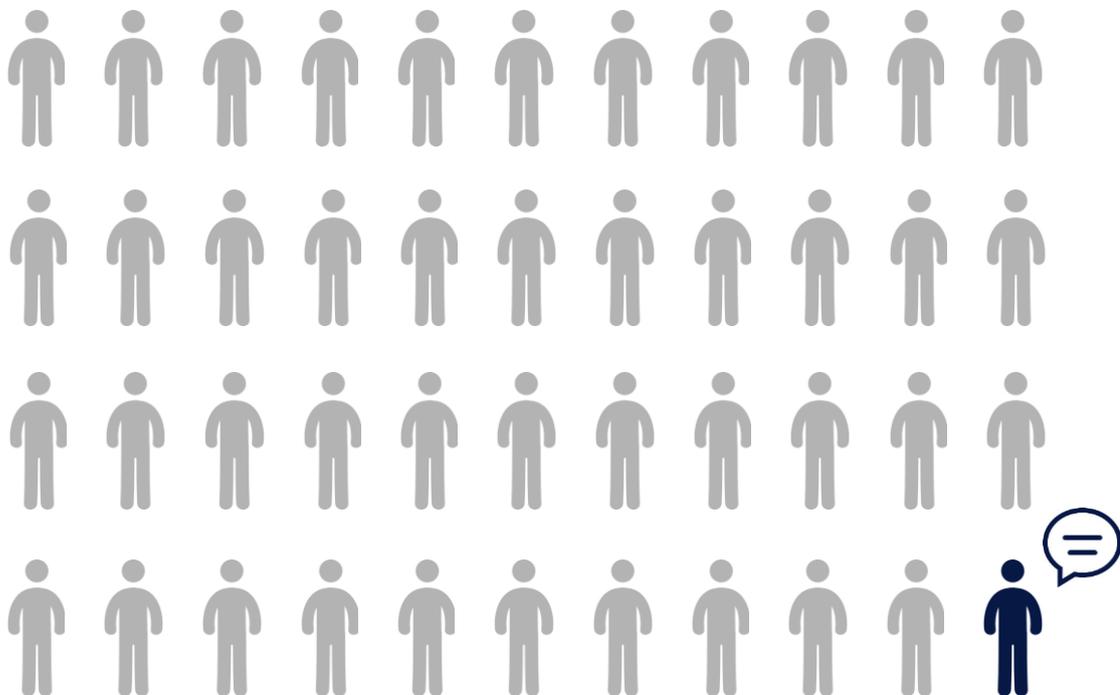




Government of **Western Australia**
Department of **Health**

Healthcare experiences of adults living with a rare disease in Australia

Results from the Australian Rare Diseases Survey



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Rare Voices Australia (www.rarevoices.org.au)

Genetic and Rare Diseases Network (www.geneticandrarediseasenetWORK.org.au)

Genetic Alliance Australia (www.geneticalliance.org.au)



This report is a summary of the following peer-reviewed article:

[Survey of healthcare experiences of Australian adults living with rare diseases.](#)

Molster C, Urwin D, Di Pietro L, Fookes M, Petrie D, van der Laan S, Dawkins H
Orphanet Journal of Rare Diseases, 24 March 2016 11:1, pp 1-12

Executive summary

Rare diseases include any disorder or condition that occurs at a low frequency within the general population. This can range from diseases that affect 1 in 2000 people (e.g. Cystic fibrosis) to diseases that affect fewer than 1 in a million people (e.g. Tricho-hepato-enteric syndrome). Although the individual diseases are rare, combined there are more than 6,000 distinct rare diseases that are estimated to affect 6% - 8% of the population. People living with rare diseases have a range of healthcare needs, but there is little evidence as to whether Australian healthcare systems are meeting these needs.

In 2014 an online survey was conducted to explore the healthcare experiences of Australian adults living with a rare disease. The survey was developed by the Western Australian Department of Health, together with Genetic Alliance Australia, the Genetic and Rare Disease Network, Genetic Support Network Victoria and Rare Voices Australia. In response to the survey invitation, 746 Australian adults living with a rare disease, or their carers, answered questions about their experiences of diagnosis, use of healthcare resources and involvement in research.

