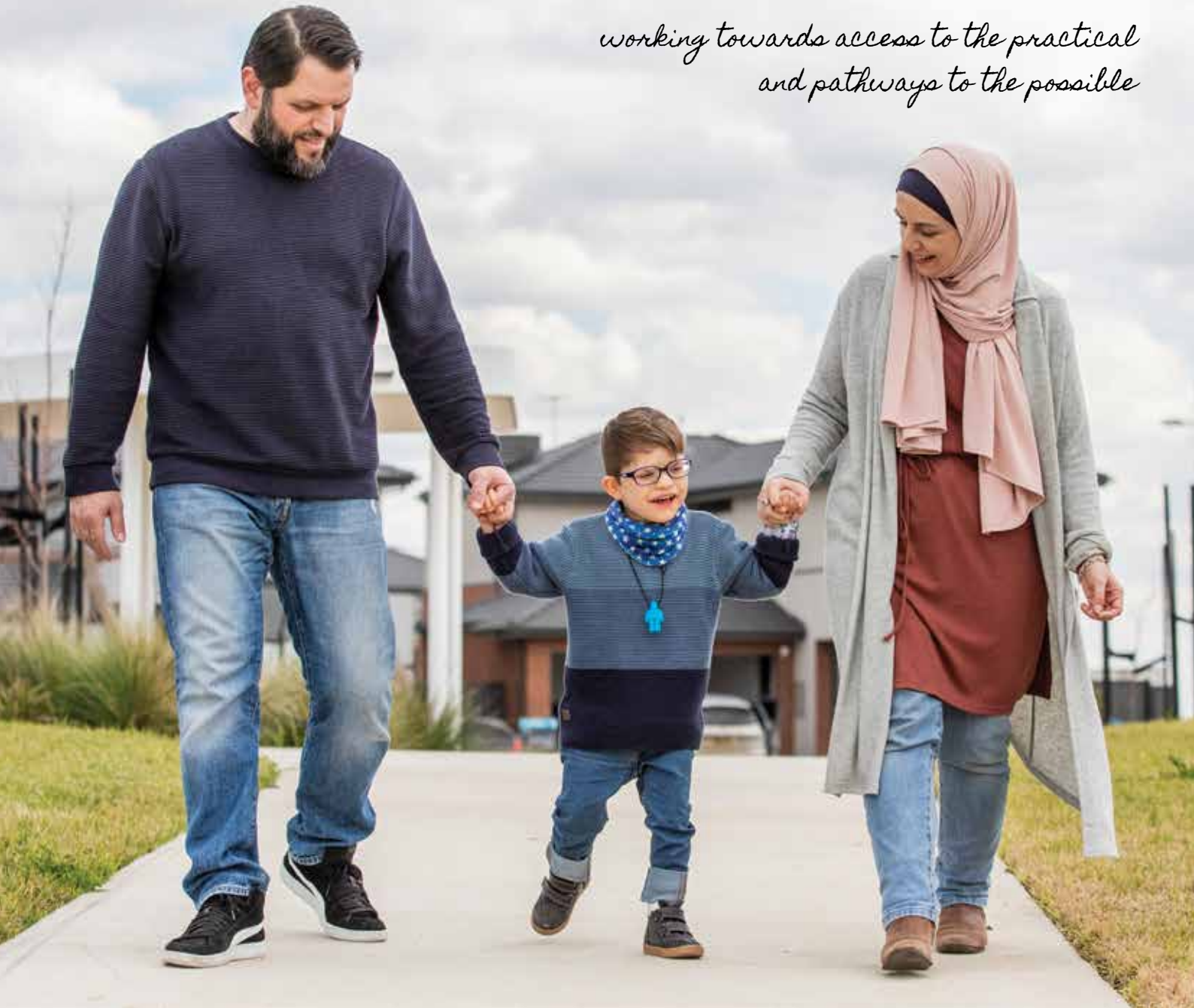
gsnv
Genetic Support
Network Victoria

2022 ANNUAL REPORT

working towards access to the practical and pathways to the possible



working towards access to the practical and pathways to the possible

Welcome to the Genetic Support Network of Victoria Inc.

(GSNV Inc.) Annual Report for period 1 January 2022 to financial year ending 31 December 2022.

As we reflect on the year in review, 2021, we highlight some of our major achievements, challenges and outcomes, including an overview of governance and financials.

The 2022 GSNV Annual Report cover photo was taken as part of the Diagnosis Day lived experience video project, a collaboration between the GSNV and Melbourne Genomics Health Alliance (MGHA).

This seven-episode series follows six amazing Victorian families through their diagnostic odyssey. This photo depicts Yusuf and his parents Selima and Kasum.

All persons included in this photo have given their permission for the image to be published and we thank them for sharing their story with us.

Genetic Support Network of Victoria

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2022
A Year of Catch Up

It's quite challenging to write about 2022.

It feels like it flew by and it was hard to take a moment to remember the world had moved on from the time warp that was 2020 and 2021.

We played catch up, we reconnected, we tentatively ventured back out into the world and then we sped it all up. We tried to make up for what we thought we'd missed and we tried to squeeze three years into one. It was exhausting and relentless and yet we as a community, endured, we found a way... as we always do.

