gsnv

Genetic Support Network Victoria



2024 ANNUAL REPORT

Working towards access to the practical and pathways to the possible.

gsnv
Genetic Support
Network Victoria

Welcome to the Genetic Support Network of Victoria Inc.

Welcome to the Genetic Support Network of Victoria Inc. (GSNV) Annual Report for the period 1 January 2024 to 31 December 2024.

As we reflect on the year in review, 2024, we highlight our major achievements, challenges and outcomes including an overview of our governance and financials.

The 2024 GSNV Annual Report cover photo:

Pictured L-R: Dominic Cy (aged 9), Chloe O'Neill (aged 17)(front), Zoe Emmerson (aged 10), Patrick Corrone (aged 11), Damien Cy (aged 9).

Image description: Pictured are 5 young lived-experience participants who contributed their experiences of living with genetic and rare disease in the Science Within Us, a digital education resource on exploring our genetic identity. The GSNV thanks the children and their families for contributing their lived-experience to support the education of genomics and genetic health in Victorian primary and secondary schools.

Image credit: Susie Roczo-Farkas for the Science Within Us launch event.



VICTORIA State Government | Department of Health

gsnv Genetic Support Network Victoria

Genetic Support Network Victoria
ABN: 510 07032 760
INC NO: A0037552M

The Royal Children's Hospital
50 Flemington Rd, Parkville. VIC 3052

Email info@gsnv.org.au
Phone (03) 8341 6315
Web www.gsnv.org.au

gsnv
Genetic Support
Network Victoria

CONTENTS

2024 – SHIFTING SANDS	4
ABOUT THE GSNV	4
THE GSNV IN CONTEXT	5
VISION AND MISSION	5
CEO MESSAGE	6
PRESIDENT'S MESSAGE	7
OUR COMMITTEE	8
OUR ORGANISATION	9
OUR VALUES	9
OUR VOLUNTEERS	9
OUR ADVOCACY FOCUS	10
CORPORATE GOVERNANCE	10
OUR TEAM	11
DIVERSITY AND INCLUSION	12
UNDERSTANDING UNCONSCIOUS BIAS WEBINAR	13
COMMUNICATIONS & ENGAGEMENT	14
OCTOBER PRACTICAL WELLNESS MONTH	15
SECTOR AND CONSUMER REPRESENTATION	16
2024 SUPPORT GROUP ENGAGEMENT SURVEY	17
RESEARCH INVOLVEMENT	18
THE SCIENCE WITHIN US	21
THANK YOU	22
FINANCIAL REPORTS	23





2024 – SHIFTING SANDS

2024 was another huge year for GSNV and for our community. Many achieved amazing outcomes in the face of continuing challenges. There were inaugural conferences, resource development, research engagement, publications, consultation participation, Government, MSAC and PBAC submissions and more. Our community of Support Group Leaders travelled to local and international conferences and meetings, all amongst juggling the multiple demands of family and carer priorities, mobilising teams, volunteers and organisations, all, to advocate for community needs and rights . None of which is easy.

In closing out 2024, I would like to acknowledge the hard, relentless work and commitment to positive change by all our stakeholders. Our community continues to grow and we've welcomed fellow travelers and friends from across the globe, the country, different stakeholder groups and communities. I'm looking forward to what we can achieve together in 2025.

The GSNV thanks our amazing and resilient genetic, undiagnosed and rare disease community who continue to support our advocacy efforts.

There remains much to do to ensure equity of access and experience, appropriate and accessible pathways and practical implemented practice for each individual, family and community members in all aspects of experience to impact quality of life.

It is a privilege to stand with our community, friends and alongside organisational allies dedicated to the rights and agency of individuals and their families living with genetic, undiagnosed and rare disease in Victoria and across Australia.

ABOUT 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.

The GSNV is a Victorian-wide service supporting all genetic, rare and undiagnosed conditions with an inclusive approach for people with conditions and those who support them.

Our work is underpinned by clarity of strategic direction, values and principles and a focus on Education, Advocacy and Support.

The GSNV continues to honour the purpose for which we were created and our commitment of service to the genetic, undiagnosed and rare disease community.





THE GSNV IN CONTEXT

The world around us continues to shift and 2024 brought forward the many challenges established in the COVID years. In Victoria, pressure on our health system continued to build, resourcing and expertise continued to outstrip supply and our ability to impact was within a context of financial constraint and priority setting across all Victorian portfolios.

We found ourselves in an interesting environment. Never, has our community been more sought out to engage with, asked for our views, opinions and expectations, actively invited to participate in research through Community Advisory Groups and, to offer our voice and lived experience.

However, the impact is hard to measure, and even when measured is disappointing. A reminder that we need to hold our partners – genetic and health services, institutions, governments, industry, support services and others accountable for change, not, congratulate them for being inclusive. I think they want to be, but working out how remains the most difficult and elusive piece of the change model.

The opportunities of genomics and precision medicine are, more than ever, reaching across the health landscape and equitable benefits will be harder to achieve. This will challenge us over the next few years.

Shifting our focus from equitable access to equitable experience, which includes access, includes opportunities to achieve a diagnosis, includes cultural safety, includes being able to access the support structures, systems and networks that are needed will drive our advocacy objectives in 2025.

VISION AND MISSION

2024 was the first year of the GSNV's new Strategic Plan. Our plan was developed in consultation with our community and stakeholders and will guide our work to the end of 2026.

Our vision remains the same, with changes to our mission to reflect how the environment has changed. Our mission reflects our commitment to be driven by the community, to recognise our community as lived experience experts and to working in partnership with all the stakeholders who can impact quality of life across, with and for the community. This new mission and strategic objectives are now guiding our work:

- Patient and Community Led Agendas and advocacy
- Connected Community and Stakeholders
- Increasing capacity to optimize relationships for community and stakeholders
- A strong and agile GSNV

gsnv
Genetic Support Network Victoria

Our Vision

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

Our Mission

Champion the long-term sustainability and value of support strategies and systems including genetic, undiagnosed and rare disease patient support groups.