Many common experiences emerged from the study. For many of the respondents, obtaining a diagnosis for their disease took a long time (more than five years), and most had to see three or more doctors to get a confirmed diagnosis. Almost half of the respondents had experienced an incorrect diagnosis. More than half of respondents reported that they did not receive enough information about their disease at the time of diagnosis, and they found it difficult to understand the information they were given. The respondents who were referred to a website were most likely to say that they had understood the information, and most respondents preferred receiving information about their disease via a website.

Respondents used health services more frequently than the general population and two-thirds indicated that they received sufficient medical support. However, many respondents noted that social, financial and psychological support relating to their rare disease was lacking. Finally, most respondents indicated that they would like to be involved in research into their disease, with one third of participants saying that they had already contributed to research on their condition. Registries facilitate patient access to research and clinical trials, but only one-fifth of respondents said that they knew of a patient registry for their condition, despite a general willingness to join one.

The experiences of these Australian adults show that living with a rare disease can be a difficult and emotional journey. Based on the responses of the survey participants, the study identified areas in which the health care experience of adults living with a rare disease could be improved. Recommendations for the Australian health system include:

- explore the establishment of centres of expertise for rare diseases, to integrate and coordinate access to health services for diagnosis and ongoing care
- provide information and guidelines to health professionals to improve communication with patients at the time of diagnosis
- investigate whether a national patient registry is required in Australia for all rare diseases

Introduction

Why was the survey conducted?

People living with rare diseases have said they have a range of needs when it comes to healthcare, including: early and accurate diagnosis; useful, timely and accurate information to inform their decision-making about ongoing care and treatment; access to a range of services including general practitioners (GPs) and hospital-based services; coordinated care; information sharing among health professionals; health professionals who are educated about rare diseases; and involvement in research such as clinical trials of drug treatments and other therapies.

However in Australia, as elsewhere around the globe, there is little evidence on whether the healthcare needs of people living with rare diseases are being met. The Australian Rare Diseases Survey aimed to explore the healthcare experiences of adults living with a rare disease, in relation to four key topics, namely:

- experiences of diagnosis
- perceptions of the information provided at the time of diagnosis
- availability and use of health and support services, and
- involvement in research on their rare condition.

Who designed and implemented the survey?

The study was conducted by the Office of Population Health Genomics, Western Australian Department of Health in partnership with Genetic Support Network Victoria, Rare Voices Australia, the Genetic and Rare Disease Network, and Genetic Alliance Australia. The survey was largely based on a similar study conducted by [Rare Diseases UK](#) .

Who answered the survey?

The Australian Rare Diseases Survey was conducted online between 21 July 2014 and 1 September 2014. People had to be aged 18 years or older to participate.



Emails containing a link to the survey were sent to patient support groups and individuals on the mailing and contact lists of all five study partners.



The recipients of these emails were asked to forward the survey link on to any others they knew who were eligible to complete the survey.



In total, 746 responses were received from adults in Australia living with a confirmed rare disease. For 80 individuals, their relative, carer or support worker answered the survey on their behalf.

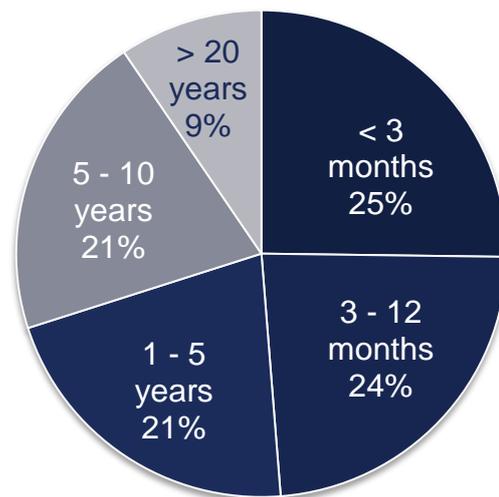
Experience of diagnosis

Diagnosis of a rare disease often took a long time, more than one doctor was required, and sometimes the initial diagnosis was incorrect. All of these may contribute to unnecessary or delayed treatment, poorer health outcomes, reduced quality of life, unnecessary hospital admissions and inefficient use of health system resources.