We experienced lots of change at the GSNV. We welcomed and farewelled team members, board members and long-time friends chosing to move on to their next adventure. I acknowledge with gratitude Paul Fennessy and Kirrily Hasham from the Victorian Department of Health and Rebecca Purvis who moved on from the GSNV Committee of Management. I also acknowledge Isaac Hockey and Sammy Bowden who joined our team, contributed wonderful work and then found new chapters calling them.

Standing together has never been more important for the genetic, undiagnosed and rare disease community. So many stakeholders have reached out in 2022, wanting to engage with the community voice, some so well and others not so well. But we are learning together, what works and when, co-creating, co-designing and co-delivering. I have never been so encouraged by our community and all our supporting stakeholders.

Thank you to all in the community who have walked with us, we are privileged to count so many outstanding people and organisations as friends of the GSNV.

The GSNV was established in 1997 in response to a changing complex environment; in recognition of the importance of a broad consumer voice in genetic health; acknowledgement of a gap in existing support for many rare, undiagnosed and genetic conditions and to increase awareness of the challenges faced by people with genetic conditions and those who support them.

We are a state-wide service and we serve all genetic, rare and undiagnosed conditions – taking an inclusive approach to what is common across all conditions; for people with conditions and those who support them. Our work is underpinned by clarity of strategic direction, values and principles and is developed with a focus on Education, Advocacy and Support.

We will continue to honour the purpose for which we were created and our commitment of service to the genetic, undiagnosed and rare disease community.

Vision and Mission

Our vision and mission were reviewed in 2020 through a six-month consultation to create a new strategic plan.

Consultation included people with lived experience and those who support them, our volunteer community, patient support organisations, health professionals and other key stakeholders.

Facilitated discussions focussed on important topics such as equity and access, integrated care, data, partnerships, diagnostics and treatment, research and clinical trials.

Our Vision

- People living with genetic, undiagnosed and rare conditions flourish and live their best lives.

Our Mission

- To have support included as an integral part of the health and wellbeing care continuum.
- To drive fairness and equity of access across testing, care pathways, treatments, research, clinical trials and support.
- To be a voice and force for positive change for genetic, undiagnosed and rare disease communities.

The GSNV In Context

As our world opened up, there was a national and international wave trying to make up for the previous two years of challenge. Many delayed projects recommenced, collaborations gained momentum and implementation challenges replaced research challenges or rather added to research and health system challenges.

Research projects continued the trend of increasing community engagement and in some cases we experienced movement towards co-creation and co-deisgn which is incredible pleasing.

The GSNV continued to focus on the Department of Health National Strategic Action Plan for Rare Diseases and to ensure that the work of the GSNV and Victoria contributed to the implementation of the plan. The pillars of Awareness and Education, Care and Support and Research and Data remain relevant and there is still much to do.

We also focussed on the relationships forged and improved during the COVID years when the world felt smaller, to build on and engage more with important international partners and projects such as Rare Disease International, EURORDIS – Rare Disease Europe, APARDO – Asia Pacific Alliance of Rare Disease Organisations and the CGN4RD - Collaborative Global Network for Rare Disease.

2022 wasn't just an environment of change for the GSNV. As part of the Victorian and National landscape, the year included elections and a change of Federal Governments and change of Ministers across both the Federal and Victorian State government and in Victoria. We would like to acknowledge the support for the genetic, undiagnosed and rare disease community from Minister Greg Hunt who's legacy will impact for many years in newborn screening, carrier screening and genomic implementation.

CEO Message

Thank you to all those who supported the work of the GSNV in 2022, to all who engage with us in conversation, exploration and genuine interest, to those who offered and provided guidance and support, to those who listened and shared, to those who attended and were generous of their time and spirit, to those who gave us feedback and ideas, who challenged and made us better. You are all part of the GSNV family and our story – we serve you all and appreciate you and all you do. We value the friendship and generosity of our community and stakeholders, who don't have to do that, we know they have their own areas of focus and yet, they are always there for us.

There were some really important consultations happening in 2022 and as always our community turned up. We were part of the National Medicines Policy consultation, the Newborn Screening federal consultation, submissions to secure MBS item numbers for Genetic Counsellors, to change legislation for MITO donation, for medicare access to carrier screening, for access to medicines through the pharmaceutical benefits schedule and so much more.

Some of our support and advocacy was successful and some was not, and we go on – we build on our successes and learn from our mistakes together. We are very conscious of our responsibility to represent and to serve our community and again, thank you for allowing us to do so.

As always, our possibilities are exciting and together we keep forging ahead towards a better life for all people impacted by genetic, undiagnosed and rare disease.

GSNV Chief Executive Officer

President's Message

Reflecting on 10 years with the GSNV Committee

Wrapping up 2022 and looking ahead to 2023 I realised that 2023 would mark 10 years for me being part of the GSNV Committee of Management. I first heard of the GSNV from the inimitable Louisa De Pietro at a presentation to my cohort of The Master of Genetic Counselling at The University of Tasmania. Louisa explained the role of the GSNV in advocacy and representation for people impacted by genetic conditions like her own and I immediately wanted to know more. I, and a few other students, attended the AGM that year and became general members. The following year I was nominated to Treasurer for the committee. After I graduated, I maintained my role by zoom from Aotearoa New Zealand. Well before the pandemic, the GSNV was stiving to make participation accessible online and in person.