Build stakeholder capacity to improve engagement and collaborative outcomes.

Be an enabler of a collaborative and connected support sector voice to explore issues/opportunities and co-design strategies and solutions.

Listen and learn from people living with GUaRD and their families and support groups to represent, lead and advocate for positive change.

CEO MESSAGE



Monica Ferrie
CEO

Each year it is my privilege to share a message in our Annual Report. I feel the weight of all the amazing community members doing outstanding work as I write. I know it's not about any single one of us, not about individual achievement but our collective commitment to improving quality of life for all of us.

In 2024, armed with our new strategic plan, we prioritised listening and seeking to better understand our community, the practical and the deeper shared challenges level. You told us through research, consultation and engagement that quality of life was your number one priority, the health journey is only a part of your story. We sought to honour your priorities and sharing by showcasing the amazing work of the patient support sector, creating more opportunities for connection through Rare Disease Day, our October wellness series, and the establishment of RareNET – our monthly network forum and importantly, increased representation opportunities on projects and in advisory groups.

Its also important to highlight a significant policy advancement in 2024. Diagnosis is the gateway to care so action in the newborn screening area was noted with hope by our community.

We saw positive change in the area of newborn bloodspot screening (NBS). NBS is a cornerstone of healthcare in Australia providing options and pathways for parents in a time period where treatments make a difference.

The community was pleased with the establishment of the expanded NBS programme to provide Australians with access to testing for more conditions where impact is critical and timely.

Yes, it would be excellent to move faster but we as a community are encouraged by the momentum we see in NBS and adding conditions to the national NBS panel. Federal and state cooperation and appropriate funding has also meant that states are ready to implement as they are now part of a revised decision making pathway.

There are many conditions not yet present on the NBS panels and this work will continue in 2025.

Finally, I would like to say thank you to all of you and also to the GSNV Team for the amazing work over the year. Hollie, Daniele, Malavika and Susie, our volunteers, our students and all our stakeholders. Key projects delivered in 2024 are covered in our yearly highlights but I would like to acknowledge the launch of The Science Within Us platform for schools in genetic education. We have received incredibly positive feedback since the launch and look forward to reporting its success over 2025. A special shout out to Hollie Feller who delivered this outstanding result.

I expect the shifting sands of 2024 to continue in 2025, we are going to be continually challenged to ensure equity of experience for all and finding collaborative solutions to create the change we need.

Monica Ferrie
CHIEF EXECUTIVE OFFICER

PRESIDENT'S MESSAGE



Rachel Pope-Couston
PRESIDENT

I would like to thank everyone who has contributed to another amazing year for the GSNV, from every member who engaged with our services, events and social media, to the incredible staff at The GSNV and the members of The GSNV Committee of Management.

I feel very proud to be contributing in my own small way as President of the Committee, and to be working alongside a dedicated and passionate group of people who are using their unique personal and professional experiences with rare disease to benefit The GSNV.

Every perspective is important and reflecting on the goals for the committee for 2024 which included expanding the committee with new members to broaden our perspectives, I believe we have delivered on that goal wonderfully.

It has been my pleasure to witness the work the staff at the GSNV have completed and the new initiatives they are undertaking, and to build new connections in the committee.

Working as a genetic counsellor in clinical genetics in public health, the resourcing pressures, and increasing need for services, seem to be building exponentially with the funding and vision for rare care lagging behind which can quickly lead to feeling overwhelmed and even a little hopeless or burnt-out.

I know many in our rare disease community are experiencing these feelings too. Some of the most powerful antidotes to these feelings are hope and community connection, both of which my role at The GSNV provide for me, and many of the events facilitated by the GSNV also try and make space for members to build community and hope for the immediate and longer-term future.

I do truly believe there are many reasons to be hopeful as we look towards 2025 and the upcoming opportunities to have a voice in the direction and focus of rare care initiatives, and I look forward to participating in the contributions The GSNV will make on behalf of it's members.

Rachel Pope Couston
PRESIDENT

OUR COMMITTEE



What a year 2024 was for the GSNV!

We had a great start of the year with Rare Disease Day. Not only did we celebrate the shared journey with our colleagues from UsherKids, but Monica and the team shared some of the main results from the research study Living with Rare Disease in Australia. This has been a long-time coming piece of work, and the committee is very proud of this achievement. This work was presented to other main stakeholders and genetics community at the HGSA Annual Scientific Meeting.

Another very exciting project took off mid last year. Led by Hollie Feller, the program The Science within Us was launched, a program that will improve the lived experience and genomics health literacy of school-aged children across Victoria. This has been a phenomenal piece of work, and we can't wait to see the positive changes that this program will bring to our community.

Our committee has witnessed the effort and great work that GSNV has put this year in completing these projects. We have been discussing new initiatives and MBS funding in genomics and the ongoing need for consultation with the communities for a more successful implementation of these initiatives. In 2025, we will keep the conversation going for actions to be taken in this space.

We welcomed Caitlin Howley this year. Caitlin not only shares her enthusiasm for the work GSNV is doing for the community but brings a wealth of experience in research policy around genomics.

I am looking forward to another year of progress (slowly but steadily!) and I would like to thank our fellow committee members for their valuable contributions to the GSNV.

Marta Cifuentes Ochoa

SECRETARY

The members of the Executive and Committee who held a position at any time during or since the 2024 AGM are:

President	Rachel Pope-Couston
Vice President	Julie Cini
Treasurer	Giorgina Maxwell
Secretary	Marta Cifuentes Ochoa
Committee Members	Keri Finlay Caitlin Howley



Julie Cini
VICE PRESIDENT



Giorgina Maxwell
TREASURER



Marta Cifuentes Ochoa
SECRETRY



Keri Finlay
GENERAL MEMBER



Caitlin Howley
GENERAL MEMBER

Having been a previous employee of the GNSV, I have first-hand experience of the important and varied work that the organisation carries out. Through the role of board member, I find I learn so much about the broader genomics space and I am proud to be a part of informing the strategic direction of the GSNV. It is clear that over the years the GSNV has become a strong well respected leader in this space and this is predominantly due to the hard work of the GSNV team.