Nearly one third of respondents waited five or more years to get a diagnosis



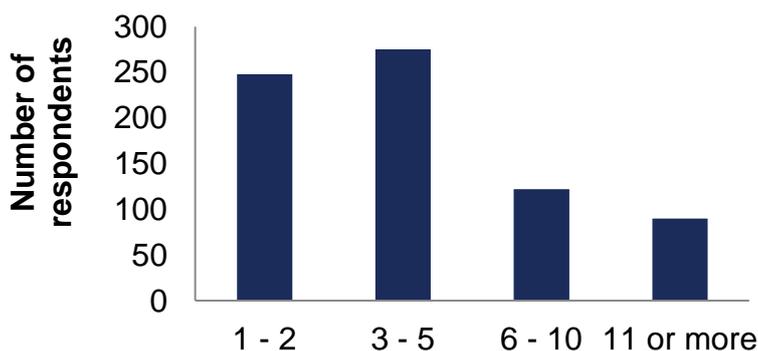
Time to diagnosis



"It's heartbreaking and exhausting. The first hurdle is convincing doctors that you have been to [psychologists] and it is not psychological."

When asked about the diagnostic process, 7% of respondents indicated that doctors had told them it was "all in their mind" or that the problem was not physical but a psychological one (e.g. stress, depression, anxiety).

Number of doctors seen to get a confirmed diagnosis



Two thirds of respondents had to see three or more doctors to receive a diagnosis





Respondents reported the situation where their disease or condition is multi-systemic and therefore they see specialists in different areas for the symptoms they have, but nobody “connects the dots” and looks at them as a whole person.

“My illness affects a lot of body systems. Since the medical system is set up with specialists in particular body systems there is no medical practitioner who will look at me as a whole.”

“It’s a very confusing and contradictory path. One specialist will say ‘Yes I believe you have X syndrome’ then a week later ‘No, you don’t have this’. They move from condition to condition and it’s a real process of elimination.”

Some respondents spoke of the implications for them of a long diagnostic process, such as not receiving treatment in time to prevent irreversible health outcomes. Others spoke of the diagnosis being a “process of elimination” where disease after disease is considered and then discounted as the cause of the symptoms being experienced.

45%

of respondents had received at least one incorrect diagnosis



There were some perceptions that GPs are not knowledgeable enough about rare diseases and that this lengthens and makes the diagnostic process more difficult.

Other respondents acknowledged the value of having a “good” GP.

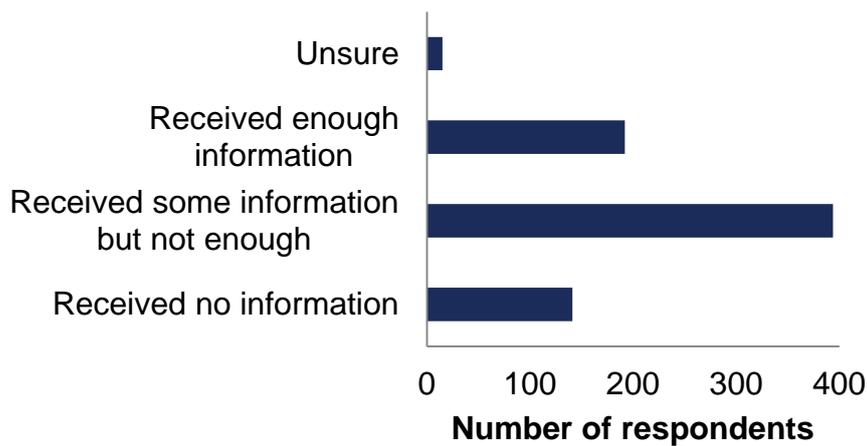
“My GP knows me very well and was very quick to recognise the symptoms I described. He took me seriously, referred me immediately to a neurologist and discussed his tentative diagnosis which was later confirmed.”

Perceptions of information received at diagnosis

Some respondents reported that the information they received at the time of diagnosis was “a lot to take in” and “too much information to take in at once”. More often however, respondents perceived they did not receive enough information at the time of diagnosis.

“I understood what I was told, but there was a lot I wasn't told. The diagnosis was given in a 10-minute appointment which is insufficient time to advise someone about diagnosis of a rare, life-changing illness.”

