

Suffering in silence: Assessing rare disease awareness and management in Australia

In November-December 2019, The Economist Intelligence Unit surveyed 103 practising healthcare professionals from Australia, as part of an Asia-Pacific analysis of understanding and challenges faced in managing rare diseases.

Our survey respondents from Australia comprised general practitioners or primary care physicians (45.5%), specialist physicians (34.2%), nurses (8.0%) and pharmacists (12.3%). Respondents predominately worked in private (49.7%) and public or teaching healthcare institutions (46.5%).

Results in context: Australia's health system

- Australia's population was estimated to be 25.2m in 2019.¹ The average life expectancy was an estimated 82.8 years in 2018, projected to increase to 83.6 years by 2023. Australia's population is ageing more slowly than many other OECD countries. This may be attributed to high net immigration and a plateau in the declining fertility rate.² Cancers, cardiovascular diseases, chronic obstructive pulmonary disease, Alzheimer's and dementia are the leading causes of death among Australians.
- According to OECD data, health expenditure was 9.3% of GDP in 2018, and the government increased the allocation for healthcare, aged care and sport by 4.7% in the 2019/20 fiscal year, to A\$104bn. The taxpayer funded

national public healthcare system, Medicare, covers residents for free hospital care and subsidises 85% of the cost of outpatient care. Total voluntary spending accounted for 30.7% of health expenditure in 2018, and an estimated 44.5% of the population have private hospital insurance.²

- There were 91,342 practising medical practitioners in 2016, with 11.5 nurses and four medical practitioners for every 1,000 people. Australia has an estimated 3.7 hospital beds per 1,000 population, with 62,000 beds in public hospitals and 34,300 beds in private hospitals.

Prioritising rare diseases: Early days for coordinated health policy in Australia

- The burden of rare diseases in Australia is not well defined. A 2010 study based on medical records in Western Australia found that 2% of the total population were admitted to hospital with a rare disease. The study notes that the true burden of such conditions is likely higher because data were available for only 467 diseases. Moreover, an indeterminate number of people with such conditions might well not have been hospital inpatients in 2010, opting instead for outpatient, primary care, or no medical services at all. The 2% of Western Australia's population with such conditions accounted for 10% of all hospital discharges and 11% of hospital costs.³

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¹ United Nations Population Division, World Population Prospects 2019. Estimates 1950–2020.

² The Economist Intelligence Unit. Industry Report Healthcare Australia. 3rd Quarter 2019.

³ Caroline Walker *et al.*, "The collective impact of rare diseases in Western Australia," *Genetics in Medicine* 2017;19:546–552.

- The definition of “rare disease” used in Australia is five cases per 10,000 people, as set out by the 2017 orphan drug regulation (which itself adopted the European definition).⁴
- Despite low awareness of Australia’s rare disease definition (Figure 1), our survey respondents recognised the value of such a definition, with over 80% saying it is important or very important. Respondents deemed the following elements to be significant in any definition:
 - o A low prevalence
 - o Difficulty of diagnosis
 - o Impact on quality of life or disability
 - o Availability of treatment
- Australia does not have a specific list of rare diseases for which it supports treatment. Instead the government uses a Life Saving Drugs Programme. This currently funds 16 medicines that are likely to improve survivability for 10 given conditions, but where data may be insufficient to receive approval and subsidies through regular channels.
- The Australian government launched the country’s first National Strategic Action Plan for Rare Diseases in February 2020.

Low awareness and knowledge among Australian healthcare professionals

- Healthcare professionals in our survey rated rare disease knowledge in Australia as middling (an average of 2.98 for themselves and 2.84 for peers on a scale of 1 to 5).
- This may not be surprising as only 30% of our survey respondents report seeing a new rare disease patient more than once every 6 months; 40% see a new patient once a year or more, and 8% report never having seen a rare disease patient.
- A previous 2017 survey of Australian paediatricians showed that, on average, under half believed that rare diseases were adequately covered during different levels of medical training and 28% felt unprepared to treat such patients.⁵

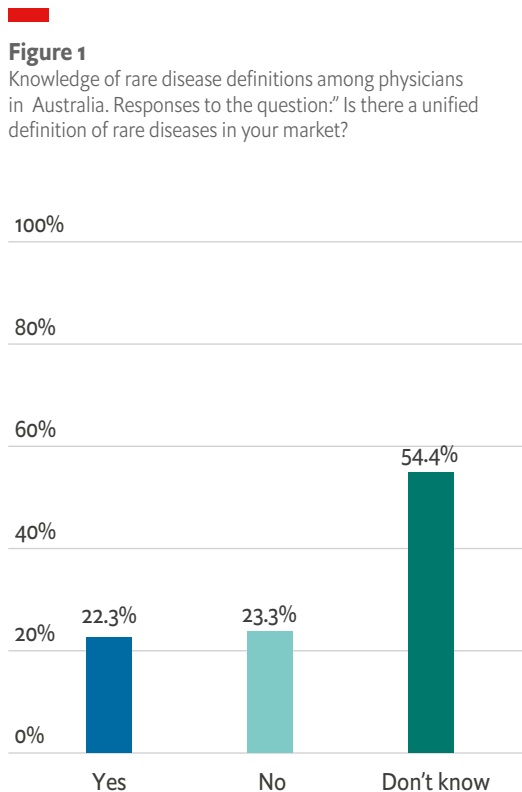


Figure 1
Knowledge of rare disease definitions among physicians in Australia. Responses to the question: “Is there a unified definition of rare diseases in your market?”

⁴ Australian Department of Health, *Orphan drug designation eligibility criteria*, 2018; Government of Australia, *Therapeutic Goods Regulations 1990* (updated 1 January 2017), 2017.