Keri Finlay, General Member

I am really enjoying being part of the GSNV Committee of Management, particularly for the opportunity to review the GSNV's progress and discuss the organisation's impacts for improving outcomes for patients with genetic conditions, in the context of the genetics and genomics landscape in Victoria, nationally and internationally.

Caitlin Howley, General Member

OUR ORGANISATION



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

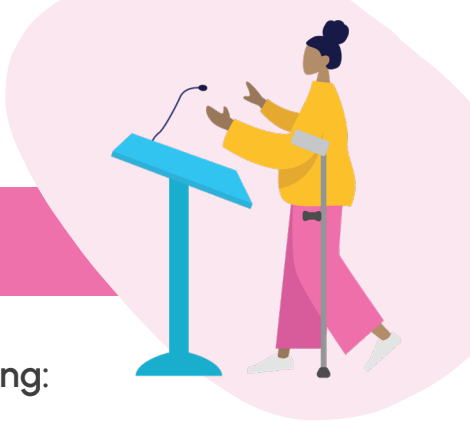
- We collaborate for outcomes
- We engage with communities to learn
- We provide opportunities for people to network
- We share lived experience
- We link people to resources and support

OUR VOLUNTEERS

In 2024, the GSNV received support from volunteers to achieve our key projects across the year such as the monthly eNewsletter, Care for Rare, promoting the October Practical Wellness Month event, assisting with the Support Group Engagement Survey and providing hands on support for the launch of The Science Within Us.

This year, the GSNV participated in the University of Melbourne's Masters of Genetic Counselling Fit for Practice program. Working with first year students, the GSNV delivered the Support Group Engagement Survey. Fit For Practice provided an opportunity to coach and mentor Masters of Genetic Counselling students to build awareness of the challenges and opportunities faced within the genetic, rare and undiagnosed community.

We are incredibly grateful and appreciative of all volunteers and their ongoing commitment to improving the lives of the genetic, undiagnosed and rare disease community.



OUR ADVOCACY FOCUS

We are committed to advocacy that drives change including:

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 that 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

CORPORATE GOVERNANCE

The Committee prioritised its leadership to strengthen the Board and improve diversity. Strong governance continues to remain a focus and all relevant policies were reviewed and updated accordingly.

The GSNV is grateful to have dedicated Committee members with a very high meeting attendance record and great engagement.

In 2024, The Committee adopted a change of meeting frequency to bi-monthly via Zoom. 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.



OUR TEAM

The GSNV Committee acknowledges and values the hard work, support, flexibility, agility and support of the team: Hollie Feller, Daniele Noel, Malavika Vasanth and Susie Roczo-Farkas.

The GSNV operated within our budget with a maximum of 2.0 FTE. We actively recruited volunteers to support our work and support group projects as in the past.

The GSNV Executive and Committee would like to highlight the contribution of the volunteers over the past year. The Committee recognises 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 Victorian Clinical Genetics Services (VCGS) governed by a Memorandum of Understanding, the GSNV adopts the policies and procedures of VCGS and receives corporate services including Finance, HR, Payroll, Facilities and IT Support.

We extend our thanks for the continued support and professionalism of the VCGS team.



Monica Ferrie
CEO



Hollie Feller
PROJECT OFFICER



Daniele Noel
COMMUNICATIONS AND VOLUNTEER COORDINATOR



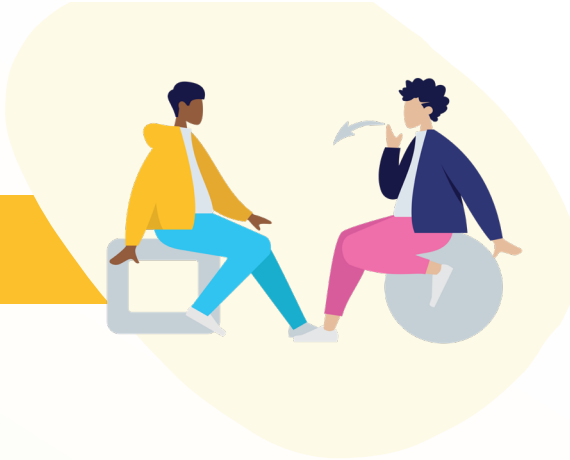
Malavika Vasanth
PROJECT AND POLICY COORDINATOR



Susie Roczo-Farkas
RESEARCH PROJECT OFFICER

EFT Allocation as at 31 December 2024			
NAME	**POSITION**	**CLASSIFICATION**	**EFT**
Monica Ferrie	Chief Executive Officer	PT	0.4
Hollie Feller	Project Officer	PT	0.6
Daniele Noel	Communications and Volunteers	PT	0.5
Malavika Vasanth	Project and Policy Coordinator	PT	0.4
TOTAL FTE as of 31 December 2024			**1.9**
TOTAL Budgeted FTE			**2.0**
Maximum FTE during 2024			**1.9**
Susie Roczo-Farkas – LINEAGE project (Research Project Officer)			**0.6**

DIVERSITY AND INCLUSION





Malavika Vasanth

"Inclusion isn't something we add on later, it's something we try to build in from the beginning. Whether we're designing a resource or running a workshop, we're always asking: who might be missing, and how can we bring them in?"

At GSNV, diversity and inclusion are at the core of everything we do. We are committed to ensuring that our initiatives, resources, and support systems are accessible to everyone, regardless of their background, ethnicity, or genetic condition. Over the past year, we focused on expanding our understanding of diverse community needs, embedding inclusive practices within our work, and strengthening our partnerships with key stakeholders. Our goal is to create an environment where individuals and families feel heard, respected, and supported in their journey.

