



CARE AND SUPPORT

VISION: The best possible health and wellbeing outcomes for Australians living with a rare disease.

CRITICAL ENABLERS:

Multi-stakeholder involvement and engagement.	Collaborative governance and leadership.	State, national and international partnerships.	High quality, comprehensive collection, and effective use, of rare disease data.
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HOW WE ACHIEVE PROGRESS:

- Further develop RVA as the national peak organisation for rare diseases.
- Build on existing strengths to formalise centres of excellence throughout Australia.

WHY IS THIS IMPORTANT?

- Early diagnosis enables the best clinical care, treatment, access to services, peer support, increased reproductive confidence and access to participation in clinical trials. Yet diagnostic delay and misdiagnosis is common in rare diseases and impacts the person and family living with a rare disease physically, psychological, emotionally and financially.
- There is a real need for rare disease care and support to be less fragmented, more integrated, person and family-centred. A rare disease affects every facet of a person's life, not just their health. A lack of clear referral pathways makes it difficult for people to navigate their way through the different health, disability and other systems.
- Rare disease organisations provide valuable peer support, information, resources, and advocacy yet often face challenges with resourcing, sustainability and capacity.
- There are limited treatment options for rare diseases and even when a treatment does exist, financial support may not be available, limiting access. Reimbursement of health technologies for rare diseases is challenging, even for an approved medicine (for a more common condition).
- Time is often critical in rare diseases. Timely and equitable reimbursement is essential for people living with a rare disease to benefit from new and transformative health technologies.





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PRIORITIES

Rare disease care and support that is integrated, while being person and family-centred.

- Clear pathways throughout health, disability and other systems.
- Support policy work with a cross-jurisdictional, cross-sectoral working party.
- Services respond to the changing needs of people living with a rare disease and their families.
- Services support people living with a rare disease through life-stage transitions.
- Develop the capacity of rare disease organisations to represent and advocate for people.
- Embed the voice of people living with a rare disease throughout structures and systems that impact rare diseases.

Diagnosis of a rare disease is timely and accurate.

- Further investment into a diverse range of specialist diagnostic responses; including existing undiagnosed disease program models to achieve national coverage.
- Equitable access to a range of diagnostic tools/tests, supported by policy.
- National coordination of a range of screening and diagnostic tools/tests jointly funded by Commonwealth and state/territory governments.
- Address urgent funding gaps within the Newborn Bloodspot Screening (NBS) National Policy Framework.
- People with an undiagnosed rare disease are identified and have priority access to a specialised diagnostic response.
- Support people with a suspected but undiagnosed rare disease on their diagnostic journey.

Increased reproductive confidence.

- Those with an increased chance of being carriers of rare diseases have equitable access to peri-conception genetic testing and counselling.
- Provide women with certain chronic conditions, thus an increased chance of having babies with rare congenital anomalies, with access to pre-conception and peri-natal care.
- Develop non-directive education materials for use by individuals and families surrounding access to peri-conception genetic testing.
- Continue to support people through the range of possible outcomes following peri-conception genetic testing.

Equitable access to the best available health technology.

- Align with and build on the National Health Genomics Policy Framework for the systematic, equitable and timely delivery of genomic services.
- Develop policy that supports timely and equitable access to new and emerging health technologies. As technology advances, policy will require ongoing review.
- Broaden the principles underpinning Australian Health Technology Assessment (HTA) processes to acknowledge the challenges associated with assessing health technologies for rare diseases.
- Build rare disease expertise within the Office of HTA (OHTA).
- Funding and reimbursement pathways are fit-for-purpose and sustainable for current and new health technologies.

- Ensure all rare disease submissions are flagged as complex and may require additional scoping and engagement to address potential uncertainties. Raise awareness of the HTA Access Point.
- Rare disease organisations work with the HTA Consumer Evidence and Engagement Unit to take a more active role in HTA processes; including submitting applications for public reimbursement of a technology eligible for assessment.
- Ensure people living with a rare disease have equitable access to medicines with demonstrated clinical benefit for a rare disease, including those that are already funded for another condition.
- The Therapeutic Goods Administration (TGA) and OHTA continue to work together to develop clear processes and pathways for sponsors considering submitting applications for the repurposing of medicines already approved for use in treatment of other conditions.

Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

- Ensure people receive the community, clinical and digital mental health supports they need.
- Rare disease care and support systems address mental health and wellbeing.
- Develop the capacity of rare disease organisations to provide wellbeing and peer support.

