



The 2nd
**Rare Disease Asia
Conference 2016**

17 - 19 November 2016
Hotel Pullman Bangsar,

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WA Rare Diseases Strategic Framework 2015-2018

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Department of Health

WA Rare Diseases Strategic Framework 2015-2018



health.wa.gov.au

Background – identifying need

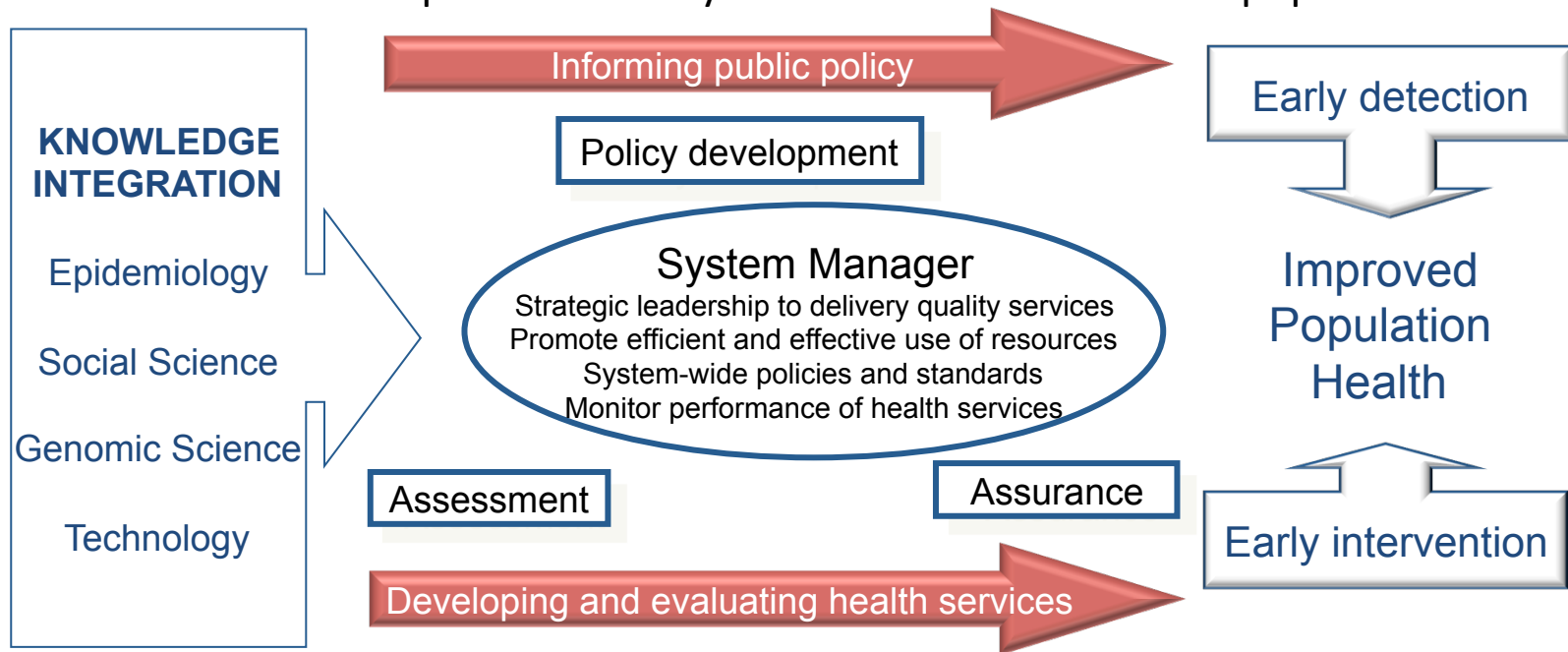
Development

Implementation

Factors for success

Background – OPHG & rare diseases

Primary purpose = translate genomic and other knowledge and technologies into the WA public health system for the benefit of the population