Building Capacity for Culturally Inclusive Support

One of the key focuses this year was empowering support workers and community leaders to better engage with multicultural and diverse communities. A significant milestone in this effort was the "Understanding Unconscious Bias – Creating a Safe Space for Support" webinar, co-hosted with the Centre for Culture, Ethnicity & Health (CEH). This session provided valuable insights into how unconscious bias can affect communication and trust building in support work. It equipped participants with practical strategies to address these biases, fostering more inclusive environments in health and community services.

Benchmarking Online Support Groups: MDI Research Project

In collaboration with the Melbourne Disability Institute (MDI) and the University of Melbourne, the Benchmarking Communities of Support research project came to a successful conclusion last year.

The report delves into key elements such as best practices, community needs, governance structures, and platform choices that foster successful engagement within online support networks. The findings will be shared in a comprehensive report, set for release in June 2025. This report will include actionable recommendations for support groups to enhance the inclusivity of their online communities.

Alongside the report, we are developing a plain language resource to make the research findings more accessible and easier to implement. This tool will provide practical guidance for support groups looking to apply these insights and improve their own online platforms.

Internal Initiatives and Continuous Learning

Internally, we remain committed to continuous learning in the areas of cultural understanding and inclusivity. Over the past year, our team has constantly improved their understanding of inclusive language, accessibility, and cultural sensitivity. These efforts have been embedded into our daily operations, ensuring that all our materials, communications, and services reflect our values of respect, equity, and inclusion.

This commitment to inclusivity is reflected not only in the language we use but also in how we design our resources. Whether simplifying complex medical language or ensuring that materials are visually accessible to people with diverse learning needs, we aim to make our resources as inclusive as possible.

As we move forward, GSNV will continue to prioritise diversity and inclusion in every aspect of our work. We recognise that the journey toward full inclusivity is ongoing, and we are committed to continually evolving our practices to better meet the needs of the communities we serve. By actively listening to and learning from those with lived experiences, we aim to foster a future where everyone affected by genetic conditions has access to the resources, support, and care they deserve.

LOWITJA INSTITUTE

eccv

Australian Multicultural Health Collaborative
OUR CULTURES OUR LANGUAGES OUR HEALTH

Reflection on the

UNDERSTANDING UNCONSCIOUS BIAS WEBINAR

18th July 2024

By Malavika Vasanth



Last year, I had the privilege of partnering with the Centre for Culture, Ethnicity & Health (CEH) to co-host a webinar on *"Understanding Unconscious Bias – Creating a Safe Space for Support."*

This collaboration was a meaningful opportunity to bring attention to the challenges faced by multicultural communities in accessing support for genetic conditions. It also aligned deeply with my work at GSNV, where inclusivity and cultural competence are at the core of everything we do.

As the Policy & Projects Coordinator at GSNV, my role often involves developing resources and strategies to create more accessible and supportive environments for individuals living with genetic conditions. During the webinar, I shared insights on how unconscious bias can unintentionally affect the support experience, especially for families from culturally diverse backgrounds. My focus was on providing practical guidance for support workers on how to create spaces that not only acknowledge but celebrate these differences, ensuring that everyone feels heard, respected, and supported.

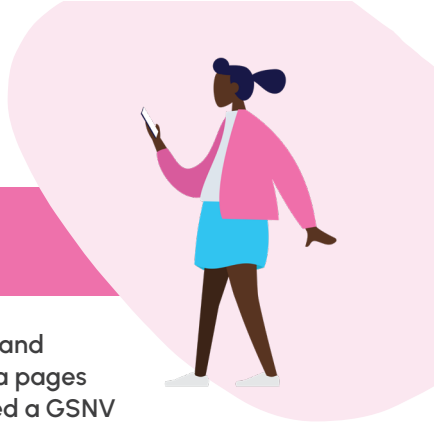
One of the most impactful parts of the session was discussing a real-world case study that demonstrated how cultural misunderstandings and language barriers can severely limit engagement in support groups. The case illustrated the importance of building trust and actively listening, which are often overlooked but essential for creating a safe environment. It was a powerful reminder that inclusivity isn't just about making resources available; it's about making sure they are delivered in a way that resonates with those they are intended to serve.

This experience with CEH was an invaluable reminder of how necessary it is to confront our own biases and work towards creating more empathetic and effective support spaces. The feedback we received from webinar participants reinforced the importance of this work, with many sharing their own experiences and asking how they could apply the strategies discussed in their own practices.

At GSNV, we are committed to creating resources that are culturally sensitive and inclusive, and this collaboration with CEH was a perfect extension of that mission. I'm proud of what we were able to achieve through this webinar, but more than that, I'm excited about the ongoing conversation it has sparked within our community. The work doesn't stop here, this is just one step in our continued journey to enhance inclusivity in the support we provide.

Reflecting on this collaboration, I am reminded of the power of learning, listening, and evolving together. The experience was both humbling and inspiring, and I'm eager to continue this process with CEH and other partners, ensuring that all individuals and families facing genetic conditions receive the support they deserve.

If you missed the webinar or would like to revisit the discussion, a recording is available on the CEH website. I encourage everyone to watch and reflect on how unconscious bias might be influencing their own work.



COMMUNICATIONS & ENGAGEMENT



Daniele Noel

In 2024, communication strategy and tactics improved the reach and growth of GSNV across multiple channels. The GSNV social media pages grew in engagement and followers as communications established a GSNV voice and implemented greater thought leadership on rare disease and genomics advocacy both locally and internationally.

For our community, the GSNV shared the many positive activities and initiatives of support groups through our eNewsletter Care for Rare. This saw positive engagement with our monthly newsletter resulting in strengthened awareness and engagement with the organization, its events and activities.