I contributed from my role as Treasurer for 6 years before being nominated to President, a role which is my pleasure and privilege to still hold at the end of 2022. Across these years I have seen the GSNV move from strength to strength thanks to the tirelessly caring and committed GSNV staff and the input of committee members. In 2013, the GSNV was using the Victorian Department of Health funding to stretch across everything needed, topped up with membership fees. Now, the GSNV is able to reach even further into the rare disease community with measures like making membership free and delivering the incredible Genomics in Schools program. I am proud to have been even a small part of this growth and I look forward to many more years of being involved in any way that I am endorsed to be by the other members of the committee and our GSNV community.

Rachel Pope-Couston

President

Our Committee

The GSNV and the GSNV Committee of Management ramped up to full speed with Rare Disease Day 2022 with the theme 'Show Your Colours'. It was a fantastic event and a wonderful reminder of why it is so important to have a committee whose experiences come from across the spectrum of the rare disease rainbow. The GSNV committee strives to be a space where people can show their true colours and express their opinions on the direction and representation of the GSNV and it was a pleasure to be part of these vibrant conversations throughout the year.

Since last year, the GSNV Committee has had some changes. It was sad to see Rebecca Purvis, long-term member of the GSNV Committee, step down. Rebecca has served the GSNV Committee since she joined as Vice President in 2019. We wish Rebecca all the best for her PhD and beyond. We also welcomed our new Vice-President, Julie Cini, and treasurer, Natalie McCloughan.

We would like to thank our fellow committee members for their valuable contributions to the GSNV and we look forward to another productive year.

Rachel Pope-Couston

President

Marta Cifuentes Ochoa

Secretary

Our Volunteers

Our volunteer program remains an opportunity for support groups to source suitable volunteers to help them with various tasks.

Volunteers are generally students from the Masters of Genetic Counselling Program, or students interested in applying for the Masters and seeking hands-on experience in the genetic health and support community.

Our GSNV volunteers play an integral part in keeping the GSNV office and activities running smoothly with opportunities created specifically for volunteers as often as possible.

Who We Are

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

President	Rachel Pope - Couston
Vice President	Julie Cini
Treasurer	Natalie McCloughan
Secretary	Marta Cifuentes Ochoa
General Committee Members	Giorgina Maxwell, Radostina Breedt, Sean Ong, Kristiina Siiankoski

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

The GSNV Committee is responsible for the governance of GSNV Inc. and establishes the key strategic priorities and organisational performance indicators. Key business objectives are delegated to the GSNV CEO and staff.

– As always, it's been a pleasure to be a GSNV committee member over the past year. I'm always in awe of the way so many key stakeholders - individuals, families, support groups, researchers, politicians, industry – are brought together through events such as Rare Disease Day and the breadth of wisdom shared.

Georgina

Our Organisation

The GSNV is an organisation underpinned by values and principles. These impact both our priorities and practice.

In 2020, as part of our strategic plan development, we reviewed our values and these remain unchanged. This Annual Report reflects how we live our values.

Our Values

Fairness and Equity

- We believe that every individual has an equal right to access information and services that impact the possibility and potential to live their life.
- We believe in equal opportunity to make an informed choice and to have that choice supported.

Empowerment

- We strive to educate and inform people to empower informed decision making
- We seek opportunities to provide the voice of lived experience
- We build capacity by developing and delivering education and support strategies

Respect

- We believe in the possibility and potential of all life
- We respect choice and the right to choose
- We seek diverse opinions and approaches
- We listen and learn from lived experience

Integrity

- We are ethical, tolerant and strive to deliver

Connectedness

- We collaborate for outcomes
- We engage with communities to learn
- We provide opportunities for people to network
- We share lived experience
- We link people to resources and support

Our Advocacy Focus

We are committed to advocacy that drives:

- Rare Disease to be considered as an entity in the same way that cancer is
- Engagement with lived experience community to build evidence that can be utilised broadly and purposefully including active engagement and co-design opportunities for people with genetic, undiagnosed and rare conditions in research, clinical trials and projects
- Representation means representation of the community not representation of an individual's ideas and opinions – representation must be supported
- Consumers as legitimate partners not a 'project' requirement, or 'good practice'
- Support as an indispensable component of an inclusive health and wellbeing system jointly considered in practice, planning and policy development
- Access and equity for all – for support, of information, in NDIS, of expertise, of care, of consideration, in research, in clinical trials, in treatment, in environment and in all things – with particular focus on disadvantaged communities including rural and regional Victoria, Indigenous, multi-cultural and refugee communities
- Recognition and action to build mental strength for people with genetic, undiagnosed and rare conditions and those who support them
- Recognition and action for people ageing with a genetic, undiagnosed and rare condition.
- The Importance and recognition of identity and individuality in care, decision making and support – Differences are of value and inclusiveness for vulnerable populations is expected and required
- The right to knowledge, education and information for decision making and choice
- A collaborative approach from local, State, National and international organisations
- Our place as the Victorian peak support organisation for people with genetic, rare and undiagnosed conditions and those who support them

The GSNV Team

I would like to thank Louisa Di Pietro, Hollie Feller and Kari Klein's valued support, hard work, flexibility and agility and also an outstanding year of contribution.

