



MACARTHUR SYMPOSIUM: RARE DISEASES

6 OCTOBER 2017

VC'S SPEAKING NOTES

Acknowledgement of country

Western Sydney University is proud to be hosting this event together with the Australasian Mastocytosis Society (TAMS) and the South Western Sydney Local Health District

Thank the organisers for the opportunity to open the symposium and welcome people to the Macarthur Clinical School:

Opened in August 2017, the Clinical School:

- Will provide one of the key training sites for the School of Medicine. Students receive hands-on clinical experience and mentoring by the hospital's senior specialists and other health professionals from their first year.
- Houses Campbelltown Hospital's postgraduate training unit for nursing and allied health professionals.
- The opening of the Clinical School coincides with the 10 year anniversary of WSU's School of Medicine.

Explain your interest and connection to the subject of rare diseases:

- Chair of Rare Voices Australia from 2013-2015
- "Rare Voices Australia (RVA) is Australia's non-profit, national peak organisation advocating for all who live with rare disease. RVA provides a strong common voice to advocate for health policy and a healthcare system that works for those with rare diseases".¹
- RVA has played a key role in helping to highlight and advocate the critical issues affecting the estimated 1.2 - 2 million Australians live with aRD.
- Despite this large number of Australians affected by rare diseases, Australia faces many challenges in addressing rare diseases and we have a lot of progress to make to catch up to other countries across the world.

¹ <https://www.rarevoices.org.au/page/7/our-purpose>

- In July 2017, RVA Australia released its key policy document *Call for a National Rare Disease Framework: 6 Strategic Priorities*.
- The Framework identifies a number of the critical issues affecting rare diseases and provides suggested areas for action necessary to addressing and managing rare diseases in Australia.

Critical issues around rare diseases (as outlined by RVA and the McKell Institute)

1. Rare diseases lack formal recognition as a national health priority

- Australia lacks healthcare policy and a healthcare system that works for those with rare diseases.
- Our health systems are designed to respond to diseases that affect larger numbers of patients rather than rare diseases.
- We lack a national rare disease framework which consequently has us falling behind countries such as the UK, France, Germany, Canada, Taiwan and more.

2. Our current data collection and information systems are largely inadequate

- The impact of RD remains largely hidden due to inadequate information systems.
- The prevalence and incidence of RD in Australia is largely based on estimates as we lack systematic patient data collection (i.e. registries) for most RD in Australia.
- The lack of standardised registries for most disorders means we lack basic information on:
 - The prevalence of rare disease
 - The number of patients receiving treatment
 - The quality of care and health outcomes
 - The overall burden of rare diseases to the nation

3. Lack of national coordination of care, services and dialogue for patients with rare diseases

- Policy and healthcare provisions remain fragmented across the states and we lack a coordinated healthcare system for sufferers of rare diseases.

4. Funding system for new rare disease therapies is in need of reform to ensure equitable access to healthcare for all rare disease sufferers

5. Australia out of line with international standards

- Australia needs to be brought closer into line with international standards around:
 - The definitions of rare diseases
 - Evidence requirements for treatment
 - Assessment of cost-effectiveness of treatment²

² McKell Institute Report, Funding Rare Disease Therapies in Australia, November 2014

Implications for Australia

- Consequently, a lack of prioritisation, coordination, and data and evidence of patient numbers leads to negative impacts and challenges such as:
 - a. **Rare diseases not being recognised as a national priority**
 - b. **Delayed diagnosis, inaccurate diagnosis, sub-optimal patient care and limited access to treatments for sufferers:**
 - 30% of people with a rare disease are impacted by a diagnostic delay of more than 5 years³
 - 50% had at least 1 misdiagnosis⁴
 - c. **Poor and inequitable access to services and treatment for rare diseases:**
 - McKell Institute notes that:
 - Australians are generally waiting anywhere from 2-4 years longer for access to government funded treatments for rare diseases than in comparable countries
 - Some medications remain unavailable eight years after becoming available overseas
 - Currently there are limited treatment options for many rare diseases in Australia with many treatments only available in countries outside of Australia.
 - The costs of services and treatments for rare disease sufferers can become uneconomical and unaffordable, while reimbursement pathways for treatments are not always equitable - treatments for rare disease can only potentially be considered for the Life Saving Drug Program once they have been rejected for funding on the PBS due to cost-effectiveness.
 - The nature of some rare diseases means that individuals may have to see a range of specialists and seek a range of services for treatment – currently many services are not provided in a fully coordinated manner and are fragmented.
 - d. **Inefficient use of resources and added costs:**
 - A lack of a coordinated approach leads to inefficient services and higher financial costs based on the provision of unnecessary tests and admissions and poor quality and inconsistent health care provision.

What is needed to improve the situation? As advocated for by RVA, we require:

(A) Improved data collection:

- RVA are advocating for the establishment of a national RD Registry Strategy and an Australian RD Registry Alliance to

³ Molster, C et al (2016) Survey of healthcare experiences of Australian adults living with rare diseases. Insert reference

⁴ ibid

encourage collaboration, shared knowledge, and best practice and to achieve better coordination among existing RD registries and stakeholders.

- The introduction of a registry will also be crucial to supporting research (including clinical trials and translational research) into rare diseases, particularly for benchmarking in research and clinical practice, as well as assisting with future government policy and funding.

(B) A nationally coordinated and collaborative approach for:

- **Dialogue, information exchange, and coordinated and collaborative action on RD.**
 - Gaining political traction in critical areas for rare diseases
 - Raising awareness (Launch of Fair for Rare Campaign by RVA in February 2017)
- **Access to services, diagnosis, care and treatment**
 - Australia requires a new approach that considers the full scope of rare disease management⁵ and ensures that rare disease is responded to in all current funded services and programs (eg the NDIS, chronic health, early intervention, and mental health)
 - Ensuring that care for patients is coordinated and that access to services is equitable and economically accessible
 - Increasing awareness amongst patients with rare diseases of the services that are available to them
- **Research**
 - RVA notes the need for:
 - improved research efforts and coordination
 - a national program of research on rare diseases
 - policy incentives to support research into rare diseases
 - RVA has played an important role in influencing the research agenda with regard to the MRFF priority to rare cancers and rare diseases
 - Under the MRFF, the Government will invest \$13 million to stimulate clinical trial and registry activity with priority to be given to under-researched health priorities, such as rare cancers and rare diseases.

(C) Stable, long-term government funding and support to sustain networking and coordinated efforts

CONCLUSION

- Australia still has a long way to go in its recognition and management of rare diseases
- Events such as the symposium are critical to raising the profile of rare

⁵ McKell Institute Report, Funding Rare Disease Therapies in Australia, November 2014

diseases and helping to address the challenges just discussed.

- Importance of engagement and collaboration, including with Rare Voices Australia.