2024 was the year the GSNV returned to in-person events such as **Rare Disease Day** and nearing the end of 2024, the launch of **The Science Within Us.**

Our advocacy initiatives were further strengthened with support groups meeting monthly for the GUARD Collaborative RareNET monthly forum, October Practical Wellness month delivering new workshops and webinars supporting learning of diversity and inclusive practice.

2024 also saw the delivery of the Support Group Engagement survey highlighting the priorities of our community and the opportunities that presented for the GSNV to enhance its service delivery and strengthen its communications channels all, to continue its mission to build positive relationships with community and advocate for PLWRD on a local, national and international level.

Social Media Engagement

2024 Social Media analytics demonstrated an increase in followers and engagement across all social media platforms. The communication strategy applied the use of a consistent GSNV voice and branding to share the organisation's and the community activities, campaigns and advocacy efforts. As a result, the GSNV's organic reach has increased across all platforms.

f	1.2k	Total Followers
	714	Total Followers
in	613	Total Followers

eNewsletters

600 SUBSCRIBERS

Care for Rare – The GSNV's monthly eNewsletter for the genetic, undiagnosed and rare disease community.

Targeted to Support Group readers, Care for Rare's content includes updates on the advocacy activities of the GSNV at state, federal and international level. The newsletter also shares highlights of the GSNV team and their respective activities across research, education, volunteers and resources, diversity and inclusion.

This shift in content design demonstrated the GSNV's focus on building the capacity of our support group stakeholders, informing our community as thought leaders and recognizing the work and impact of our support group community in the GUaRD sector. The GSNV continues to deliver targeted content both Care for Rare and **In Focus** – a half yearly update to government stakeholders.

Rare Disease Day 2024 – 29 February 2024

Rare Disease Day 2024 saw a return to in-person event for the GSNV. Offering a panel discussion featuring support group leaders, Madelene Rich – CHARGE Syndrome Australia, Julie Cini – VP, GSNV Board, Caitline Howley – Genomics Research Project Officer, Australian Genomics and Heather Renton – SWAN Embassador and Consumer Advocate. The panel led by Monica Ferrie explored the recently commissioned report *About Us - Living with Rare Disease in Australia* which highlighted the voices of people living with rare disease in Australia and their needs and wants for themselves and their families across the diagnostic and health management journey. Survey data reveal common themes across quality of life, financial impacts relating to medicines and treatments and mental health.

RARE DISEASE DAY®

OCTOBER PRACTICAL WELLNESS MONTH



Each year, the GSNV's Practical Wellness Month is hosted throughout the month of October, as it coincides with national mental health awareness month and Carers' Week.

In 2024, the program coordination was led by GSNV's Communications & Volunteer Coordinator, Daniele Noel with contributions from the GSNV team and assistance of student volunteer, Naduni Elvitigalage. The month-long program hosted a total of five workshops designed to build the capacity of support group leaders to deliver mental health and wellbeing support to their communities.

Each year, the program seeks to introduce contemporary themes and respond to the challenges faced by support groups and individuals living with rare disease. This year, new workshops were introduced on Trauma-informed care, co-designing Mental health toolkits for the rare community and, how to build and manage online communities. New workshops were successful with full capacity attendance and delivered by health professionals, advocates and researchers with expertise in genetic and rare disease.

The new workshops added to the 2024 October Practical Wellness Month program included:

Trauma informed Care within the Diagnostic Environment presented an hour-long comprehensive exploration into understanding trauma informed care as a practice embedded into the delivery of genetic counselling services and mental health services to improve dialogue and interactions with PLWRD. Featuring a panel discussion, genetic counsellors Lyndon Gallacher and Anita Gorrie shared insights and practical strategies along with Psychologist Anthony Hurst to support the diverse audience of health professionals, individuals and support group leaders in attendance to apply this practice into their interactions to improve the patient experience.

Building and Managing Support Groups featured research by Dr. Jennifer Beckett, School of Culture and Communications at The University of Melbourne on the value of online communities and the practical experience of Huntington Victoria's CEO, Tammy Gardiner on the challenges and opportunities of adopting online communities as a support channel for the community. Dr. Beckett's report provided recommendations and insights into how to establish, moderate and ensure governance of online support groups. The workshop was valuable for new and established support groups and offered guidelines to improve and ensure the mental health, safety and wellbeing of all online participants.

RareNET October Forum hosting a mental health and wellbeing theme featuring Matt Bolz-Johnson EURORDIS-Rare Diseases Europe and Lauren Roberts, Rareminds provided comprehensive presentations on the Rare Minds Matter survey and the EURODIS Mentally Healthy Toolkit co-designed with the rare disease community in the UK. Both their work demonstrate the possibilities for improved and targeted mental health services for the Australian rare disease community. Hosted by the GUARD Collaborative's monthly RareNET Forum offered an advocacy channel for support group voices to ensure mental health remains an advocacy priority for the GUaRD community.

The GSNV acknowledges and thanks all presenters for giving of their time and expertise to deliver such valuable workshops for our Rare Disease community.

Workshop Attendee Overview



Understanding Vicarious Trauma – *The Caregiver's Journey*
OCTOBER 9TH
4 Attendees

