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. 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. 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.

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.

• 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 has committed to putting in place the country’s first National Rare Diseases Framework and Action Plan, which is currently being drafted.

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?”

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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)

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**Figure 2**
Survey responses reporting average proportion of patients managed with the optimal to sub-optimal care in five Asia-Pacific markets.

- Managed with the best evidence-based care
- Not managed with the best evidence-based care due to lack of clinical practice guidelines
- Not managed with the best evidence-based care due to lack of regulatory approval of medicine
- Not managed with the best evidence-based care due to lack of funding for testing/treatment
- Not managed with the best evidence-based care for other reasons

### Taiwan
- Managed with the best evidence-based care: 38.2%
- Not managed due to lack of clinical practice guidelines: 19.1%
- Not managed due to lack of regulatory approval of medicine: 14.3%
- Not managed due to lack of funding for testing/treatment: 14.1%
- Not managed for other reasons: 14.3%

### South Korea
- Managed with the best evidence-based care: 28.5%
- Not managed due to lack of clinical practice guidelines: 24.0%
- Not managed due to lack of regulatory approval of medicine: 14.8%
- Not managed due to lack of funding for testing/treatment: 16.0%
- Not managed for other reasons: 16.7%

### Japan
- Managed with the best evidence-based care: 24.8%
- Not managed due to lack of clinical practice guidelines: 23.8%
- Not managed due to lack of regulatory approval of medicine: 19.0%
- Not managed due to lack of funding for testing/treatment: 18.3%
- Not managed for other reasons: 14.1%

### China
- Managed with the best evidence-based care: 23.7%
- Not managed due to lack of clinical practice guidelines: 22.4%
- Not managed due to lack of regulatory approval of medicine: 17.4%
- Not managed due to lack of funding for testing/treatment: 19.6%
- Not managed for other reasons: 16.9%

### Australia
- Managed with the best evidence-based care: 42.6%
- Not managed due to lack of clinical practice guidelines: 19.9%
- Not managed due to lack of regulatory approval of medicine: 12.9%
- Not managed due to lack of funding for testing/treatment: 15.7%
- Not managed for other reasons: 8.8%
• 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

**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.

• 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

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