

# Living with a Genetic, Undiagnosed and Rare Disease Through the COVID-19 Pandemic: A Longitudinal Journaling Study



**Australian  
Genomics**



A  
collaborative  
research  
project  
undertaken  
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## EXECUTIVE SUMMARY

The aim of this exploratory study was to identify how COVID-19 impacted the lives of Australian adults living with a Genetic, Undiagnosed or Rare Disease. We collected monthly journals chronicling lived experience from June 2020-May 2021 and identified three timely waves of targeted support that would help sustain this community through crisis situations:

**1st wave:** facilitate finding a framework to structure day-to-day life

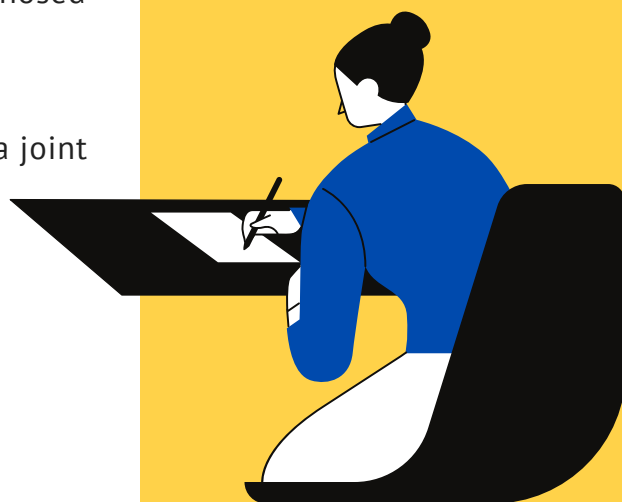
**2nd wave:** consistency in communication of health information

**3rd wave:** support for mental wellbeing

This knowledge allows us to develop a first line response to a crisis of this nature at it's outset. Resources for developing new routines and adapting and resetting as change keeps coming. It seeks accountability and consistency from the health sector to develop plain English, accessible and consistent information and the implementation of mental health support and protection mental health wellbeing from the beginning of such an event. Covid -19 changed the way we lived with uncertainty from the outset. The pandemic impacted every aspect of our lives from wellbeing, socialising to accessing healthcare. . For people in vulnerable populations, such as those with Genetic, Undiagnosed and Rare Disease, their carers, and the support sector, the experience was heightened. Recognising this inequity, this nationwide study was co-designed, co-led and analysed in a joint venture by Australian Genomics and The Genetic Support Network of Victoria.



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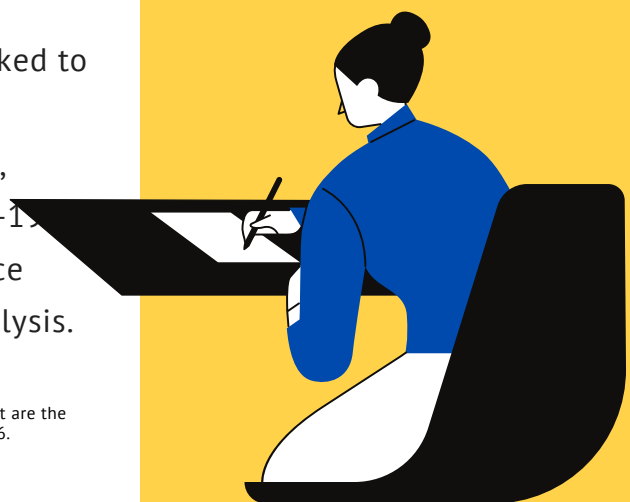
## AIM:

To identify how the rapidly changing COVID-19 environment impacted the lives of the Genetic, Undiagnosed and Rare Disease community across Australia. We investigated:

- **How has the COVID-19 pandemic impacted on the wellbeing and resilience of people in the Genetic, Undiagnosed and Rare Disease community and what coping mechanisms have they used?**
- **How has the COVID-19 pandemic affected access to health and social care services?**
- **What lessons can be learnt for future health and social care service provision for people in the Genetic, Undiagnosed and Rare Disease community?**

## METHODS:

We used a phenomenological, longitudinal, qualitative, research study and collected monthly open-ended journals, between June 2020 – May 2021, from the Genetic, Undiagnosed and Rare Disease community living in Australia. Participants were asked to keep a journal in any form they preferred (e.g. digital text, handwritten) and also in any format of preference (e.g. text, pictures) to share their lived experiences during the COVID-19 pandemic. Data analysis was deductive, using the Resilience Scale for Adults (1) (RSA), and inductive using thematic analysis.