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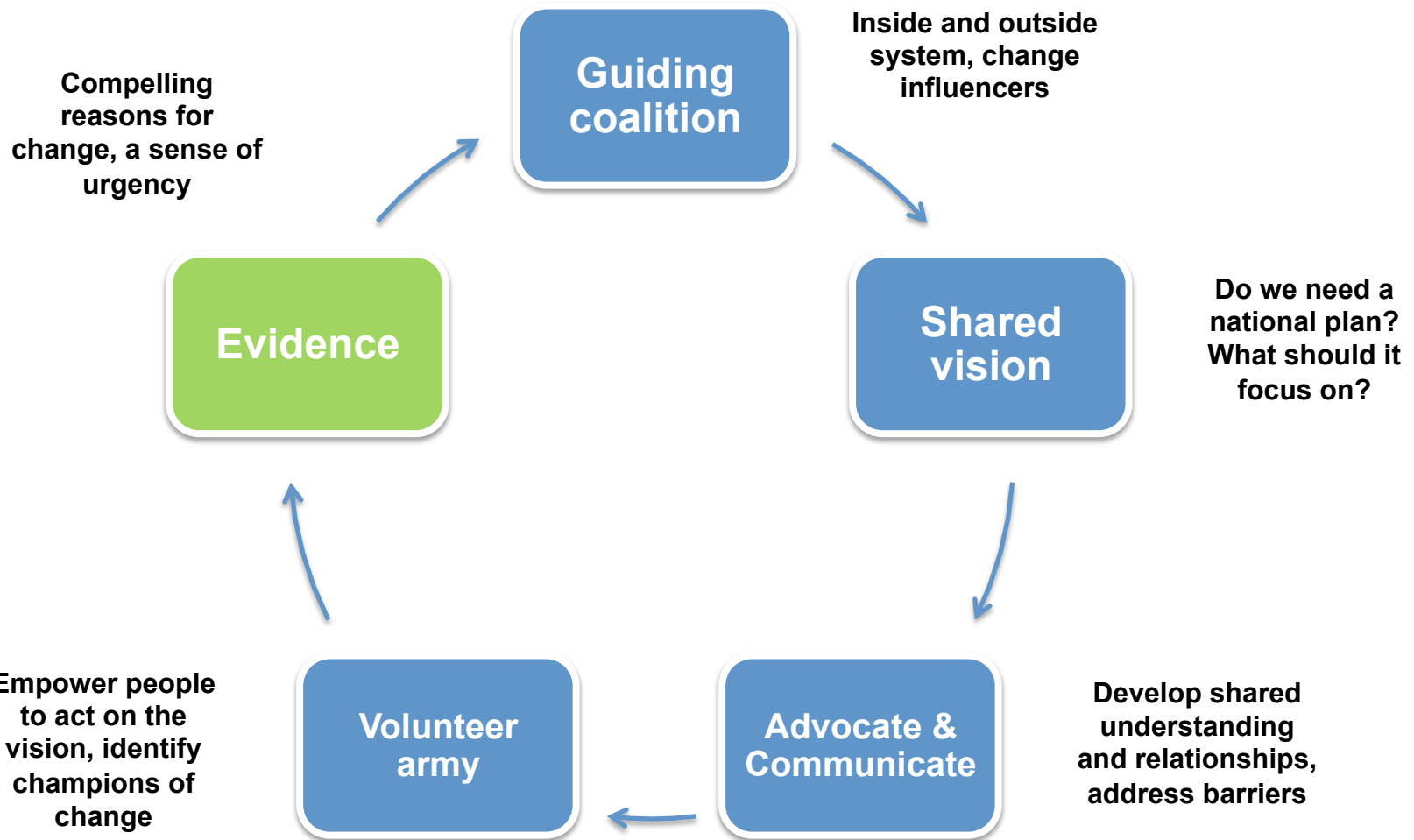
Background – OPHG & rare diseases

Horizon scanning

Genetic diseases  Rare diseases



Unmet need in Australia – no state or national plans



Adapted from Kotter's 8 steps to change

Shared vision – need for a national plan?