We would also like to thank Keri Finlay who left us in 2021 for her wonderful contribution to GSNV and our Community over many years.

The GSNV operated within our budget with a maximum of 2.5EFT (equivalent full-time) with all staff members classed as part-time. We actively recruited volunteers to support our work and support group projects as in the past.

The GSNV Committee of Management and team would like to highlight the contribution of the volunteers over the past year. They have been outstanding. The Committee would also like to recognise the hard work of the GSNV team, who demonstrate such dedication, passion and commitment to our vision and mission.

Through our commercial arrangement with the VCGS, the GSNV adopts MCRI corporate services policies and procedures who provide our Finance, HR, Payroll, Facilities and IT support. We extend our thanks for their continued support and professionalism. Our organisation would also like to extend our gratitude to Martin Delatycki, Medical Director of VCGS for his ongoing support and engagement.

EFT Allocation As At 31 December 2021

Name	Position	Classification	EFT
Monica Ferrie	Chief Executive Officer	PT	0.4
Louisa Di Pietro	Education and Advocacy Strategist	PT	0
Hollie Feller	Project Assistant	PT	0.6
Natalie McCloughan	Communications and Administrative Officer	PT	0.2
TOTAL FTE as at 31 December 2022			1.2
TOTAL Budgeted FTW			2.0
Maximum FTW During 2022			2.0

The GSNV Volunteer Program

We currently have 43 active volunteers and approximately 10 support groups we have assisted.

Potential volunteers apply through the GSNV website and are then invited for an orientation and training session alongside the official MCRI onboarding process. Jobs are then circulated among volunteers as they come available and allocated on a first come basis, but also taking experience and volunteer skillsets into account where appropriate.

Some examples of volunteer activities requested and undertaken by volunteers in 2022 include social media post creation, administration, database support, event support and much more. With over 300 hours completed across the year.

With a focus on system efficiencies being implemented in the last months of 2022 and into 2023, we will be adopting new practices to record volunteer information and matching them to support groups. It was noted though that there was a general decline in support group activities post-pandemic requiring volunteers and as such the level of volunteer engagement had significantly declined.

The volunteer program supported the following patient support groups in their work. Some examples of volunteer-support group matches in 2021 were:

NODSS Narcolepsy and Overwhelming Daytime Sleep Society

USHER KIDS AUSTRALIA

CHARGE SYNDROME AUSTRALASIA

SSPA Short Statured People of Australia

ausDoCC australian disorders of the corpus callosum

Syndromes Without A Name

AAAF Australia Alopecia Areata Foundation Inc. Alopecia Areata It's life changing

Children's Tumour Foundation Conquering NF

LEUKODYSTROPHY .org.au AUSTRALIA

Membership

2022, remained a year of increased hardship for some of our members and their communities as was 2021.

The GSNV Committee of Management, together with the GSNV team made the decision to institute free membership on an ongoing basis so as to ensure that all those in need of our support and advocacy efforts had equitable access to all that we offer.

As we did not engage members to renew for their membership we established that our active subscriber list would consitute evidence of membership.

Corporate Governance

The GSNV commenced 2022 with 8 committee members. The Committee remained committed to strengthening the Board and improving diversity.

Our Committee was consistent throughout the year. Strong governance continues to remain a focus and all relevant policies were reviewed and updated accordingly. The GSNV is fortunate to have dedicated Committee members with a very high meeting attendance record and great engagement.

It is not unusual for the Committee members to meet and discuss areas of importance between meetings or simply to support each other. We are very grateful for this environment.

The GSNV Executive and Committee continued to meet via Zoom on the third Tuesday of each month. This continues in 2023 when we hope to have a least one face to face meeting. Each meeting has a central theme and supports the strategic plan and business objectives.

Reporting on actions delivered and actions planned is a key feature with documentation provided in advance to the Committee.

The Year In Review

WE COMMUNICATED WITH

633
SUBSCRIBERS

10
NEWSLETTERS

10
GENETIC HUB NEWS

WE CONNECTED THROUGH

7
EVENTS

43
VOLUNTEERS

300+
VOLUNTEER HOURS

WE ENGAGED WITH

320+
TEACHERS

55,000+
STUDENTS

7400+
VISITORS

1076
FOLLOWERS

61
NEW FOLLOWERS

637
FOLLOWERS

141
FOLLOWERS

144
FOLLOWERS

Sector and Consumer Representation

Participation, consultation and representation are critical elements of the work of the GSNV. We are committed to listening to, engaging with the genetic, undiagnosed and rare disease community and stakeholders and providing genuine representation of the diverse views, ideas, needs and expectations.

