gsnv
Genetic Support
Network Victoria

GENETIC SUPPORT NETWORK OF VICTORIA

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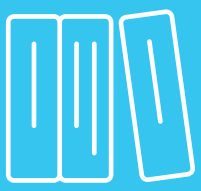
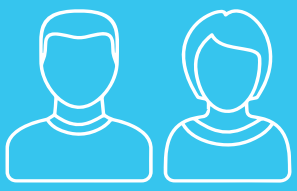
A YEAR IN REVIEW 2021



AWARENESS & EDUCATION



CARE & SUPPORT

RESEARCH & DATA

2021 IN CONTEXT

2021 continued to challenge with the uncertainty and pandemic fatigue that accumulated as the year continued. Many individuals, families and communities have been stretched to simply survive. As always, extraordinary times, highlight the best and worst of humanity. Patient Support Groups are most definitely amongst the best of humanity. Patient Support Groups have worked tirelessly to support their communities, to advocate and champion change, equity of access, and to highlight the challenges for people living with genetic, undiagnosed and rare disease in a time totally crowded by pandemic news. I am endlessly humbled and grateful for the commitment, support and advocacy delivered with and for our genetic, undiagnosed, and rare disease community. The GSNV is proud to serve and support our sector.

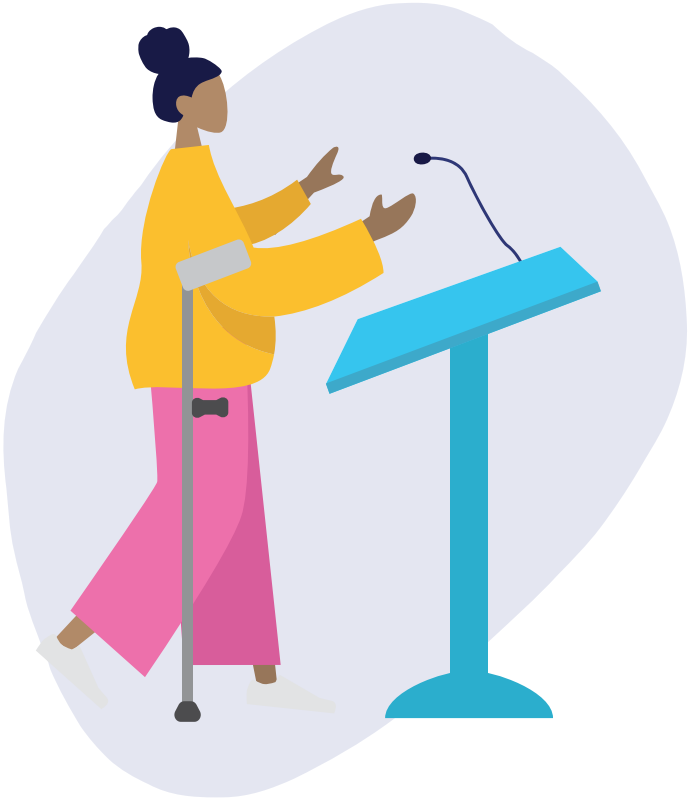
We have now been working on the implementation of the Federal Department of Health National Strategic Action Plan for Rare Diseases since launch in February 2020, almost two years ago. We would like to thank the outgoing Federal Health Minister, the Honorable Greg Hunt MP for his commitment to improving the lives of our community through many initiatives and importantly, investment. We would also like to acknowledge the important reviews and inquiries over the past year, and we look forward to working with all stakeholders on implementation in 2022 and beyond.

I am delighted to share our 2021 Year in Review to provide a snapshot of how we at the GSNV have contributed to the delivery of the plan objectives this year. This review also flags some of our project priorities for 2022.

We have chosen to present our work within the National Strategic Action Plan for Rare Diseases framework. The plan is also consistent with the National Genomics Health Policy Framework and the Genetic and Genomic Healthcare in Victoria 2021. The GSNV Projects are focused on Victoria but can reach a national audience through GUARD Collaboration Australia.

As always, there is much to do, and we will continue to focus on serving collaboratively and through consultation. We have included a list of connections, networks, and collaborations as part of this report. We hold ourselves accountable to our community, collaborating partners and the Department of Health in Victoria.

