoption, where the family records of the adoptee were sealed, was standard practice. As a result of this practice many adopted individuals today still have limited or no knowledge of their family history. Changes in legislation which came into effect in the 1980's-1990's meant that adoptees born after this period had access to their birth parent's names. However, despite this many of these adoptees still struggle to gain sufficient information on their family health history. Adoptees born overseas often still have no access to information about their birth parents.

The implications of this lack of knowledge regarding family health history in a genetic setting remains largely unknown. There is limited research to date into the difficulties faced by adoptees when navigating the health system, with an unknown family history. The perspectives of adoptees regarding genetics, what they know and what they want to know, has also not been explored. There is also limited research into ways in which genetic health professionals manage restricted knowledge of family history in assessing genetic risk.

he GSNV were contacted by a member of Adoption Origins Victoria, and adoption support group, to develop a fact sheet for adoptees about genetics and accessing family history information. This prompted the GSNV to propose a number of research projects around adoption and genetics to the Master of Genetic Counselling students. From this two research projects were developed, looking at adoption, genetics and family health history from two different perspectives.

The first project will aim to understand the perspectives of adoptees regarding their interest and exploration of genetics, and family health history through semi-structured interviews. The second project will use a short survey followed by semi-structured interviews to explore the experiences of genetic health professionals with adopted clients.

We hope that through this research we will be able to develop a better understanding of issues faced by adopted individuals in terms of genetics and family health history. We hope that the findings from both projects may be used to inform genetic health professional practice in assisting adopted clients, as well as clients with limited knowledge of their family history.

Recruitment for these projects begun in March 2017.

SNAPSHOT

THE NEUROMUSCULAR SUPPORT GROUP FOR YOUNG ADULTS SECOND INFORMAL MEETING

By Hannah Pennington

The Second Neuromuscular Support Group for Young Adults was held at VicHealth, Carlton on Thursday 28 January, 2016.

The evening began with an introduction from me, Hannah Pennington – welcoming my guests and thanking them for attending. My genetic counsellor, Adrienne Sexton (a genetic counsellor from the Royal Melbourne Hospital) introduced our first speaker, Professor Ingrid Winship (a Professor of Genetics and Executive Director of Research with Melbourne Health) who gave us an insightful talk on the services and testing options that are offered within the Royal Melbourne Hospital for young adults who suffer from neuromuscular conditions. She also outlined the obvious gap in medical services for those particular individuals.

Our second speaker for the evening was the vivacious Karni Liddell. Karni was flown in from Brisbane to attend our meeting, as she and I have both been unable to attain a conclusive diagnosis for our neuromuscular conditions. Karni spoke about her personal experience with spinal muscular atrophy, and her struggles as a child and young adolescent to adult with the condition. Karni is also a former Paralympic swimming competitor, winning a bronze medal at both the 1996 Atlanta Games and the 2000 Sydney Games.

Next to speak was my friend, Fin Kelly. I met Fin through Adrienne Sexton at the Royal Melbourne Hospital in 2014. Adrienne put us in contact after she suggested that I chat with other individuals with similar symptoms and age to myself. Fin suffers from ARSACS (Autosomal recessive spastic ataxia of Charlevoix-Saguenay) and has been experiencing symptoms for his entire life; they include difficulty coordinating movements and muscle wastage, which we have in common. Fin wrote a poem to express his feelings in regards to his condition; it was incredibly beautiful and moving. Our final speaker for the evening was Emma Bradhurst. Emma and I met during the first Neuromuscular Support Group for Young Adults in 2015. Emma was put in touch with the support group as she also sees a geneticist at the Royal

Melbourne Hospital as she suffers from Cerebral Ataxia. Emma's condition affects her gait and muscle coordination, which are also symptoms that I share.

Emma spoke about her diagnosis as an adult, and how it was unknown to her throughout her childhood and early stages of adulthood, that she has a neurological condition. She also spoke very passionately about her belief in her faith, which assists her in the daily struggles of her disorder. The purpose of this particular meeting, previous and (hopefully) future meetings are to promote the importance of supporting one another through our difficulties as young adults living with neurological conditions. It is my hope that we continue to strive to ensure others feel supported and cared for, regardless of their disability.



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 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 and Human Services (DHHS) Victoria who continue to provide recurring funding for our important work. We thank in particular Dr Paul Fennessy and Margaret Howard (DHHS) for an ongoing supportive relationship with the GSNV and for their wisdom year in, year out.

We are thankful for the professional relationships including support and advice from all our stakeholders including people with genetic conditions, clinical services, universities, support groups and so many others. We thank Prof Martin Delatycki for his support of the GSNV staff as operations line manager, and providing that all important sounding board to the Group Leader.