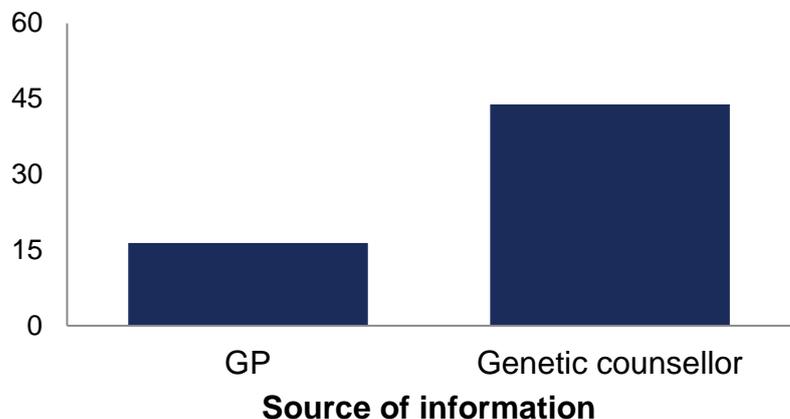
Information at the time of diagnosis



Most respondents received too little information at the time of diagnosis

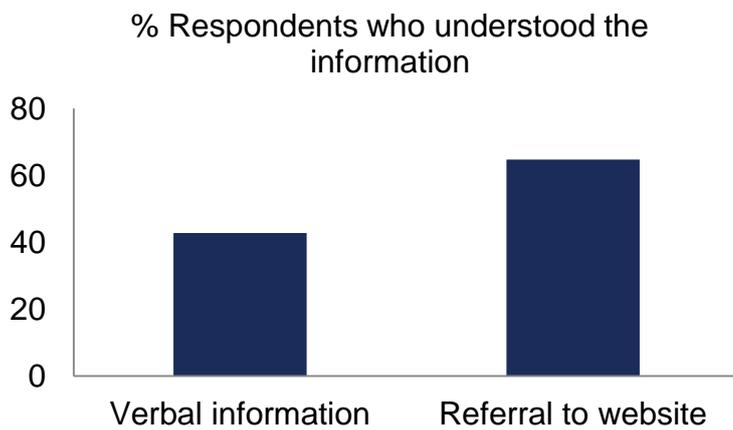
Those who received information from a genetic counsellor were more likely to say they received enough information

% Respondents who received enough information



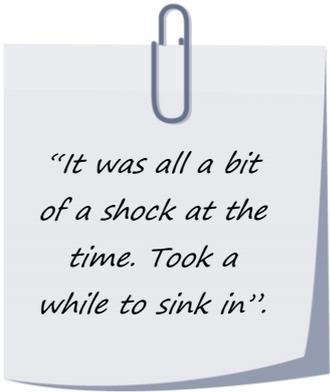
“It is a complex [condition] so it takes a while to understand it.”

Some respondents thought that the information they received was “medical jargon/terminology”, being too “complicated” and “technical” and not explained in layman’s terms. Some suggested that the fact they had not heard of the disease they were diagnosed with, and that it was rare and complicated, contributed to their inability to understand the information provided at the time of diagnosis.



Respondents who were referred to a website were more likely to say that they understood all of the information provided

Psychological and emotional responses to diagnosis also affected the ability of some respondents to take in and retain information. In particular this included being shocked and/or overwhelmed.

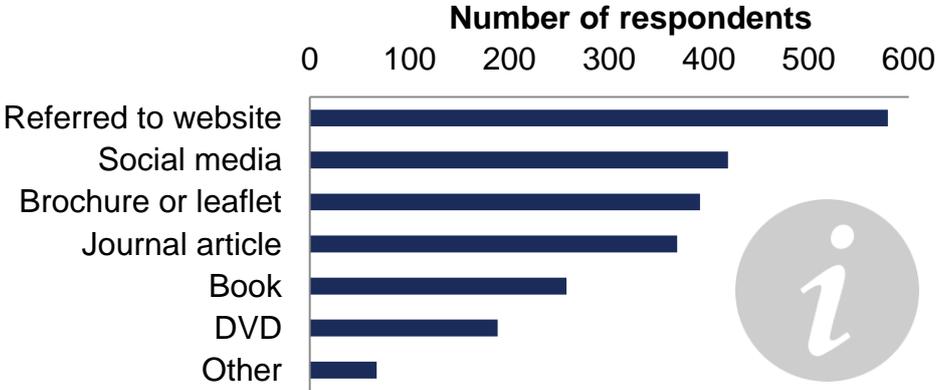


- The type of information that respondents had wanted to receive at the time of diagnosis included:
- ✓ Disease characteristics
 - ✓ Impact of the disease on daily life
 - ✓ Treatment options
 - ✓ Support options
 - ✓ Self-management
 - ✓ Research options

However, respondents didn't necessarily know what to ask at the time that they were diagnosed. Included at the back of this report is a list of questions that patients can ask that might help them get the information they need at the time of diagnosis.

