Introduction

Our extraordinary times continue, as does the amazing work of Patient Support Groups. I am 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. I am delighted to share our 2021 Mid-Year Review to provide a snapshot of how we have contributed to the delivery of the plan objectives to date this year. This midyear review also includes a look at our project priorities for the balance of 2021.

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 Collaborative 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.

We continued to collaborate purposefully as part of the GUARD Collaborative Australia, (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. As GUARD Collaborative Australia has a separate strategic plan and program of work and is working towards common goals and objectives for our community nationally, a GUARD snapshot is included after the GSNV review.

PILLAR 1 – AWARENESS AND EDUCATION

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

GSNV Current Work and Projects Delivered

- Genomics in Schools project to be launched for delivery into Victorian Schools in Term 3, 2021
- Teachers Guide and resources completed including health professional and lived experience videos and discussion props.
- #MyGenesandMeProject – community awareness campaign launched to build health literacy.
- Putting Patients at the Centre project launched through social media with lived experience vignettes – building awareness of the issues and experience faced by patients and families in the health system.
- Getting to know your electorate project – introducing Members of Parliament to support groups within their electorate and some localised statistics regarding rare disease populations and support groups.

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.

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

GSNV Current Work and Projects Delivered

- GSNV Website and Genetic Link updated to ensure ease of access to information
- Directories added to Genetic Link 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 at the GSNV, GSNV YouTube, Linked-In and other Social Media
- Building and sharing evidence base through surveys, international, national and local experience
- Patient Pathways Project – case management telehealth nurse now available for people living with rare disease
- 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

We are seeking to influence and educate the future genetics and genomics workforce about support needs for people with genetic, undiagnosed and rare disease identified by listening to and engaging with our community. Our projects seek to both influence the current and future workforce as well as to actively engage as representatives of patient expectations to inform workforce strategy.

GSNV Current Work and Projects Delivered

- Fit for Practice Placements in Support Groups for students from the Masters in Genetic Counselling
- Masters Projects Student Supervision – Masters in Genetic Counselling and Masters in Genetic Health Internships and Research Projects
- Active membership of InGeNA – National Genomic Industry Alliance workforce project team exploring skill and knowledge expectations and requirements.
- Organisation and delivery of professional development sessions for GSNV volunteers

PILLAR 2 – CARE AND 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

We are focused on supporting people, families and support groups

- 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 the needs and expectations of our community and collaborating to deliver them

GSNV Current Work and Projects Delivered

- Strategic Representation and engagement in health, social services and government sectors
- Fit in Practice Placements in support groups for students from the Masters in Genetic Counselling
- Genetic Link – The Support Group Life Cycle Resource Bank is now available. An Education section has been created to allow access to resources for Teachers
- Share for Rare – Skills sharing bank of experts for support groups to access skills they need
- Access to Rare Connect Network through GSNV website
- Grant identification and writing support through Grant Writing Bootcamp
- Events – Rare Disease Day, Who's on Your Team workshop
- 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
- Convening the Mental Health Working Groups – a national collaboration that has developed a strategic plan and priority actions specifically to serve our community.
- Understanding needs and priorities through COVID-19 – C19Journals project has developed its first report and is seeking publication
- 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

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

GSNV Current Work and Projects Delivered

- Strategic Representation, advocacy and relationships including Clinical Hubs, Victorian Genomics Clinical Advisory Committee
- Relevant representation including the Parliamentary Inquiry into approval processes for new drugs and novel medical technologies in Australia, MSAC and PBS submissions and letters of support.
- Patient Pathways Project – Telehealth nurse case management
- Active engagement 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 through Consumer Advisory Group
- Engagement with international projects – Global Commission to end the Diagnostic Odyssey

2.3 Facilitate increased reproductive confidence

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

GSNV Current Work and Projects Delivered

- Active participation in National Carrier Screening Reference Group
- Promoting carrierscreening.org.au

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

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.