# RESULTS:

Participants: We recruited 29 people and received an average of  $n=9.7$  submissions each month. Most participants were aged 18-29 years ( $n=10$ ) with six people aged 30-45 years, nine were 46-59 years and two between 60-75 years. The majority were based in metro locations ( $n=24$ ) with 12 people based in Victoria, 11 in New South Wales and just two in both Queensland and Western Australia. People played more than one role in the community with most having a Genetic, Undiagnosed and Rare Disease ( $n=18$ ), ten people were carers and nine were part of the peer support sector for people with Genetic, Undiagnosed and Rare Disease and/or their carers. Most participants ( $n=10/17$ ) solely used a word processor facility to record their journal, one participant hand wrote their journal and submitted photos of their writing, another participant sang their submission and sent a video, and several participants ( $n=5/17$ ) submitted multimedia journals that combined photographs, graphics, and text (figure 1)

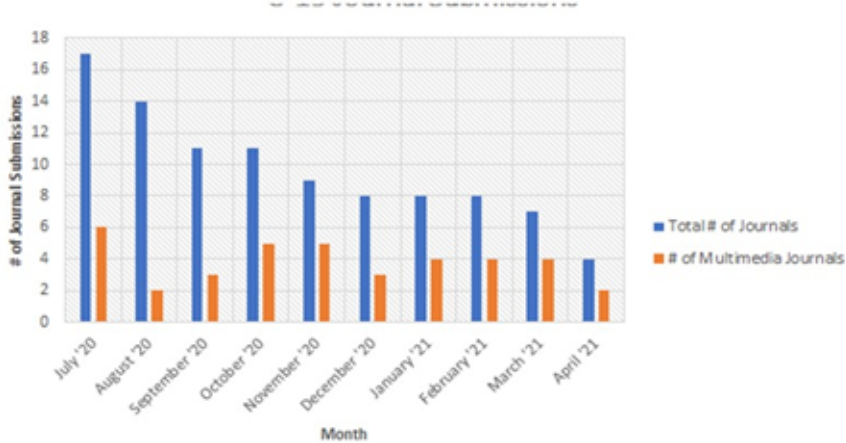
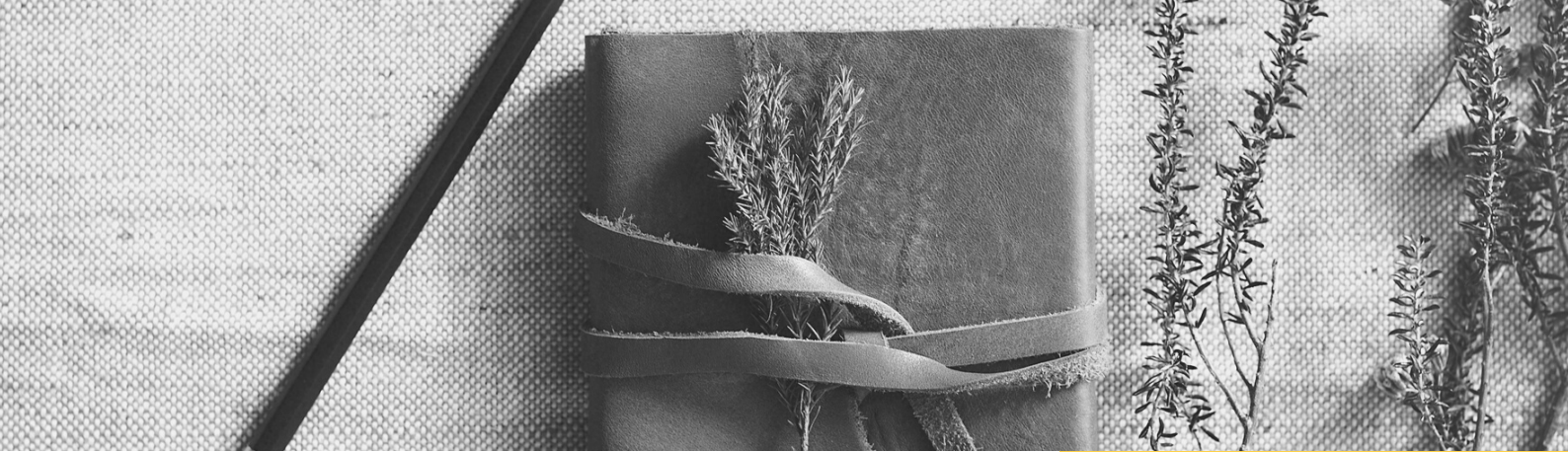


Figure 1: Journal submissions by month







# THEMES:

COVID-19 related themes came most commonly from journals where people were based in New South Wales and Victoria rather than the other states.