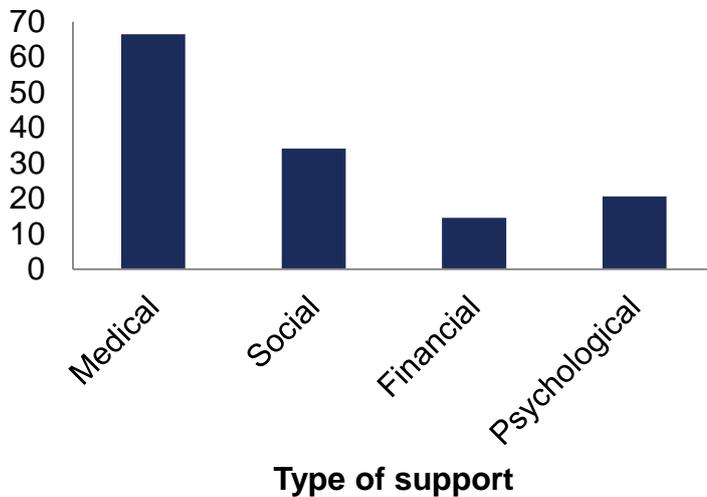
Preferred format for information

The preferred format for information was referral to a website



Availability and use of healthcare services

% Respondents who agreed that they received sufficient support



Most respondents agreed that they received sufficient medical support, but fewer agreed that they received sufficient social, financial and psychological support

The need for psychological support for people living with rare diseases is clear. Respondents spoke of the impact of the diagnostic process on their mental health. The emotion most commonly mentioned by respondents was “frustration”.

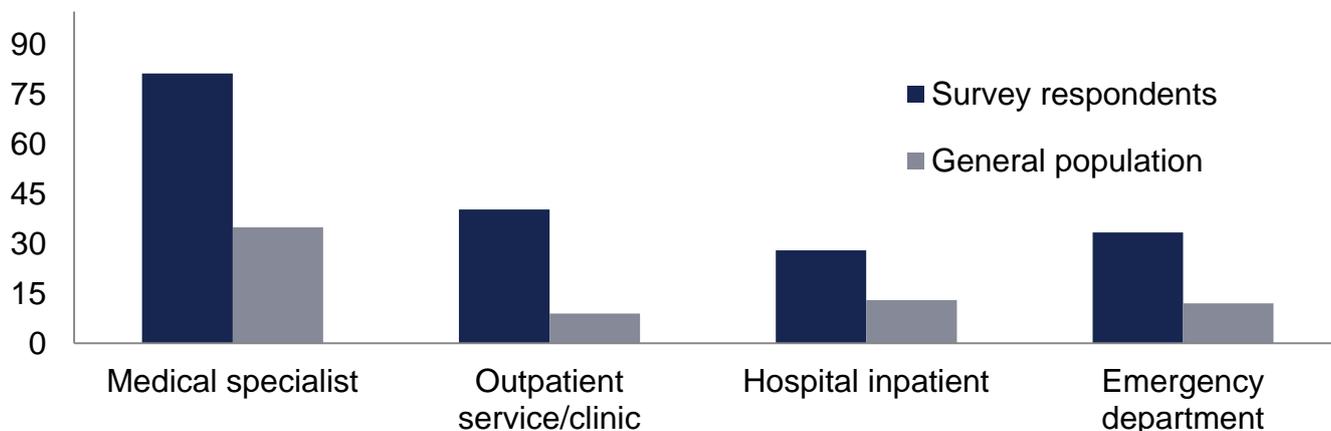
“Apart from the financial cost, the time, energy, emotional and psychological resources required to persist in this process...are unsustainable.”

“It’s exhausting to need to keep pushing and I think the mental health support needs to be increased.”

Others said it was stressful, confusing, disheartening, overwhelming, lonely, difficult, upsetting, depressing, time consuming, expensive, humiliating, traumatic, terrifying, draining; and that no or little counselling, emotional or mental health support was provided during the diagnostic process.