In 2022, we engaged with a large number of organisations and committees through direct consultation, submissions, attendance at events and driving engagement alongside our essential grass-roots community engagement.

Consultations and Submissions

MSAC ID 1737
Consultation survey on MSAC application newborn bloodspot screening for sickle cell disease and thalassemia

We recommended the conditions be added to newborn screening due to the clear clinical benefits of early diagnosis

MSAC ID 1637
Expanded reproductive carrier screening of couples for joint carrier status of genes associated with autosomal recessive x-linked conditions

We sought subsidised and therefore equitable and affordable access to carrier screening to inform choice across many conditions

MSAC ID 1675
Whole genome sequencing for the diagnosis of mitochondrial disease

We believe whole genome sequencing for mitochondrial disease will improve access to information and services for impacted families

MSAC ID 1737
Genetic testing for childhood hearing impairment

Subsidised access to expanded testing improving outcomes for children experiencing deafness and their families

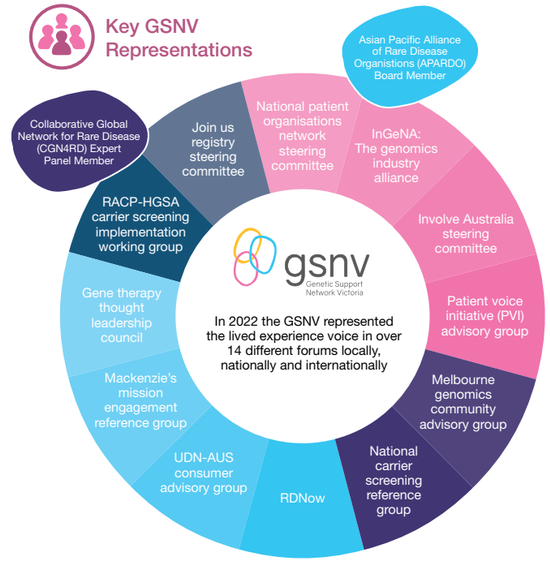
MSAC ID 1710
Newborn bloodspot screening for X-linked adrenoleuko-dystrophy

Access to newborn screening will reduce the trauma resulting from misdiagnosis that often occurs for children with this condition

Response to MBS genetic counsellor medicare benefits schedule review advisory committee draft final report August 2022

An MBS item number would allow patients to be seen by genetic counsellors in a more timely manner thus improving outcomes

Key GSNV Representations



GSNV Collaboration on Newborn Screening Advocacy

NewBorn Screening remained an area of focus for the GSNV and our community actively engaged in the national consultation around newborn screening and forwarded a collaborative response to the federal government. The collaboration to create a comprehensive response was enlightening for us all, as the process built common understanding and shared challenge across the community. We remained focussed on supporting the addition of SMA to newborn screening panels and to a positive assessment for sickle cell disease.

> The last 18 months has been a marathon in the advocacy space for Newborn Screening (NBS) for Spinal Muscular Atrophy (SMA). After the positive recommendation from the federal government in 2020, the hard work continued to successfully advocate to each state for the permanent implementation for NBS inclusion for SMA.
>
> I reached out to the GSNV to assist me in this advocacy, as I knew there was a bigger picture at play and that if we did this correctly then other organisations in the future wouldn't perhaps have such a long drawn out process. Monica and I joined a working group with Novartis and Parker and Partners and had weekly meetings with relevant stakeholders to target those in each state that needed to be across this important issue. We met with countless Ministers and ministerial officers and Department of Health representatives, to state the case for SMA and to identify and overcome any barriers for the implementation of the program. It's been a huge task, and finally we will see the implementation of newborn screening in each state starting from May 2023.
>
> Hopefully the work that we have done in this space pioneers the way for other patient organisation groups to have a much easier road than we did, and that lives are saved by the early implementation of newborn screening programs in the future.
>
> — Julie Cini, CEO, SMA Australia

Highlights from a productive year

Diagnosis Day

On the 17th of October, GSNV and Melbourne Genomics Health Alliance (MGHA) hosted a premiere event of the Diagnosis Day lived experience video series at Graduate House, University of Melbourne. A seven-episode series that follows six amazing Victorian families through their diagnostic odyssey.

Many people first engage with GSNV as they seek support and guidance following a genetic diagnosis. At this stage, most people are looking for a support group where they can receive trustworthy information and meet people going through a similar experience. This support from others in the GUaRD community has a material impact on how a person with a genetic condition, their family, their carer, and their friends navigate a diagnosis.

That's why we developed Diagnosis Day. We are passionate about co-design - and not very good behind a camera - so this series is a product of a collaboration with the Melbourne Genomics Health Alliance. The series features diagnoses of PKU, thalassemia, Pitt-Hopkins syndrome, Niemann Pick-Type C, and hyperoxaluria. What shines in this series is that lived experience is the absolute truth. Equitable access to a genetic diagnosis is vital in reducing the time that families spend navigating the 'unknown' and more time to live life at the highest standard of attainable health. We can not achieve equitable access without listening to and learning from the patient voice. We are ineffably grateful to the families for opening their doors and hearts to share their stories. It was a highlight of this project having all the families together to watch the series in full for the first time. It was heartwarming and heart-rending.