The GSNV continued to collaborate purposefully as part of the GUARD Collaborative Australia in 2021, (GSNV, SWAN and GAA) to form a joined-up alliance ensuring resources are maximized, voices are joined and heard and our sector remains nationally strong and represented. GUARD Collaborative Australia has a strategic plan and works towards common goals and objectives for our community nationally, a GUARD snapshot is included after the GSNV review. Further information can be found at GUARD.org.au



Pillar 1: AWARENESS & EDUCATION

1.1 Increase every Australian's awareness of rare diseases including, where applicable, relevant protection measures.

GSNV Projects Delivered:

• ***Genomics in Schools*** project launched for delivery into Victorian Schools in Term 3, 2021 and resources downloaded by more than ***150 teachers*** across metropolitan, regional, and rural Victoria. Estimated reach – ***44,000 Victorian students***.

• ***#MyGenesandMeProject*** – community awareness campaign launched to build health literacy about what genetics means to individuals.

• A library of lived experience was made available to continually ensure we are ***'Putting Patients at the Centre'*** - building awareness of the issues and experience faced by patients and families in the health system and accessing services.

• Getting to know your electorate project – introducing ***Victorian MP's*** and ***Federal MP's*** with Victorian electorates to their genetic, undiagnosed and rare disease community and support groups within their electorate.

• Presentations to stakeholders across ***Government, Industry, Researchers, Agencies***.

We are committed to introducing every Australian to the 2,000,000 Australians living with genetic, undiagnosed, and rare disease.

1.2 Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.

GSNV Projects Delivered:

We are committed to improving the experience of patients and families navigating the health and social services systems.

• ***GSNV Website*** and ***Genetic Link*** updated to ensure ease of access to information.

• ***Directories added to Genetic Link*** with currency maintained for searching support groups linked to genes and conditions, searching support groups and resources for health professionals, and searching support for patients, families and carers.

• ***Delivery of expert webinar*** for our community answering ***COVID-19 vaccination*** questions.

• ***Communications*** – Bits and Pieces, What's On the GSNV, GSNV YouTube, LinkedIn and other Social Media.

• ***Building and sharing evidence base*** through surveys, international, national and local experience.

• ***Patient Pathways Project*** – access to case management telehealth nurse.

• ***Share for Rare Program of Work:***

- ***Support Group Lifecycle Project*** resources available through Genetic Link.
- Collecting and sharing ***Best Practice Resources*** from support groups on Genetic Link.

1.3 Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.

GSNV Projects Delivered:

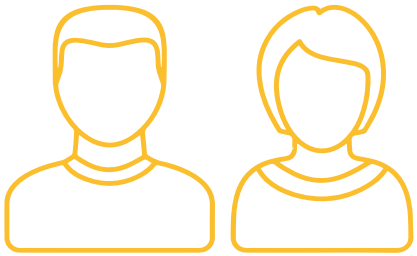
The current and future genetics and genomics workforces must understand the needs and expectation of our community. We seek to both influence the current and future workforce with knowledge, information and awareness.

• ***Masters in Genetic Counselling*** students participate in ***Fit for Practice placements in Patient Support Groups***.

• ***Masters of Genetic Counselling and Masters in Genetic Health*** student projects supervision and presentations.

• ***Working Group membership of InGeNA*** — National Genomic Industry Alliance workforce project team. Delivering a report on the skill and knowledge competencies required in industry to support genomic advancement.

• Organisation and delivery of ***professional development sessions*** for GSNV volunteers.



Pillar 2: CARE & SUPPORT

2.1 Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family centred.

It takes a village to support people living with genetic, undiagnosed and rare disease.
It takes a committed and coordinated community to support the village.
We are ***focused on supporting*** individuals, families, support groups and villagers.
- By ***engaging with our community*** to listen to and identify needs.
- By ***empowering our community*** to seek and find what they need to support the best lives they can live.
- By ***strengthening the sector*** through support, tools, resources, volunteers and advocacy.
- By ***advocating for simple and connected pathways*** for patients through representation and engagement.
- By ***educating sectors*** about what the needs and expectations of our community and collaborating to deliver them.

We are focused on supporting individuals, families, support groups and villagers by engaging, empowering, strengthening, advocating for and educating.

GSNV Projects Delivered:

• ***Strategic Representation*** and ***engagement*** in health, research, social services and government.

• ***Genetic Link*** – Includes the ***Support Group Life Cycle Resource Bank*** and resources to inform health professionals seeking to find resources and supports for patients.

• Access to the ***Patient Pathways Program*** – a nurse acting as case manager accessed through telehealth. This provides practical navigation assistance and support.

• ***Share for Rare*** – Bank of experts for support groups to access skills they need.

• Access to ***Rare Connect Network*** through GSNV website.