Respondents were more likely to have seen a medical specialist, been an inpatient at hospital, used an outpatient service or used an emergency department, compared to the general population.

% Respondents who used a service in the previous 12 months



Since many rare diseases originate in childhood, some respondents first accessed paediatric care, and had to transition to adult health services as young adults. Of the 746 respondents, 115 (15%) had used paediatric services for their rare disease.

57%

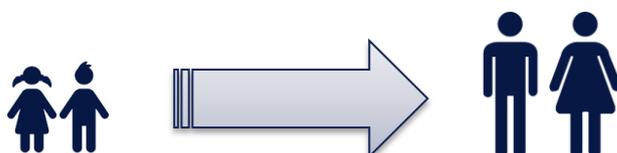
of respondents who had used paediatric services were **satisfied** with the care they had received

However, 53%

of respondents who had used paediatric services had **experienced problems** in the transition from paediatric to adult services

Some of the problems encountered by respondents in the transition from paediatric to adult services included:

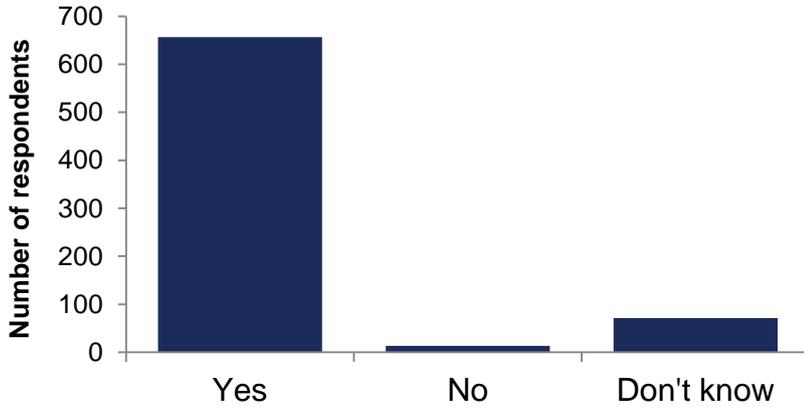
- lack of knowledge of the disease among adult services
- delays or lack of clarity around referral pathways
- lack of involvement of parent(s)
- lack of coordination of care
- lack of personalised care
- inability of health services to cope with intellectual disability



Involvement in research

In this study respondents indicated an overwhelming desire to be involved in research into their condition.

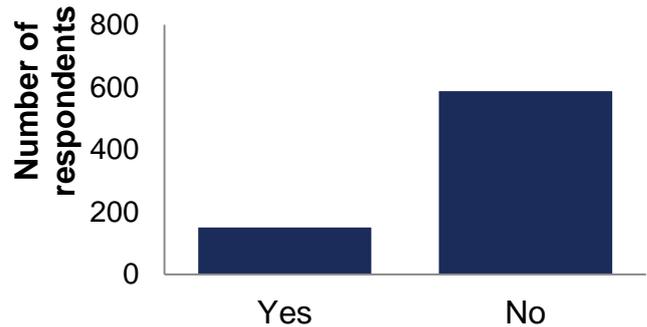
Would you join a patient registry?



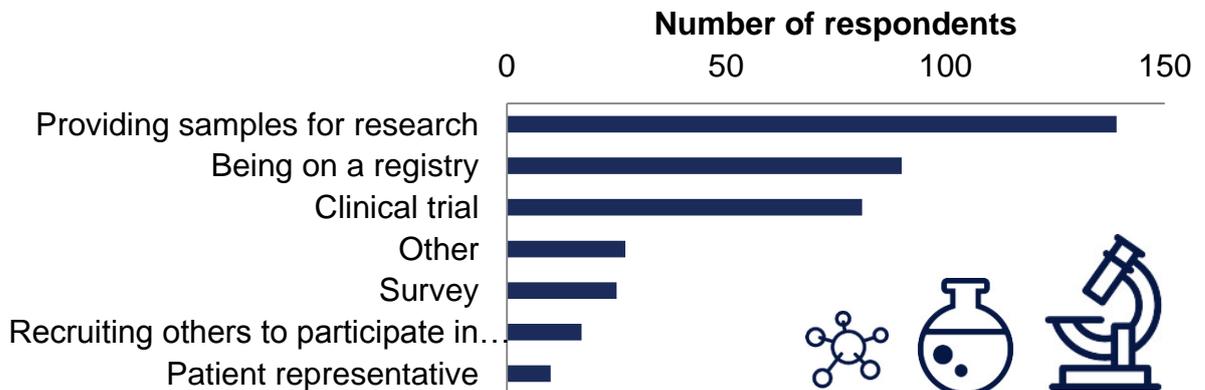
89% of respondents said that they would join a patient registry for their condition if one existed

But only 20% knew of a patient registry for their condition

Are you aware of a registry for your condition?