The Accidental Counsellor
OCTOBER 14TH
10 Attendees

Trauma Informed Care Within the Diagnostic Environment
OCTOBER 17TH
30 Attendees

RareNET Forum
OCTOBER 23RD
16 Attendees

Building and Managing Effective Online Support Groups
OCTOBER 30TH
22 Attendees

SECTOR AND CONSUMER REPRESENTATION



Committees and Engagement - Current

Director – Rare Disease International
Board Member (Treasurer) Asia Pacific Alliance of Rare Disease Organisations (APARDO)
Department of Health Victoria – Health Information Sharing Management Committee
Taskforce Member - International Rare Diseases Research Consortium (IRDiRC) Framework to assess impacts associated with diagnosis, treatment, support, and community integration
Working Group Member – HTAi Rare Disease Working Groups x 2 Understanding the Global HTA environment
Member GenSCAN Ethics, Legal and Social Issues (ELSI) Working Group (NBS) GenSCAN Operating Committee
Steering Committee member – HTAi Developing Countries working group
Member - RDNow Undiagnosed Disease Program – Consumer Engagement Committee
Chair – Genomics Industry Alliance (InGeNA) Community Advisory Group
Chair - Embedding Genomics Project Advisory Group
Steering Committee Member – Join Us Research Register
Member – Carrier Screening Reference Group Australia
Member – Patient Voice Initiative Advisory Group
Member – Victorian Genomics Clinical Advisory Group
Member - Deakin University Faculty of Health - Consumer and Research Network
Consumer Member – HGSA-RCPA Carrier Screening Clinical Guidelines Group
Member - Undiagnosed Disease Network – Australia (UDN-AUS) Community Advisory Group
Member – MCRI Expert Reference Group - Consumer Engagement in Research
Member – LINEAGE International Advisory Group (Genomic data usage)
Member – Expert Working Group - Genomics acceptability - societal preferences, public opinion and analysis
Member - LINEAGE Community Advisory Group (Targeted project)
Member - Consumer Involvement in Research Network at MCRI
Member - James Lind Alliance Priority Setting Partnership for Vision Loss Steering Committee
Member - Melbourne Genomics Health Alliance Statewide Paediatric Project Consumer Advisory Committee
President - Communities Council on Ethnic Issues
Member - Eastern Health Community Advisory Committee
Member- MCRI accessibility working group

Research – Within 2 years

Director - Rare Disease International
Associate Investigator - Australian Genomics, Involve Australia
Associate Investigator - Exploring the lived experiences and needs of peer support providers for couples and/or individuals who have received a prenatal variation in sex characteristics test result.
Chief Investigator - Ethical governance for clinical and genomic datasets. (LINEAGE)
Associate Investigator - "The Epi-Genetics Newborn screening (EpiGNs) Program
Associate Investigator - Living with a Genetic, Undiagnosed or Rare Disease: a longitudinal journaling study through the COVID-19 pandemic".
Primary Investigator – Masters Project Exploring the Expectations of People Accessing Genetic Services
Chief Investigator – Embedding Genomics in Primary Care
Primary investigator Masters project :Exploring attitudes and experiences of genetic health professionals towards online genetic support groups

2024 SUPPORT GROUP ENGAGEMENT SURVEY



In 2024, with the implementation of The GSNV's Strategic Plan 2024-2026, the Support Group Engagement Survey demonstrated the organisation's key strategic goals of connecting community and stakeholders and, ensuring a strong agile GSNV and support sector.

Key Objectives:

Build a positive relationship with Support Groups

Build a two-way communication channel between the GSNV and Support Groups

Implementation of The GSNV's Support Group Engagement Survey is an opportunity for the organisation to improve engagement with its Support Group Leaders (SGL) stakeholders.

As an advocacy organisation the survey outcomes highlight that increasing the awareness of The GSNV to this stakeholder group must be made a priority to ensure improved reach and targeted campaigns showcase the advocacy, education and capacity building it aims to achieve for its diverse community.

There was a total of 49 participants of the survey who responded to questions:

SUMMARY OF FINDINGS

The data provides the basis for recommendations and future directions for the GSNV.

- The data whilst small, shows the diversity of the Support Group environment.
- Diversity and inclusion were areas of focus for some Support Groups with many interested in learning more about what the GSNV is doing in this space.
- When comparing the feedback from different services and communication channels offered by the GSNV, the eNewsletter was the most engaged platform followed by the Website/Genetic Link.
- For the most part, when people did not engage with an activity offered by the GSNV it was because they were unaware of it.
- When people did engage the feedback was generally positive with people finding the information relevant and useful.

As a Fit for Practice Student, the development of this project set the foundation for the hours I spent with The GSNV. Having clear timelines and objectives meant I had a structure to work from and enabled me to do many tasks independently.

Although challenging at first, the autonomy offered by The GSNV's Project Lead, Daniele Noel allowed me to take control of the project and ultimately means I am leaving feeling like I have made a valuable contribution to the team, and the sector more broadly.

MIKAELA PRICE - STUDENT EXPERIENCE

Overall, the participants had positive feedback about the responsiveness and initiatives of the GSNV and felt their voice was heard. There were several ideas provided by participants about how the GSNV can better support and provide value to them. The most common responses related to financial guidance.

The GSNV still has work to do to engage and build trust with its members and implement targeted communication strategies that support increase uptake of the survey.

RESEARCH INVOLVEMENT





Susie Roczo-Farkas

Research plays a vital role in shaping the health journeys of people living with undiagnosed, rare, and genetic conditions.

In 2024, GSNV strengthened its commitment to meaningful research involvement - participating as investigators, Community Advisory Group members, and trusted engagement partners. We recognise that impactful research begins with, and belongs to, the communities it aims to serve. Through our work in governance, co-design, and national initiatives, we helped ensure that research not only informs healthcare but becomes part of a more connected, compassionate, and patient-led system. By embedding lived experience at every stage, we're helping to transform research into a tool for advocacy, equity, and lasting change.

Project: LINEAGE Study

Community Perspectives on Genomic Data Governance

(Funded by Medical Research Future Fund [MRFF] [2022-2027])

LINEAGE Study

LINEAGE unites legal, ethical, scientific, and community expertise to develop a national framework for the ethical and responsible governance of genomic data in Australia.

Under the leadership of Monica Ferrie (Chief Investigator) and Susie Roczo-Farkas (Project Lead) at GSNV, the Patient Perspectives team ensures that the needs and perspectives of the community are heard, respected, and influential in shaping the future of genomic governance. Alongside Alison McIvor (SWAN), Emma Bonser (GAA), and Heather Renton (Founder and former CEO of SWAN), the GUARD collaborative is empowering individuals affected by genetic, undiagnosed, or rare diseases (GUaRD) to be actively involved in this study, which brings together bioethics scholars, legal academics, social scientists, clinical geneticists, genomic scientists, First Nations Scholars, and others.

