

## General Comments

This has been an extraordinary year for everyone, the general community has learnt many lessons in what's important; the value of connection, how to live with restricted freedoms and the need for sharing responsibilities and support. These are things that the genetic, undiagnosed and rare disease communities already knew, that are part of everyday lived experience. It's been tough and I'm so proud of the work of the GSNV and the support sector during this year.

The GSNV began working from home in February and is yet to return to our office. We have perfected zoom, gotomeeting, Microsoft Teams, well some of us....This year has tested our flexibility, our responsiveness and our innovation. This report is a snapshot of where we have been and how we have served in 2020. We hold ourselves accountable to our community and the Department of Health and Human Services in Victoria.

As with our midyear review, we have chosen to present our work in the framework of the National Strategic Action Plan for Rare Diseases which inspired so much hope for us and people with genetic, undiagnosed and rare disease and those who support them. The plan is also consistent with the National Genomics Health Policy Framework and the Genetic and Genomic Healthcare in Victoria 2021.

We continued to collaborate purposefully as part of the GUARD Collaboration, (GaRDN, 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. 2021 is going to be a very exciting year for the GSNV and our community. We have developed a new strategic plan which we will share in the new year, with our annual work plan.

# PILLAR 1 – AWARENESS & EDUCATION

## 1.1 INCREASE EVERY AUSTRALIAN'S AWARENESS OF INCLUDING, WHERE APPLICABLE, RELEVANT PROTECTION MEASURES

The GSNV Projects are focused on Victoria but can reach a national audience through our GUARD collaboration with GaRDN, GAA and SWAN and our national networks



### GSNV work & current projects

- **Genomics in Schools** project now to be launched in 2021
  - o Speaking at VCE Science Teachers Conference 2020 and 2021
  - o Teachers Guide and resources completed (95%)
- **#MyGenesandMeProject** – community awareness campaign ready to launch in 2021
- **Mackenzie's Mission Carrier Screening Research Project**
  - o Chair of Engagement Reference Group
- **Carrier Screening Reference Group**
  - o Member of national group

# PILLAR 1 – AWARENESS & EDUCATION

## 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

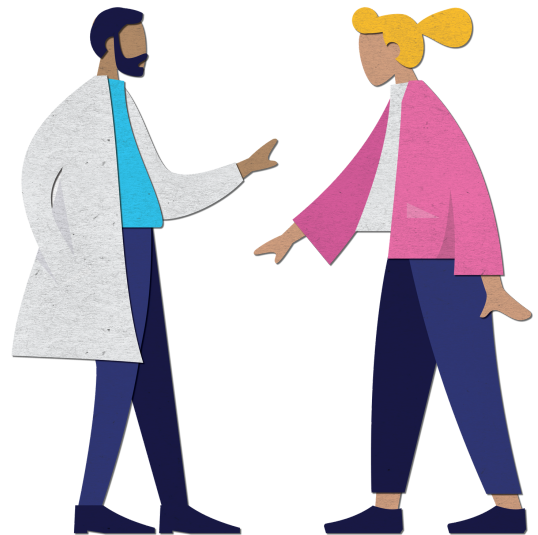


### GSNV work & current projects

- **GSNV Website and Genetic Link updated** to ensure ease of access to information
- **Communications** –Bits and Pieces, Whats On, Social Media
- **Building evidence base through surveys**, international, national and local experience
- **Patient Pathways Project** – case management telehealth nurse now available for people living with rare disease
- **Critical Conversations** – Commenced project to develop resources; what to say and ask, when for Patients, Families and Health Professionals. Delivery 2021
- **Share for Rare Program of Work**
  - o Support Group Lifecycle Project – Delivery 2021
  - o Collecting and sharing Best Practice Resources on Genetic Link

# PILLAR 1 – AWARENESS & EDUCATION

## 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

### GSNV work & current projects

- **Genomics in Practice** Placements for students from the Masters in Genetic Counselling
- Masters Projects Student Supervision – **Masters in Genetic Counselling and Masters in Genetic Health**
- Engagement with **InGeNA** – National Genomic Industry Alliance nominating workforce as a key area of focus and project work
- **Critical Conversations** – What to say, when, for Patients, Families and Health Professionals



## 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 what the needs and expectations of our community and collaborating to deliver them

### GSNV work & current projects

- **Strategic Representation** and engagement in health, social services and government
- **Genomics in Practice Placements in support groups** for students from the Masters in Genetic Counselling
- **Care for Rare Program of Work**
  - o Wellness Week- including Mental Health First Aid, Peer Support Training, NDIS support event and Right to Safety Program
- **Genetic Link** –The Support Group Life Cycle Resource Bank
- **Share for Rare**-Skills sharing project
- **Grant identification and writing support**
- **Events** – Rare Disease Day, Consumers in a Pandemic
- **Critical Conversations** for Patients and Professionals Project
- Develop a **database of psychologists** with expertise to support patients and families in our community
- Comprehensive sector **database of support organisations** – accurate resource for clinicians and patients
- Administration of **Intersex Variation Peer Support Grant**
- Active and high performing **volunteer base**
- Understanding needs and priorities through COVID-19 – **C19Journals project**
- **The GSNV “wants to know” surveys**
- Engagement with the **Pinnacle Support Group** website