FINANCIAL REPORT

ON BEHALF OF THE EXECUTIVE AND COMMITTEE OF THE GSNV INC. WE ARE PLEASED TO REPORT ON THE FINANCIAL DETAILS FOR THE FINANCIAL YEAR 1 JANUARY 2016 – 31 DECEMBER 2016.

We have continued support from the Department of Health and Human Services (DHHS) 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.

I am pleased to report that the GSNV has been awarded an ongoing increase in funding from the DHHS of \$50,000 to assist us to achieve our outcomes. 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 2016 Financial Year with a surplus of \$7,159.15. This surplus is carried forward to the current 2017 Financial Year.

FINANCIAL SUB-COMMITTEE

The Financial Sub-Committee (FSC) for 2016 went through several changes, ending the 2016 financial year determined to build a strong and effective sub-committee for 2017. As Treasurer and Convener of the FSC I would like to thank Kay Timmins, Shona Malberg, Doreen Floyd, Katarina Radonic and Bill Ellerton for their service at various points across the financial year.

In 2017 we are planning to concentrate on creating and guiding fundraising projects that can be completed by the GSNV Inc. staff and volunteers.

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 was approved by members and Consumer Affairs Victoria. This means that this 2016 Finance Report will be the first year to be a complete calendar year and will encompass financial reporting from 1 January 2016 to 31 December 2016.

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 2016 financial year.


Rachel Pope-Couston

STATEMENT OF INCOME & EXPENSES

STATEMENT OF INCOME & EXPENSES FOR THE MONTH ENDED 31 DECEMBER 2016
70680 GENETIC SUPPORT NETWORK (V)

	Actual 2016		Budget 2016	
	For this Period	Year to Date	Year to Date	Year to 31/12/2016
BROUGHT FORWARD BALANCE @ 1/01/2016		43,953.77		0.00
INCOME				
DHS GRANT	12,750.00	178,000.00	202,999.96	178,000.00
NHMRC CONSULTING	0.00	0.00	0.00	0.00
LORD MAYOR'S TRUST	0.00	0.00	0.00	0.00
MEMBERSHIP FEES	0.00	0.00	0.00	0.00
DONATIONS	0.00	3,023.00	0.00	0.00
CONFERENCE FEES	0.00	0.00	0.00	0.00
SUNDRY INCOME	20.00	3,841.00	2,000.04	2,000.00
Total Income	12,770.00	184,864.00	205,000.00	180,000.00
EXPENDITURE				
SALARIES & RELATED COSTS	7,763.38	154,280.59	158,000.00	158,000.00
COMPUTER HARWARE	0.00	1,986.82	2,500.00	2,500.00
COMPUTER SOFTWARE & EXPENSES	0.00	253.85	500.00	500.00
POSTAL SERVICES	0.00	414.19	1,500.00	1,500.00
PRINTING, STATIONERY & PHOTOCOPYING	1,631.39	6,383.86	15,000.00	15,000.00
BOOKS & SUBSCRIPTIONS	0.00	807.28	1,000.00	1,000.00
TELEPHONE CALLS	0.00	0.00	700.00	700.00
SMALL GRANTS	0.00	1,800.00	2,500.00	2,500.00
SPECIAL FUNCTIONS – OTHER	0.00	1,247.65	3,800.00	3,800.00
STAFF TRAINING & CONFERENCES	0.00	2,662.53	2,000.00	2,000.00
TRAVEL	0.00	3,298.33	4,000.00	4,000.00
CORP SERVICES – IT/HR/FIN	2,500.00	30,000.00	30,000.00	30,000.00
OTHER ADMINISTRATIVE COSTS	13,500.00	18,523.52	5,000.00	5,000.00
Total Expenditure	25,394.77	221,658.62	226,500.00	226,500.00
OPERATING SURPLUS/(DEFICIT)		7,159.15		-46,500.00
CARRIED FORWARD @ 31/12/2016				

AUDIT STATEMENT



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Murdoch Childrens Research Institute
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W vcgs.org.au ABN 51 007 032 760


Audit Statement

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

The accounts of the VCGS are audited annually by KPMG who are our External Auditors. The accounts for 2016 have been audited by KPMG in accordance with this practice. In the Calendar Year 2016 GSNV recorded a surplus of \$7,159.15 which will be carried forward to the current 2017 Calendar year.

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

Best Wishes,



Maria Telford BA, CPA
Financial Accountant
Murdoch Childrens Research Institute/
Victorian Clinical Genetics Service.

**genetic
support
network
of victoria**

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