Finally, it has been a pleasure working with MGHA to create a series that not only puts the patient at the centre, but spreads the word that genomics justifies time, resources, and expertise as its development can significantly improve patient health and wellbeing outcomes.This collaboration of consumer-industry is just the first step in collaborations that are necessary for tangible health reform.

Practical Wellness Month of October

In the post-pandemic environment, creating space for our community members to upskill in practical wellness and mental health was still significantly important. The month of October as the national Mental Health Awareness Month was our home to provide a series of conversations and events over four weeks and to affirm the GSNV's commitment to upskilling support group leaders to have agency in making their communities stronger, more connected, well governed, and help people attain the highest standard of health.

This year the GSNV facilitated four online workshops that fostered the development of practical skills surrounding themes of ageing with a rare disease, compounding grief and loss, supporting siblings of someone with a rare condition, and peer support. Each workshop was led by external experts who are pioneers in their field.

We extend our gratitude to Christine Walker (Chronic Illness Alliance), Melanie Schroder (Suicide Programs), Kate Strohm (Siblings Australia), and Sharon Terry (Genetic Alliance – USA) for sharing their expertise for the benefit of our community. Almost 50 attendees participated in the Practical Wellness Month activities. Attendees included support group leaders, consumer advocates, genetic counsellors, carers, and people with a rare condition.

APARDO Conference Presentation

In 2022, APARDO has partnered with Rare Cancers Australia and participated in two capacity building events in Singapore for Patient Support Group leaders in rare cancer and rare disease across the Asia Pacific. They have delivered webinars and conference presentations covering universal health coverage, mental health and inclusion, gene therapies and genomic technologies.

In November APARDO also convened a face to face conference in Bangkok, bringing together patient support organisations, health professionals, industry and Government from across the region.

Monica Ferrie has acted as Treasurer for APARDO over the past year: The 2023 work plan will be released early next year and it will be another exciting year as the alliance are committed to leaving no-one in the Asia Pacific behind.

Rare Disease Day

Each year GSNV hosts an event for Rare Disease Day to provide our community with a selection of conversations and workshops that influence the pathway and support mechanisms of our patient support group community. T

his year the event focused on **"Global Themes, Local Action**" where themes identified across the international environment were explored in relation to how they may shape and inform the challenges Australian Genetic, Undiagnosed and Rare Disease communities will face in 2022 and created the framework for discussion groups in the afternoon workshop session.

The first half of the event shared pre-recorded presentations by international rare disease experts from **Rare Disease International (RDI), Rare Diseases Europe (EURORDIS), Genetic Alliance America & UK and Asia Pacific Alliance of Rare Disease Organisations (APARDO), and Director of the Murdoch Children's Research Institute Kathryn North.** The event attracted 110 registrations with 65 people present for the morning session, 65 for the breakout session 81.9% of respondents attended both sessions of the event.

The key takeaway messages identified from those that attended was the ongoing support from such a large group of global organisations and the sentiment that there are a lot of people who really do care! "Collaboration is needed from all sides of the table" was a common theme in the feedback as well as, "We can do this if we work together as a team!"

It was clear that the global focus and context was appreciated and useful for the audience and the GSNV wishes to thank again all our presenters from around the world and locally for their contribution.

A Rare Event at Parliament House

On the 22nd February 2022, as part of our Rare Disease Day celebration, our community came together with health professionals, researchers, industry and our politicians at a bi-partisan evening at Parliament House.

The theme of the evening was collaborative community. Kris Pierce from SCN2A Australia was our MC and we were privileged to hear from Kate Beattie about the impact on families living with Battens Disease, Professor Steve Petrou - Director of The Florey Institute of Neuroscience and Mental Health and were welcomed by a number of our parliamentary representatives.

It was the first post-COVID event at Parliament House, so we were all delighted to be there. We raised awareness and created a platform for progress. It was a beginning and there remains much work to be done.

Research Involvement

COVID-19 Journals

The lived experience of the Genetic, Undiagnosed and Rare Disease Community Research Project.

Two studies codesigned and co-led by the GSNV and Australian Genomics have recently been completed. The COVID19 Journals study recruited 29 people from the Australian GUaRD community to share their experiences of the pandemic and lockdown. We found the support required comes in targeted waves.

The findings allow us to develop a first line response to a crisis of this nature at its outset. Resources for developing new routines and adapting and resetting as change keeps coming. It seeks accountability and consistency from the health sector to develop plain English, accessible and consistent information and the implementation of mental health support and and the implementation of mental health support, protection and wellbeing from the beginning of such an event.

(Byun M, Feller H, Ferrie M, Best S (2022) http://doi.org/10.1111/hex.13405)

The Live Well study builds on the strengths of the GUaRD community to identify practical interventions to help people to 'live well'.

Themes from the COVID19 Journals were shared with the GUaRD community in focus groups for refining and prioritising. Sixteen practical interventions were identified. The emphasis was on provision of the interventions by peer support groups highlighting the role these groups can and do play. However, this prominence also raises issues of equity and access amongst the GUaRD community.