The resilience scale code, perception of self, began to peak around the middle of the submission timeline as participants experienced struggles as activities stopped and socialisation was kept to a minimum. Family cohesion remained a constant theme throughout the study, with initial difficulties of being separated from family. Later as the restrictions continued, participants found that cohesion in family life and dynamic were impacted. The social resources construct played an important role across the pandemic as people sought out health-oriented resources and additional supplemental resources as well. Coding of social competence revealed that the limitations of social interactions as a result of the pandemic had shifted much of the social sphere to online communication which removed some of the complexities usually involved in social interactions, but for others, technologically based communication was more difficult. Lastly, turning points and coping strategies were mentioned consistently across the journals from beginning to end. Turning points in the pandemic, including the imposition of restrictions were identified by participants and coping strategies varied from activities at home to socially distanced outdoor activities.

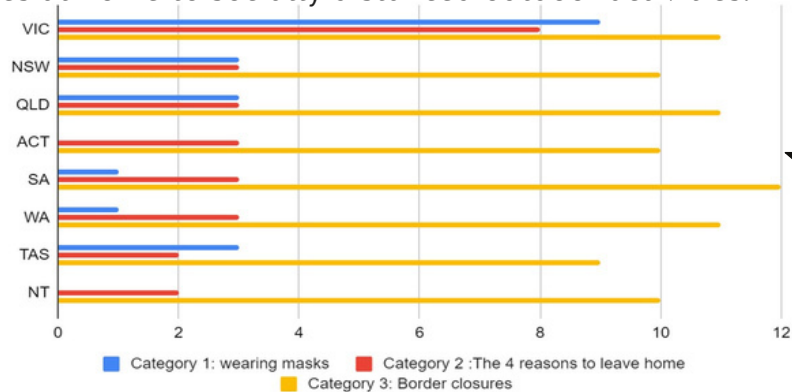


Figure 2: Restrictions by State 2020





## CONCLUSION:

Responses changed over the year. In line with the waves of the COVID-19 pandemic, there is a parallel need for waves of targeted support for vulnerable communities to cope through with crisis situations. The first support wave needs to focus on facilitating the identification of new frameworks to structure day-to-day lives. A second (and ongoing) wave needs to centre on consistency of communicating health information. Finally, a later theme is a need for mental wellbeing and coping with isolation. While consistent communication on health and social care throughout was essential.

## NEXT STEPS:

We have submitted a manuscript for peer review, Byun, M. Feller, H. Best, S. Ferrie, M, *Living with a Genetic, Undiagnosed or Rare Disease: a longitudinal journaling study through the COVID-19 pandemic Health Expectations (Under review)*. Subject to future research resources, we plan to mine the journals further and establish a follow-on study to identify potential interventions for support as prioritised by the Genetic, Undiagnosed and Rare Disease community.

*If you would like any further information about this study please contact us at [C19Journals@mcri.edu.au](mailto:C19Journals@mcri.edu.au)*





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### REFERENCES FOR FIGURE 2: COVID-19 RESTRICTIONS BY STATE

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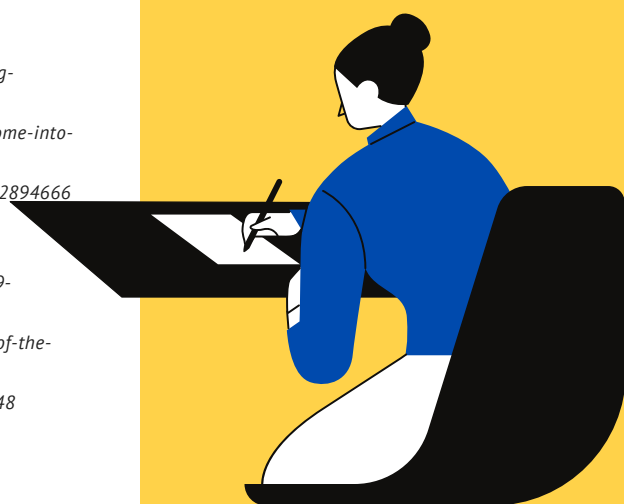
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