Why it matters:

Patients and the public are most affected by genomic data governance but have historically had limited influence. The GUARD collaborative has consistently advocated for a stronger public and patient voice. However, as Susie identified, there is a deeper need for genuine, embedded community involvement. Representation alone cannot fully capture the complexity of lived experience, particularly in a field as personal and evolving as genomic data.

Best practices in genomic governance require more than consultation; they necessitate placing community members at the core of decision-making processes. With coordinated support from GUARD, Australians—including the GUaRD community and the broader public—are not only informed but also empowered and engaged as long-term contributors, ensuring their voices are more than token representation.

IMPACT

Community-driven insights are shaping a governance framework that is transparent, inclusive, and aligned with public values

INNOVATION

Community members are empowered to actively participate with GUARD's support, moving beyond mere representation. They are given the autonomy to ask questions, shape priorities, and contribute directly to the study.

Embedding Community into LINEAGE

Susie developed and submitted a targeted proposal to reposition the Patient Perspectives stream within LINEAGE in response to this need. Rather than representing the community, the revised model brought them into the study as collaborators—designing and leading inquiry based on their own questions and priorities. This marked a strategic shift from advocacy to embedded leadership. With support from GUARD, the community now contributes directly to the study, ensuring decisions are informed by lived experience and aligned with public values.

From Proposal to Practice

The proposal was endorsed in mid-2024, leading to the establishment of the LINEAGE Consumer Advisory Group (CAG) by year's end. Members include individuals with lived experience of rare diseases, those with varying health literacy, and others without prior genomics exposure. This diversity ensures the insights shared are grounded, accessible, and reflective of the broader public.

A Growing Influence Within LINEAGE

By the end of 2024, the CAG had begun co-designing a research study exploring trust, communication, and genomic data's ethical, legal, and social issues. This work, informed by SWAN's research into parental views on data sharing, will support national conversations about public expectations in genomics. The CAG is now acknowledged as a vital consultation resource across LINEAGE project teams. Its insights will directly influence how data governance is developed and implemented, ensuring it is scientifically sound, socially responsive, and aligned with community expectations.

Useful links:

Further details about the CAG and its activities can be found at https://guard.org.au/lineage-project-guard-consumer-advisory-group-cag/

More information on LINEAGE: https://lineagestudy.org.au/

Project: Epi-Genomic Newborn screening (EpiGNs) Program

(Funded by the MRFF Genomics Health Futures Mission [2022-2027])

EpiGNs is examining if conditions linked to intellectual disability, autism, severe obesity and seizures can be identified as part of the heel prick test performed in the first year of life of 100,000 Victorian infants. Detecting these conditions from birth can lead to life-altering effective treatment and care pathways, which are not possible without early diagnosis.

In addition to the conditions currently detectable by the heel prick test in newborns, such as cystic fibrosis and hypothyroidism, the EpiGNs program is using a new approach to identify changes in the activity of genes. This is helping us to detect developmental conditions including Fragile X, Prader Willi, Angelman, Dup15q, Turner, XXY, XXXY and XXYY syndromes.

With the exception of Prader Willi syndrome, most conditions don't typically show physical signs at birth. That means early detection can prevent delayed diagnosis, lower medical expenses, and reduce stress and anxiety for families.

Finding Fragile X syndrome early is also crucial. It not only helps the affected infants by providing early access to treatments but also their mothers, who will be at high risk of having more affected pregnancies before the first newborn is diagnosed. Typically, children with Fragile X are diagnosed after they turn three years old. By that time the family may have more affected children, which can be avoided through opportunities for informed pregnancy choices.

Prader Willi and Turner syndromes can be treated effectively from infancy and there are clinical trials for gene therapy for Angelman syndrome.

Current medical treatments may also help with serious underlying conditions like intellectual disability, autism and obesity that can occur with these disorders. However, more data on the real-time costs and effectiveness of the EpiGNs program is needed for these conditions to be included in the newborn screening programs using the heel prick test.

Project: 'About Us' Study (Commissioned by GUARD Collaborative Australia, funded by generous industry sponsors and conducted by the Centre for Consumer Directed Research (CCDR).

Innovation

Commissioned by GSNV and delivered by CC-DR using their PEEK (Patient Experiences, Expectations, and Knowledge) methodology, the About Us study explored the lived experiences of Australians with rare and genetic conditions. By comparing these journeys to more common chronic conditions, the study mapped the full continuum of care—from first symptoms and diagnosis to treatment access, communication with healthcare providers, support needs, and impacts on quality of life. Both individuals and carers were included, offering rich, layered insights into what it means to navigate the Australian healthcare system with a rare disease.

Impact

In 2024, the study's findings were shared at two major events:

Rare Disease Day 2024, hosted by GSNV, was the perfect in-person event to launch the data to support group leaders and the wider community. The official launch spotlighted key themes—diagnosis delays, communication breakdowns, fragmented care, and resilience—grounded in real-life stories.

HGSA 2024 Annual Scientific Meeting (Gold Coast) offered a professional hub to engage clinicians, researchers, and industry stakeholders. Susie Roczo-Farkas presented two posters, generating significant interest in the healthcare journeys of rare disease families and their broader implications for practice and policy.

Together, these platforms translated complex, qualitative data into actionable insights—connecting community perspectives with clinical and research agendas.

Next Steps

In 2025, the findings will be adapted into a practical resource for support groups. Designed for advocacy and education, this tool will ensure the study has a lasting impact by amplifying voices, informing service design, and anchoring future research in community-identified priorities.

Upskilling: Family Engagement in Research (FER) Course

(Offered by Healthy Trajectories in partnership with CanChild Centre for Childhood Disability Research, McMaster University; Susie's training funded by LINEAGE).