It is essential that whichever practical interventions are implemented to support the GUaRD community to 'live well', they are evaluated to ensure people living with GUaRD gain maximum benefit. Central to this evaluation is the GUaRD community who should play a key role in the design and implementation of any activities aimed at impacting people with GUaRD.

Beadell I, Byun M, Feller H, Ferrie M, Best S (2023)

"Co-designing Interventions to 'Live Well': Experiences and perceptions of the Genetic Undiagnosed and Rare Disease (GUaRD) community" Journal of Community Genetics [in press]

Family Engagement in Research

Researchers and parents of children attending Royal Children's Hospital Melbourne, including our own Hollie Feller, came together to engage in a course focusing on family engagement in research. It was a collaboration between KBHN, CanChild and McMaster University. This 10 week course sought to develop an understanding of the importance of involving families in research teams and how to implement this with a balance of power and partnership, which research suggests improves the quality and relevance of research.

The course covered communication strategies, roles and responsibilities of families and researchers in research, ethics in family engagement and much more.

As this field is continuing to grow this course has upskilled GSNV and others within the MCRI campus to lead the way with family engagement in research projects.

Melbourne Disability Research Institute

In 2022 GSNV received funding through MDI to investigate support group governance and community structures though engaging support groups who utilise online platforms to support their community. The study aims to develop a best-practice governance scale to assist us in supporting support group leaders to improve their leadership and community operation. The researcher responsible for the lead in this research project is Dr.Jennifer Beckett, a member of the rare disease community with an extensive social media background.

Master of Genetic Counselling Projects

'Exploring the Expectations of People Accessing Genetic Counselling'

This Master of Genetic Counselling project has been developed to explore the expectations people have of genetic counselling prior to accessing the service and how these expectations are met in accessing genetic counselling. It will also explore the perspectives of genetic counsellors on the expressed expectations of those accessing their service. The project will be completed next year.

'The Involvement of Rare Disease Organisations in Research Projects'

This Masters of Genetic Counselling project focusses on understanding the experiences of patient support organisations (PSO) to inform future collaborations between researchers and patients, and lead to better outcomes for both parties. The study seeks to address questions around how PSOs experience involvement in research projects, the potential enablers and barriers impacting their involvement and whether support during the process is required. The project will be completed in 2023.

Education

Education is a vital part of the core work of the GSNV.

Education activities are grouped as follows:

Professional | Community | Student

Professional

MCHN conferences

In 2022 the State-wide Maternal Child Health Nurse conferences remained virtual events.The GSNV was represented through the inclusion of flyers and information brochures covering the services offered by our organisation.

A summary flyer was distributed through the exhibition website to advise nurses how the GSNV could assist them with supporting their families with genetic conditions.

Genomics in Schools

Commencing in 2019 Genomics in Schools is by far the GSNV's biggest project to date. During 2020 the first stage of the project was launched to Victorian teachers and students studying Biology in Years 11 and 12, specifically targeting the topics within the VCE study design. Over 320 teachers across Victoria requested access to the free downloadable resources which created a reach of over 55,000 students! The resources are being used in both metro and rural schools with over one third of teachers using them within rural schools.

The success of this program initiated an invitation from the Victorian Department of Education to contribute to a program of science challenges for Middle Year levels. This program, known as the Victorian Middle Years Science Challenge, is heralded by Museums Victoria and will be launched on the Scienceworks website in 2023.

During 2022 the GSNV created content for three challenges for Years 5-6, Years 7-8 and Year 9. These challenges have allowed us to continue educating students on the emerging technologies within genomics and begin a dialogue that empowers them as individuals to think differently about our population's diversity and the role genetics may play in this.

Under the leadership of Ms Hollie Feller, the GSNV has continued to accelerate this project in 2022 with the successful application in the Australian Genomics Implementation Project grant.

The grant has allowed us to commence the implementation of a national rollout of the work that has been accomplished under the two stages of the project across Years 5-12, with the development of one national science curriculum resource focused on the understanding of everyday health utility of genomics through lived experience storytelling. Two educators have been engaged to assist with this task as the team create content for release on its own website in early 2023.

The GSNV wishes to thank the ongoing volunteer commitment of the Genomics In Schools steering committee.

Post-Graduate Education

Fit for Practice Master of Genetic Counselling community placement with patient support groups.

The GSNV is proud to be directly involved in education and contributing to the training and education of health professionals in Victoria. In collaboration with the University of Melbourne (UoM), Master of Genetic Counselling, the GSNV has provided coordination of the Fit for Practice subject, Community Placements Program.

The Community Placements experience and a reflective report are a formal learning requirement of the Genomics in Practice subject.

The two tasks meet the following learning outcome/s for this subject:

- Develop an understanding of the multidisciplinary nature of health care
- Demonstrate an understanding of how support organisations can be incorporated into genetic health care
- Demonstrate an understanding of the impact of disability on an individual and their family

Community placements are designed to maximise experiential learning and promote the concept that support is an essential component of the genetic health sector but knowledge and understanding of how it is offered and where it is, is limited amongst health professionals.