GSNV Current Work and Projects Delivered

- Engage in actions to decrease the gap between the evolution of science and technology and community under standing such as starting conversations and educating our community.
- Strategic representation in particular – Clinical Hubs, Victorian Genomics Clinical Advisory Committee, Consumer Health Forum, InGeNA and Australasian Institute of Digital Health
- Actively engaged in InGeNA Access and Equity Workgroup focusing on health technology assessment
- Staying across 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 (CGN4RD)
- GUARD Collaborative Australia

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

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

GSNV Current Work and Projects Delivered

- Mental Health and Genetic Counselling Masters Project – helping us to understand the current expectations.
- Establishment of Mental Health Working Group for our Community and the establishment of a series of mental health priority projects for our community
- 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 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
- Patient Pathways Program – telehealth nurse case management

PILLAR 3 – RESEARCH AND 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

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.

GSNV Current Work and Projects Delivered

- Strategic representation in particular – Clinical Hubs, Victorian Genomics Clinical Advisory Committee, InGeNA and Australasian Institute of Digital Health
- Commenced exploration of collaborative development of national rare disease registry
- Engagement with JoinUs Research Register. The George Institute Project

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

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

GSNV Current Work and 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
- C19Journals Project partnership with Australian Genomics

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

GSNV Current Work and 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

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

GSNV Current Work and Projects Delivered

- 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

Our Projects for the Second Half of 2021 include:

- Critical Conversations – Commence project to develop resources; what to say and ask, when for Patients, Families and Health Professionals. Delivery 2021
- Care for Rare Program of Work
- Wellness Week including Mental Health First Aid, Peer Support Training, NDIS support event and Right to Safety Program
- Planning for 3 x "A day in the Life of Living with ....." webinars for the APS, RPICI & PIG
- Getting to Know Industry workshop introducing patient support groups to industry groups in pharmaceuticals, health technologies, labs and medical equipment developers.
- Convene an Ageing with Genetic, Undiagnosed and Rare Disease Working Group
- Plan webinar series for APS for 2022
- Develop template for condition specific resource to make available for psychologists and test with at least 1 condition
- Phase 2 of C19Journals project with deeper exploration of themes identified in Phase 1
- Masters Project exploring the expectations of Genetic Counsellors compared to the expectation of patients to understand gaps in 2021 and 2022
- Development of webinar series on advanced therapies to grow community understanding
- Commencement of Involve Australia project
- Completion of Master in Genetic Counselling Fit for Practice Support Group Placements
- Delivery and evaluation of Genomics in Schools program
- Directories and Database development of registries, health professionals and clinical trials
- Continued advocacy in newborn screening

The GSNV has also completed our Annual General Meeting and released our Annual Report. In February 2021, we also released our new strategic plan 2021-2024 after substantial consultation and input from our community and key stakeholders.

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

- Department of Health - Victoria
- 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 and project representative: Access and Equity, Data and Innovation and Workforce Capability
- Mackenzie's Mission Engagement Reference Group - Chair
- RDNow Consumer Engagement Committee
- Australian Patient Organisations Network – Steering Committee Chair
- 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
- Health Navigation Hub
- 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

This report provides an update on the current and project work plan for GUARD Collaborative Australia.

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 and 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 Collaborative Australia Current Work and Projects Delivered

- GUARD Website developed 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
- Consultation for national submissions on NDIS Independent Assessments and Parliamentary Inquiry into approval processes for new drugs and novel medical technologies in Australia
- Advocacy on newborn screening application assessment and implementation

Future Projects

- Survey data collection and analysis
- Report providing an evidence base for all support groups available

Pillar 2 – Care and 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 Collaborative Australia Current Work and 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

Future Projects

- Start a national conversation about multi-disciplinary expertise support and clinics
- Support for the Mental Health Working Group and priority projects

Pillar 3 – Research and 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 Collaborative Australia Current Work and Projects Delivered

- Representation on Genomics Industry Alliance (InGeNA) – Data and Innovation Project, Access and Equity Project, Workforce Project and Measuring the Benefits of Genomics Project

Future Projects

- Implement a project focused on health data literacy and public trust