• Events – ***Rare Disease Day***, ***Who's on Your Team workshop*** highlight who is needed along the patient journey and patient support team.

• Availability of a ***database of psychologists*** with expertise to support patients and families in our community available for clinicians for referral purposes.

• Comprehensive sector ***database of support organisations*** – accurate resource for clinicians and patients – maintained for accuracy and currency.

• Active and high performing ***volunteer base***.

• Activated the ***Mental Health Working Group*** – a national collaboration that has developed a strategic plan and priority actions specifically to serve our community.

• Understanding needs and priorities through COVID-19 and long-term effects to inform care requirements – ***C19Journals project*** has developed its first report and has been accepted for publication. Phase 2 – ***Interventions to Live Well*** is underway.

• Engaging with our community creating regular ***engagement opportunities***, sharing knowledge and seeking input through ***consultation and transparency***.

2.2 Ensure diagnosis of a rare disease is timely and accurate.

GSNV Projects Delivered:

• Strategic Representation, advocacy and relationships including ***Genetics Clinical Hubs***, ***Victorian Genomics Clinical Advisory Committee***.

• Relevant ***representation and engagement*** including the ***Parliamentary Inquiry*** into approval processes for ***new drugs and novel medical technologies*** in Australia, C***onsumer Consultation Committee for MSAC and PBAC***, submissions and letters of support.

• Active engagement and advocacy in ***newborn screening discussions*** and active research to develop a collaborative strategy to address ***access*** and ***implementation*** in Victoria.

• Engagement in ***Mackenzie's Mission Project*** through ***Mackenzie's Mission Engagement Reference Group***.

• Engagement in ***RDNow Project – Undiagnosed Rare Disease*** Project at the Royal Children's Hospital through Consumer Engagement Committee.

• Engagement with ***international projects*** – Global Commission to end the Diagnostic Odyssey.

• Submission to ***National Medicines Policy*** review.

We are committed to equity of access to testing and the best chance of early and accurate diagnosis for everyone.

2.3 Facilitate increased reproductive confidence.

GSNV Projects Delivered:

• Active participation in ***National Carrier Screening Reference Group.***

• Promoting ***carrierscreening.org.au***

• Representation as part of ***Human Genetics Society of Australasia*** and ***Royal College of Pathologists carrier screening implementation group***.

• Submissions to ***MSAC applications*** and ***targeted consultation requests*** for access to testing and screening.

The GSNV supports equitable and informed access to carrier screening and testing in pregnancy to support reproductive choices.

2.4 Enable all Australians to have equitable access to the best available health technology.

GSNV Projects Delivered:

Health technologies are evolving rapidly and the GSNV is committed to equitable and safe access to health technologies. We seek to inform our community about the developing technologies, the challenges of implementation, the ethical questions and active engagement in the processes of health technology assessment.

- Relevant representation and engagement including the ***Parliamentary Inquiry*** into approval processes for ***new drugs and novel medical technologies*** in Australia, Therapeutic Goods Australia review, Consumer Consultation Committee for MSAC and PBAC, submissions and letters of support.

- ***Strategic representation*** in particular – Clinical Hubs, Victorian Genomics Clinical Advisory Committee, Consumer Health Forum, InGeNA and Australasian Institute of Digital Health.

- Participated in national advocacy for ***Newborn Screening implementation of Spinal Muscular Atrophy***.

- ***Research and advocacy*** informing the newborn screening application process.

- Actively engaged in ***InGeNA Access and Equity Workgroup*** focusing on health technology.

- Engaged in establishment of ***Involve Australia***, developing ***pathways to inclusive and equitable engagement*** in genomic research.

- Participating and engaging in what's happening ***internationally, nationally and locally***.
    - Board member of the ***Asia Pacific Alliance of Rare Disease Organisations***.
    - Engagement in the ***WHO Collaborative Global Network for Rare Diseases***.
    - ***GUARD Collaborative Australia***.

2.5 Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

GSNV Projects Delivered:

We are committed to holistic healthcare. This includes mental health and the importance of connections and support as part of the care continuum.

- Establishment of ***Mental Health Working Group*** for our Community and the establishment of a ***series of mental health priority projects***, responding to the recommendations of the ***Victorian Royal Commission into Mental Health***.

- Delivery of ***Practical Wellness month*** of activities including ***Accidental Counsellor***, ***Peer Support Training***, ***Grief and Loss webinar*** and ***Right to Safety Program***.