The program has moved forward with incorporating an understanding of 'support' and the 'support sector' as an important learning outcome area and one that will heighten the student awareness of impact on families and individuals.

Under the Community Placement students work in groups to experience the realitities of host support/advocacy organisations.

The feedback from both students and support groups was extremely positive, it is always challenging to support 'new' people into organisations and we are extermely grateful to our host organisations who provide an experience which in the longer term benefits our entire community.

The Community Placement program offers an opportunity for the support sector to influence future professionals and have some input into the awareness and understanding of the huge contribution they make to genetic health care. The GSNV is privileged to be a leader in the coordination of that exchange and believe it to be even more relevant in a changing health and support landscape.

Placements and Internships at GSNV

Emily Johnson joined us from the US to complete a 12 week internship as a post graduate student with a Bachelor's degree in Genetics & Genomics from the University of Wisconsin – Madison.

Coming to us with an interest in maternal and infant health, specifically in rural and First Nations populations, she provided support to several of our key projects for the year.

Emily gathered background information on international newborn screening programs and how Australia can make amendments to the current program so that all babies have a chance at their best quality of life. This will support a Federal submission due soon.

As a key contact for Rare Diseases New Zealand Emily has, and will continue to support the development of an informative document outlining those medicines available in Australia and world-wide that are subsidised allowing for equitable acess.

Emily also assisted Isaac develop our Equity and Diversity policy statement. Emily recently returned to the US for what she hopes is a brief stay before returning to commence medical school in Sydney in 2024.

Sarah Bainbridge joined us to complete a Science and Technology Internship as part of her University of Melbourne science degree.

"I had the privilege to work on updating the NDIS resource guide for the GSNV website. I've learnt a lot with GSNV and am super grateful for the opportunities I've had during this semester!" This document is the one stop guide to the NDIS and aims to collate all the information currently available into one easy to read document on the GSNV website.

Financial Report 2022

The financial report for the period 1 January 2022 – 31 December 2022 has been provided by the Committee Treasurer on behalf of the executive and committee of the GSNV.

The GSNV continues to manage its budget well and will carry forward a small surplus into 2023. Despite the continuing impacts the COVID-19 pandemic has had on the sector the team have delivered well on their business plan remaining in budget.

As part of our agreement with VCGS our accounts were again audited by KPMG. The report findings can be found below and concluded that the GSNV accounts were in order.

We remain extremely grateful for the continued grant funding from the Department of Health, which allows us to continue our great work. The GSNV continues to responsibly manage its finances under guidance from the committee, which places us in an excellent position to support those we serve in the coming year.

GSNV
2022 High Level Summary

YTD through December 22 – **CONSOLIDATED**

OPERATING ITEM	$–000's
Vic Govt Grant	255.4K
Other Income	37.3K
TOTAL REVENUE	**292.8K**
Saleries And Related Costs	111.7k
TOTAL EMPLOYMENT COSTS	**111.7K**
Computer Software Expenses	1.9K
Computer Hardware	8.0K
Consultants	128.7K
Printing, Stationery And Photocopying	0.2K
Catering	0.1K
Other Administrative Expenses	24.8K
Other Expenses	0.3K
Staff Training And Conferences	0.3K
TOTAL OPERATING EXPENSE	**275.9K**
OPERATING RESULT BEFORE SHARED COST ALLOCATIONS	**16.8K**
Corporate Services and Facility – IT / HR/ FIN	16.8K
TOTAL SHARED COSTS	**16.8K**
OPERATING SURPLUS / (DEFICIT)	**0.0K**
	–
TOTAL DEPRECIATION AND INVESTMENTS	**–**
TOTAL SURPLUS / (DEFICIT)	**0.0K**
UNSPENT FUNDING ROLLED FORWARD TO 2023	**109.6K**

THANK YOU

I am so lucky in my role at the GSNV, I always feel like I get to work with the best people and I acknowledge you all and the work you all do.

The GSNV continues to appreciate and value your gifts of time, perspective, talent, expertise and knowledge. We thank those who have donated and those that continue to offer support to us, our community and each other.

We are especially grateful for the ongoing support of the Department of Health who continue to provide funding and support for our important work. We thank in particular Dr Paul Fennessy and Kirrily Fasham who have moved on to their next chapters and Marianne Griffin for an ongoing supportive relationship with the GSNV and their wisdom and counsel year in, year out.

We are also thankful for the professional relationship including support and advice from all our stakeholders including people with genetic conditions, clinical services, universities, support groups and so many others. A special mention to Martin Delatycki for his ongoing support and generous time.

VCGS
Leaders in Genetic Health

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Audit Statement

I confirm that the funds belonging to Genetic Support Network of Victoria are being held by the Victorian Clinical Genetics Services (VCGS).

The accounts of the VCGS are audited annually by KPMG who are our External Auditors. The accounts for 2022 have been audited by KPMG in accordance with this practice.

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

Kind regards,

Emma Amalfi CA

VCGS General Manager of Finance

Murdoch Children's Research Institute /

Victorian Clinical Genetics Services

gsnv
Genetic Support
Network Victoria

working towards access to the practical and pathways to the possible

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