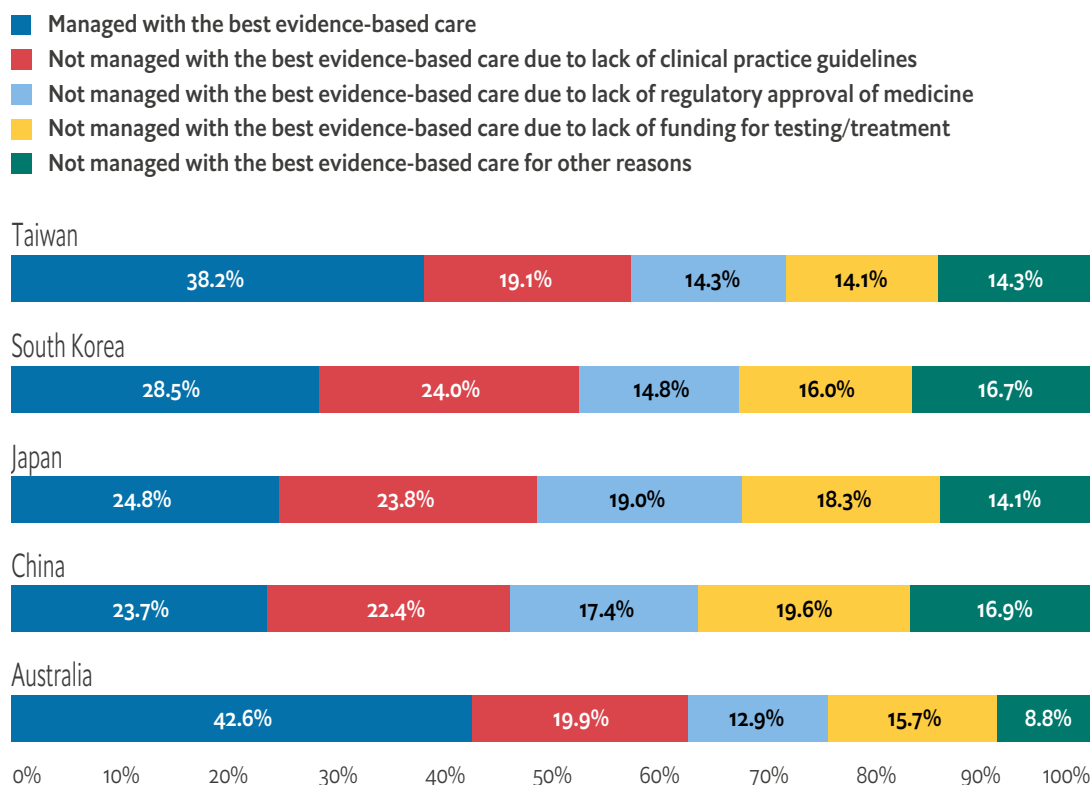
⁵ Yvonne Zurynski, et al., “Rare disease: A national survey of paediatricians’ experiences and needs,” *BMJ Paediatrics Open*, 2017; 1:e000172.

Less than half of patients receive the best available care and healthcare professionals experience multiple challenges in delivering rare disease care

- Respondents to our survey report that on average 46.2% of cases are managed with best evidence-based care, the highest proportion found in our research (Figure 2).
- Multiple areas were cited by our survey respondents as a challenge in diagnosing and managing rare diseases, most notably:
 - Reaching the correct diagnosis (23.2% report this is always a challenge)
 - Speed of diagnosis (22.1% report this is always a challenge)
 - Funding for treatment (20.0% report this is always a challenge)
 - Defined referral pathways (29.5% report this is always a challenge)
 - General population knowledge (33.7% report this is always a challenge)
 - Access to ongoing professional education (20.0% report this is always a challenge)

Figure 2

Survey responses reporting average proportion of patients managed with the optimal to sub-optimal care in five Asia-Pacific markets.



- Data from a 2016 survey of adults with rare diseases indicate that on average it takes 4.7 years to receive a diagnosis, with the mean number of doctors consulted just over five. Nearly half of respondents (49%) had a diagnosis within a year, but for 10% it took more than 20 years.⁶
- When our survey respondents were asked about the one action that should be taken to improve the lives of rare disease patients in Australia, the most common responses were:
 - o Increased awareness and education for both physicians and the general public
 - o Increased financial support for treatment, management that preserves patient function and palliative care
 - o Dedicated or defined clinical care pathways
 - o More research and data generation
- Despite this low visibility, Australian healthcare professionals recognise the value of such groups and report wishing to see more from them.
 - o 87.4% ranked patients, and 51.5% ranked patient groups as (very) important when developing care pathways for rare diseases
 - o More than 50% of respondents thought that patient organisations should play a bigger role in:
 - Promoting disease awareness.
 - Developing patient education tools.
 - Setting or designing research priorities.
 - Influencing or driving policy change.
- Numerous rare disease patient organisations are active in Australia, and most are small, volunteer-based groups. The vast majority are involved in some form of research activity, including contribution to registry data. Resources, lack of policy support and difficulty in engaging the scientific or medical community are cited challenges for these groups' work in Australia.⁷

Patient organisations are active, but there is wide variability in their effectiveness in reaching the full healthcare community

- Among our survey respondents, 56.3% did not know if rare disease patient organisations are active in Australia; 4.9% incorrectly reported that they are not.

⁶ Economist Intelligence Unit calculations based on Caron Molster *et al.*, "Survey of healthcare experiences of Australian adults living with rare diseases," *Orphanet Journal of Rare Diseases*, 2016;11:30.

⁷ Pinto *et al.* *Orphanet Journal of Rare Diseases* (2016) 11:2 DOI 10.1186/s13023-016-0382-6