One third (33%) of respondents had participated in research into their condition, with the most common form of participation being the provision of biological samples for research, followed by being on a registry and participating in a clinical trial.



Recommendations

This study of 746 survey respondents indicates that not all of the healthcare needs of Australian adults living with a confirmed rare disease are being met. Common experiences were identified that cut across disease types, suggesting that looking at rare diseases as a collective group is an efficient way for health service providers and policy-makers to respond to the public health issue of rare diseases. Below are some recommendations for improving the healthcare experiences of adults living with a rare disease.

Experience of diagnosis:

- Multi-disciplinary centres of expertise for rare diseases may help to achieve timely and accurate diagnosis.
- Education could be provided to reinforce to health professionals that when they see a patient whose symptoms they can't explain, usual practice is to ask whether the cause could be a rare disease.
- Information relating to patient support groups could be provided to patients at the time of diagnosis.

Perceptions of information received at diagnosis:

- To improve availability of information, a checklist of answers to common questions could be available at the time of diagnosis (an example is provided at the end of this report).
- A "guide to living with a rare disease" might be useful for patients and medical specialists, and may include information on topics such as:
 - what to do now that you have been diagnosed
 - how to live with the disease and manage day-to-day living
 - what changes could be made to lifestyle, and
 - how the onset of symptoms could be avoided or delayed.

Availability and use of healthcare services:

- Multi-disciplinary centres of expertise should be considered to integrate services for disease diagnosis, follow-up and management, and care coordination.
- Mental health professionals should be a key part of the diagnostic process and ongoing disease management, to make sure that psychological needs are catered for.
- The economic costs of rare diseases to the health system should be explored.
- Transition from paediatric services for adults living with rare diseases should be improved, including better preparation and planning for transition and coordination of care.

Involvement in research:

- The need for a national patient registry in Australia for all rare diseases should be explored.

To aid with implementing some of these recommendations, the Western Australian Department of Health has developed the [WA Rare Diseases Strategic Framework 2015 – 2018](#). In addition, WA Health is working closely with the other states and territories to progress national planning initiatives for rare diseases across Australia.

Checklist of questions to ask when first diagnosed

Disease Characteristics

1. What is this disease? (explanation of the disease) How common is it?
2. What causes the disease? How/why did I get it?
3. Is it genetic/inherited? Can I pass it on to my children?
4. Is the disease curable?

Impact of Disease

1. What does it mean for me?
2. How will it affect me and my health? What impact will it have on my daily life?
3. What are the symptoms? Will these change over time?
4. What is the progression of the disease?
5. Is it likely to get worse?
6. What is the prognosis?
7. What can I expect? What does the future hold for me?
8. What is the likely outcome?
9. What quality of life can I expect?
10. What is the life expectancy with this disease? How long will I live for?
11. What are my chances of survival?

Treatment

1. What are all the treatment options available? What are the alternatives?
2. What medications should I take? What are the side effects? What are the pros and cons?
3. What will the outcome of treatment be?
4. Can I have a treatment plan?
5. How can I manage pain?
6. How do I find a specialist to treat my disease?

Support

1. Is there a support group for my disease? If so, what is the name of the support group?
2. How can I connect to other people who have this disease?
3. What support, help and advice is available and how do I access it?
4. What financial/income help is available?
5. Where else can I get good information? Any websites? Printed information?
6. Where can I go for counselling?

Self-management

1. What do I do now?
2. How can I manage the disease? How do I live with the disease?
3. How can I manage my day-to-day living?
4. What changes should I make to my lifestyle?
5. How can I avoid or delay the onset of symptoms?

Research

1. What research is being conducted on my disease?
2. How can I access research such as clinical trials?

**This document can be made available in alternative formats
on request for a person with a disability.**

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