## PILLAR 2 – CARE AND SUPPORT

### 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 work & current projects

- Strategic Representation, advocacy and relationships including [Clinical Hubs](#), [Victorian Genomics Clinical Advisory Committee](#)
- Relevant Submissions and support for applications 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
- Engagement in [Mackenzie's Mission Project](#)
- Engagement in [RDNow Project](#) – Undiagnosed Rare Disease Project
- [Rural and Remote Access and Experience Report](#)
- Engagement with international projects – [Global Commission to end the Diagnostic Odyssey](#)

### 2.3 FACILITATE INCREASED REPRODUCTIVE CONFIDENCE

#### GSNV work & current projects

- Development of resource kit for support groups to inform and be able to assist families enquiring about [Mackenzie's Mission carrier screening project](#)
- Engagement in development of patient resources as part of the [national consent project](#)
- Promoting [carrierscreening.org.au](https://carrierscreening.org.au) and the work of the [Carrier Screening Reference Group](#)

# PILLAR 2 – CARE AND SUPPORT



## 2.4 ENABLE ALL AUSTRALIANS TO HAVE EQUITABLE ACCESS TO THE BEST AVAILABLE HEALTH TECHNOLOGY

### GSNV work & current projects

- Strategic representation in particular – [Clinical Hubs](#), [Victorian Genomics Clinical Advisory Committee](#), [Consumer Health Forum](#), [InGeNA](#) and [Australasian Institute of Digital Health](#), [National COVID-19 Clinical Guidelines Taskforce Consumer Panel](#)
- Staying across what's happening internationally, nationally and locally
  - o Board member of the [Asia Pacific Alliance of Rare Disease Organisations](#)
  - o Engagement in the [WHO Collaborative Global Network for Rare Diseases \(CGN4RD\)](#)
  - o [GUARD Collaborative](#)
- [MSAC](#), [PBAC](#) submissions

## 2.5 INTEGRATE MENTAL HEALTH, AND SOCIAL AND EMOTIONAL WELLBEING, INTO RARE DISEASE CARE AND SUPPORT



### GSNV work & current projects

- [Wellbeing Week](#)
  - o Mental Health First Aid
  - o Right to Safety –Women with Disabilities Partnership
  - o Peer Support Training – CFCC Partnership
  - o NDIS Support Session
- [Mental Health and Genetic Counselling Masters Project](#)
- Establishment of [Mental Health Working Group](#) for our Community
- COVID-19 Support– [C19Journals Research Project](#) alliance with Australian Genomics
- [Genetic Link](#) and resources
- [Psychologist support database](#) development
- [Patient Pathways Program](#) – telehealth nurse case management
- An introduction to [Violence Against Women with Disabilities](#) Webinar in September

## 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

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



#### GSNV work & current projects

- Strategic representation in particular – [Clinical Hubs](#), [Victorian Genomics Clinical Advisory Committee](#), [InGeNA](#) and [Australasian Institute of Digital Health](#)
- Data utilisation exploration with [Melbourne Genomics](#)
- Engagement with [JoinUs The George Institute Project](#)
- Development of comprehensive [databases](#) with clinical utility – local, national and international support groups linked to classifications and conditions

### 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 work & current projects

- Strategic Relationships with [Australian Genomics](#), [Melbourne Genomics](#), [InGeNA](#), [Australasian Institute of Digital Health Research Australia](#)
- [Surveys](#) to our community to establish gaps
- [C19Journals Project](#) partnership with [Australian Genomics](#)
- [Masters student](#) projects

# PILLAR 3 – RESEARCH & DATA

## 3.3 ENSURE RESEARCH INTO RARE DISEASE IS COLLABORATIVE AND PERSON-CENTRED

### GSNV work & current projects

- 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 work & current projects

- Involvement in [Mackenzie's Mission](#)
- Involvement in [RDNow Project](#)
- Collecting data from [Patient Pathways Program](#) to influence the clinical care process
- [Masters in Genetic Counselling](#) and [Masters in Genetic Health](#) Projects



# The Genetic Support Network of Victoria Year In Review 2020



## 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 many stakeholders including :

[Our community](#); people living with genetic, undiagnosed and rare conditions and those who support them

[All Victorian Clinical Genetics Services](#)

Department of Health and Human Services

[GUARD Collaborative Australia](#)

Genomics Clinical Advisory Group - Victoria

[Australasian Institute for Digital Health – Precision Healthcare Steering Committee](#)

Human Genetics Society of Australasia

[Australian Genomics](#)

Melbourne Genomics

[InGeNA- Genomics Industry Genomics Alliance](#)

Mackenzie's Mission Engagement Reference Group

[RDNow Consumer Engagement Committee](#)

National COVID-19 Clinical Guidelines Taskforce – Consumer Panel

[WHO Collaborative Global Network for Rare Diseases \(CGN4RD\)](#)

Rare Disease International (RDI)

[Asia Pacific Alliance of Rare Disease Organisations \(APARDO\)](#)

Royal College of Pathologists Lay Committee (RCPA) Lay Committee

[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](#)

Rehabilitation Psychology for Injury, Chronic Illness and Pain Interest Group

[Centre for Consumer Directed Research](#)