In 2024, Susie completed the Family Engagement in Research (FER) course as part of her professional development to strengthen her ability to engage meaningfully with community members in research and healthcare settings. The course brought together people with lived experience, peer support leaders, clinicians, allied health professionals, researchers, and industry to explore best practices in co-design—focusing on respectful engagement, cultural responsiveness, and creating psychologically safe spaces when working with community members.

Susie participated in the course as a representative of the Genetic Support Network of Victoria (GSNV), which provided an opportunity to raise awareness of the GUaRD community and highlight the broader work GSNV is doing to champion patient and family perspectives in the rare and genetic conditions space. The course offered a valuable, broad understanding of how different stakeholders experience and contribute to healthcare—making it a recommended experience for anyone involved in supporting patients or shaping their journey.

As part of a collaborative project team, Susie co-developed a Knowledge Translation (KT) tool—a practical resource designed to support clinicians/researchers who wish to work with the GUaRD community and community members seeking to engage in research. The toolkit streamlines the setup of co-designed projects, offering clear guidance, ethical considerations, and structured processes before, during, and after engagement.

Importantly, the course also inspired a shift in how the Patient Perspectives team contributed to the LINEAGE study. Drawing on her learnings, Susie led the formation of a Consumer Advisory Group to bring patients and the public directly into the study. It also deepened her understanding of the importance of recognising and reimbursing community members for their time, input, and expertise.

KT tool, Family Engagement Pathway in Rare Disease: https://healthy-trajectories.com.au/resources/family-engagement-pathway-in-rare-disease-research/

THE SCIENCE WITHIN US

An implementation of Genomics In Schools



Hollie Feller

On 27th November 2024, the GSNV launched The Science Within Us – an education resource for Australian primary and secondary school students.

The unique resource was piloted by small number of teachers across independent and government schools. Evaluation under the Healthy Trajectories Grant and the summary of findings will be available mid 2025. Preliminary evaluation confirms the program is easy for teachers to access and use and effectively increases student understanding of genetic concepts. The inclusion of lived experiences of children within our community gives "students an actual real -life context of what they are learning about"

gsnv Genetic Support Network Victoria

Australian Genomics

MEDIA RELEASE

A new approach to a transformative science:

Connecting genomic concepts to everyday life.

A ground-breaking education resource for school students is set to roll out nationally after the successful uptake by 320 teachers in Victoria.

The Science Within Us is a digital program that can dovetail with existing school curricula, improve students' genetic literacy and understand real-world application of genetics through powerful human stories.

It was designed by the Genetic Support Network of Victoria (GSNV) in collaboration with Australian Genomics to help students understand an area of science that is undergoing massive transformation and impacting daily life.

The Science Within Us

A digital learning resource exploring our genetic identity

The program was initially developed for Year 11 and 12 students in Victoria. GSNV Project Manager Hollie Feller said the feedback from teachers during the pilot phase was a positive sign.
"The first stage of the project was downloaded by more than 320 teachers in Victoria, with a student reach of some 55,000," she said
"That response sent a clear signal to us that the demand for such a resource was there and that we could extend it into lower secondary years and primary school."

Now expanded to cover Years 5 to 12 nationwide, **The Science Within Us** is designed to link with existing curriculum areas such as science, health, humanities, and personal development. Its content emphasises social inclusion and diversity in health, as well as the positive impacts of genomic technologies and advancements.
"Genetics can be complex", says The GSNV Chief Executive, Monica Ferrie. "What sets this program apart from others is that we start with the basics, like understanding heredity traits and how similar we are to each other.
"And, as the program integrates real-world case studies, students are shown how these scientific concepts connect them to everyday life."

With a mixed-modality approach, The Science Within Us offers equitable access to learning via an online portal for all students, regardless of location, to easily engage with the content.

Australian Genomics Managing Director Tiffany Boughtwood said introducing students to the science and real-world impact of genetics would help them understand a fascinating area of human health.
"The Science Within Us is such an innovative approach that could set students up for careers in a rapidly evolving field and create pathways for the future workforce,"
Ms Boughtwood said.

Educators and school leaders are encouraged to register to use the free resource by visiting www.thesciencewithinus.org.au

About The GSNV

The Genetic Support Network of Victoria (GSNV) was established in 1997 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.

For more information or to arrange an interview, please contact:
Monica Ferrie, CEO GSNV Monica.ferrie@vcgs.org.au



gsnv

gsnv
Genetic Support
Network Victoria

THANK YOU

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

We are especially grateful for the ongoing support of the Department of Health (DoH), who continue to provide funding and support for our important work. We thank in particular Marianne Griffin and Sonia Caruana for an ongoing supportive relationship with the GSNV and their wisdom and counsel.

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, industry, researchers and so many others. A special mention to Melody Menezes for her ongoing support and generous time.



VICTORIA State Government | Department of Health

FINANCIAL REPORTS

GSNV - 2024 High Level Summary

OPERATING ITEM	YTD through Dec-24 CONSOLIDATED $ - 000's
VIC GOVT GRANT	258.6k
OTHER INCOME	121.0k
TOTAL REVENUE	**379.6k**
SALARIES & RELATED COSTS	238.0k
TOTAL EMPLOYMENT COSTS	**238.0k**
COMPUTER SOFTWARE & EXPENSES	1.8k
COMPUTER HARDWARE	-
PRINTING, STATIONERY & PHOTOCOPYING	1.4k
CATERING	2.0k
OTHER ADMINISTRATIVE EXPENSES	127.0k
TRAVEL	1.4k
OTHER EXPENSES	1.5k
STAFF TRAINING & CONFERENCES	1.2k
TOTAL OPERATING EXPENSE	**374.3k**
OPERATING SURPLUS / (DEFICIT)	**5.3k**
TOTAL DEPRECIATION AND INVESTMENTS	-
TOTAL SURPLUS / (DEFICIT)	**5.3k**