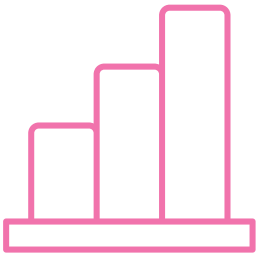
- Delivery of a ***joint webinar series*** (A Day in the Life of...) with the Australian Psychologists Society ***(APS)*** Rehabilitation Psychology for Injury, Chronic Illness and Pain Interest Group ***(RPICI&PIG)***.

- COVID-19 Support – ***C19Journals*** and ***Interventions to Live Well Research Project*** alliance with Australian Genomics.

- ***Genetic Link and resources*** for patients, support groups and health professionals.

- ***Psychologist support database*** available for referral opportunities for clinicians.

- Access to ***Patient Pathways Program***, a telehealth nurse case management assisting with service navigation.



Pillar 3: RESEARCH & DATA

3.1 Enable coordinated and collaborative data collection for facilitating the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.

GSNV Projects Delivered:

- ***Strategic representation*** in particular – Clinical Hubs, Victorian Genomics Clinical Advisory Committee, InGeNA and Australasian Institute of Digital Health.
- Support for ***collaborative development*** of national rare disease registry.
- Engagement with ***JoinUs Research Register, The George Institute Project***.
- Establishment of a ***Rare Disease Registry Directory*** on the Genetic Link.
- Active member of ***InGeNA Data and Technology group*** delivering the ***Genomic Data in Australia report***.

The possibilities of data utilisation and digital technology are exciting and important. We are committed to ensuring that data and technology are optimized collaboratively to deliver more equitable access, more successful outcomes and better lives.

3.2 Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.

GSNV Projects Delivered:

- ***Strategic Relationships*** with Australian Genomics, Melbourne Genomics, InGeNA, Australasian Institute of Digital Health, Research Australia.
- Surveys to our community to ***establish gaps and community priorities*** that inform discussion with researchers.
- ***C19Journals*** and ***Interventions to Live Well Project*** partnership with ***Australian Genomics***.
- Participation in ***research prioritisation and applications*** for the ***Medical Research Future Fund*** and ***Genomics Health Futures Mission***.

We support a national collaboration of all relevant stakeholders to identify research priorities, gaps and establish a plan.

3.3 Ensure research into rare disease is collaborative and person-centred.

GSNV Projects Delivered:

- Project Lead with ***Australian Genomics on Involve Australia Project***.
- Support and engagement in research projects such as ***JOIN US***, ***Insurance Equity***.
- Engagement in ***InGeNA alliance***.
- Input into ***Research Australia consultation*** into the future of health and medical research

3.4 Translate research and innovation into clinical care; clinical care informs research and innovation.

GSNV Projects Delivered:

- Publication of ***C19Journal research findings***.
- Involvement in ***Mackenzie's Mission carrier screening research project***.
- Involvement in ***RDNow Project*** – undiagnosed diagnostic research project.
- Collecting data from ***Patient Pathways Program*** to influence the clinical care process.
- ***Masters in Genetic Counselling*** and ***Masters in Genetic Health*** continued ***collaboration***.

2022 WILL BRING

A snapshot of what the new year will include:

In 2022, we will build on our success in implementing the National Strategic Action Plan for Rare Diseases with a continued commitment to collaboration and consultation. We are privileged to often be asked to represent our community, this is a great responsibility and we promise to tell it how we are told, to listen and share and to work together with all stakeholders to positively impact the quality of life for our community.

Projects

- Launch of Parliamentary Friends of People Living with Genetic, Undiagnosed and Rare Disease in Victoria
- Critical Conversations – Commence project to develop resources; what to say and ask, when for Patients, Families and Health Professionals.
- Ageing with Genetic, Undiagnosed and Rare Disease Working Group to identify these issues and required actions
- New Lived Experience Series to continue to give voice to our community
- Continued collaboration with our community and stakeholders
- Release of Interventions to Live Well outcomes
- Phase 2 of Genomics in Schools to build genomics literacy, workforce interest and promote exploration of ethics, family history and choice
- Masters Project exploring the expectations of Genetic Counsellors compared to the expectation of patients to understand gaps
- Delivery of webinar series on advanced therapies to grow community understanding

Engagement

To keep ourselves informed and to ensure information and activity is current, relevant and doesn't duplicate, we are engaged purposefully as active members/associates/collaborators of and with:

- Our community; people living with genetic, undiagnosed and rare conditions and those who support them
- All Victorian Clinical Genetics Services
- Department of Health – Victoria
- Department of Health - Federal
- GUARD Collaborative Australia
- Genomics Clinical Advisory Group - Victoria
- Australasian Institute for Digital Health – Precision Healthcare Steering Committee
- Human Genetics Society of Australasia
- Australian Society of Genetic Counsellors
- Australian Genomics
- Melbourne Genomics
- InGeNA - Genomics Industry Alliance – Steering Committee consumer representative
- InGeNA – Project Committee Member, Access and Equity, Workforce development and Data and Innovation
- Mackenzie's Mission Engagement Reference Group - Chair
- RDNow Consumer Engagement Committee
- Australian Patient Organisations Network – Chair Steering Committee
- WHO Collaborative Global Network for Rare Diseases (CGN4RD) – Expert Panel Member
- Rare Disease International (RDI)
- Asia Pacific Alliance of Rare Disease Organisations (APARDO) – Board Member
- Royal College of Pathologists Lay Committee (RCPA) - Lay Committee Member
- Carrier Screening Reference Group
- HSGA-RCPA Carrier Screening implementation working group
- Deakin University Faculty of Health - Consumer and Research Network
- Research Australia
- Consumer Health Forum
- Rare Voices Australia
- Victorian Council of Social Services
- Women With Disabilities Victoria
- The George Institute – Join Us Project Steering Committee
- Rehabilitation Psychology for Injury, Chronic Illness and Pain Interest Group
- Involve Australia Project Lead – Australian Genomics

GUARD Collaborative Australia

Review - 2021

GUARD Collaborative Australia is a collaboration of peak body organisations; ***Genetic Support Network of Victoria***, ***Genetic Alliance Australia (NSW)*** and ***Syndromes Without A Name (SWAN) Australia***.

GUARD members work independently of each other fulfilling their own responsibilities and roles and come together to provide systemic advocacy and support for people living with genetic, undiagnosed and rare conditions and those who support them.

Our advocacy is driven by values of fairness, equity and quality of life and focusses on change in the health and disability sectors.

Together we offer our united collaborative strength and provide assistance, support and services for genetic, undiagnosed and rare condition support groups, their members and the wider community seeking to influence and change current health, mental health, support services and disability policy and practice impacting our community.We strive for a fair, equitable and collaborative approach to health and wellbeing for all members of our population.

The work of GUARD is consistent with the National Strategic Action Plan for Rare Diseases, national and state genomics and precision health policies and individual organisation objectives.

This snapshot will be presented utilising the pillars of the National Strategic Action Plan for Rare Diseases.

Pillar 1: AWARENESS & EDUCATION

1.1 Increase every Australian's awareness of rare diseases including, where applicable, relevant protection measures.

1.2 Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.

1.3 Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.

GUARD Projects Delivered:

• ***GUARD Website*** implemented making available national information, sharing national resources and linking to state based services.

• A ***GUARD Facebook Group*** is moderated and accessible.

• Establishment of ***Community Advisory Group*** to inform the work of GUARD Collaborative Australia.

• Development of a ***Landmark Australian Patient, Family, Carer and Survey Group survey*** for 2022.

• Consultation for national submissions on ***NDIS Independent Assessments and legislation***, ***Parliamentary Inquiry*** into approval processes for ***new drugs and novel medical technologies*** in Australia, ***National Medicines Policy***, targeted consultation for ***MSAC*** and ***PBAC*** applications.

• Advocacy on ***newborn screening application*** assessment and implementation.

Pillar 2: CARE & SUPPORT

2.1 Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family centred.

2.2 Ensure diagnosis of a rare disease is timely and accurate.

2.3 Facilitate increased reproductive confidence.

2.4 Enable all Australians to have equitable access to the best available health technology.

2.5 Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

GUARD Projects Delivered:

- ***Patient Pathways Project Collaboration*** providing a case manager telehealth nurse for people living with genetic, undiagnosed and rare conditions.
- Representation on ***Genomics Industry Alliance (InGeNA) – Access and Equity Project***.
- Input into Industry submissions to ***National Medicines Policy review***.
- ***Mental Health Working Group*** support and engagement.

Pillar 3: RESEARCH & DATA

3.1 Enable coordinated and collaborative data collection for facilitating the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.

3.2 Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.

3.3 Ensure research into rare disease is collaborative and person-centred.

3.4 Translate research and innovation into clinical care; clinical care informs research and innovation.

GUARD Projects Delivered:

- Representation on ***Genomics Industry Alliance (InGeNA) – Data and Innovation Project.***