Development of national plans
Patient empowerment
Patient care, support and management
Research and translation
Networks, partnerships & collaboration

Patients, carers, families
Patient support groups
Clinicians & health services
Social & disability services
Researchers, industry
Regulators, policy makers

RESEARCH

Open Access

Key outcomes from stakeholder workshops at a symposium to inform the development of an Australian national plan for rare diseases

Caron Molster^{1*}, Leanne Youngs¹, Emma Hammond¹ and Hugh Dawkins^{1,2,3}, National Rare Diseases Coordinating Committee and National Rare Diseases Working Group

Need for a national plan



Need for a national alliance



rare voices
A U S T R A L I A



A UNIFIED VOICE

FOR ALL AUSTRALIANS LIVING WITH A RARE DISEASE

Standard definition

Education campaigns

Lobbying of government

Research infrastructure

Whole-of-lifetime services

Case coordination

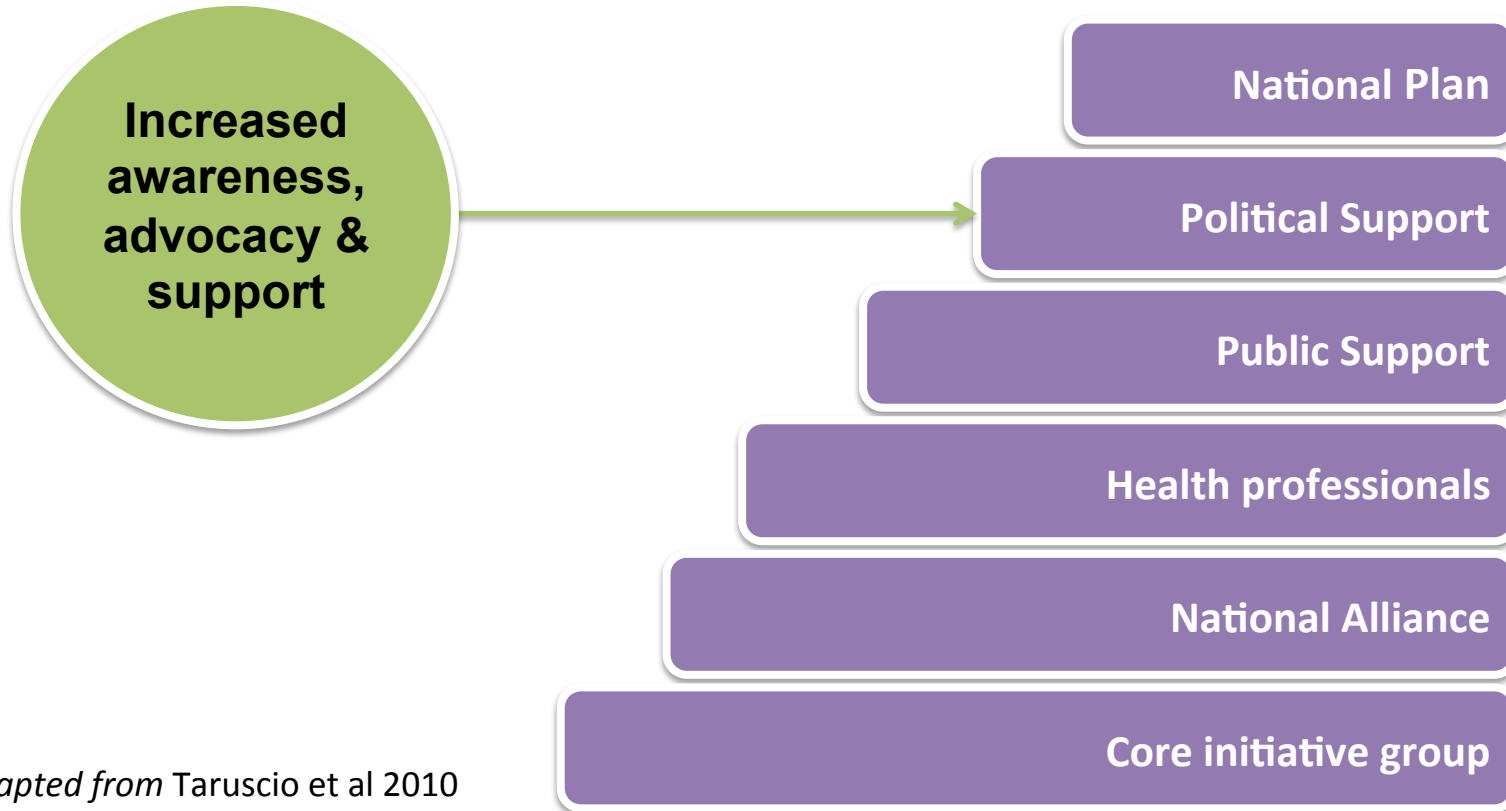
Early diagnosis

Support of health professionals

Dedicated funding

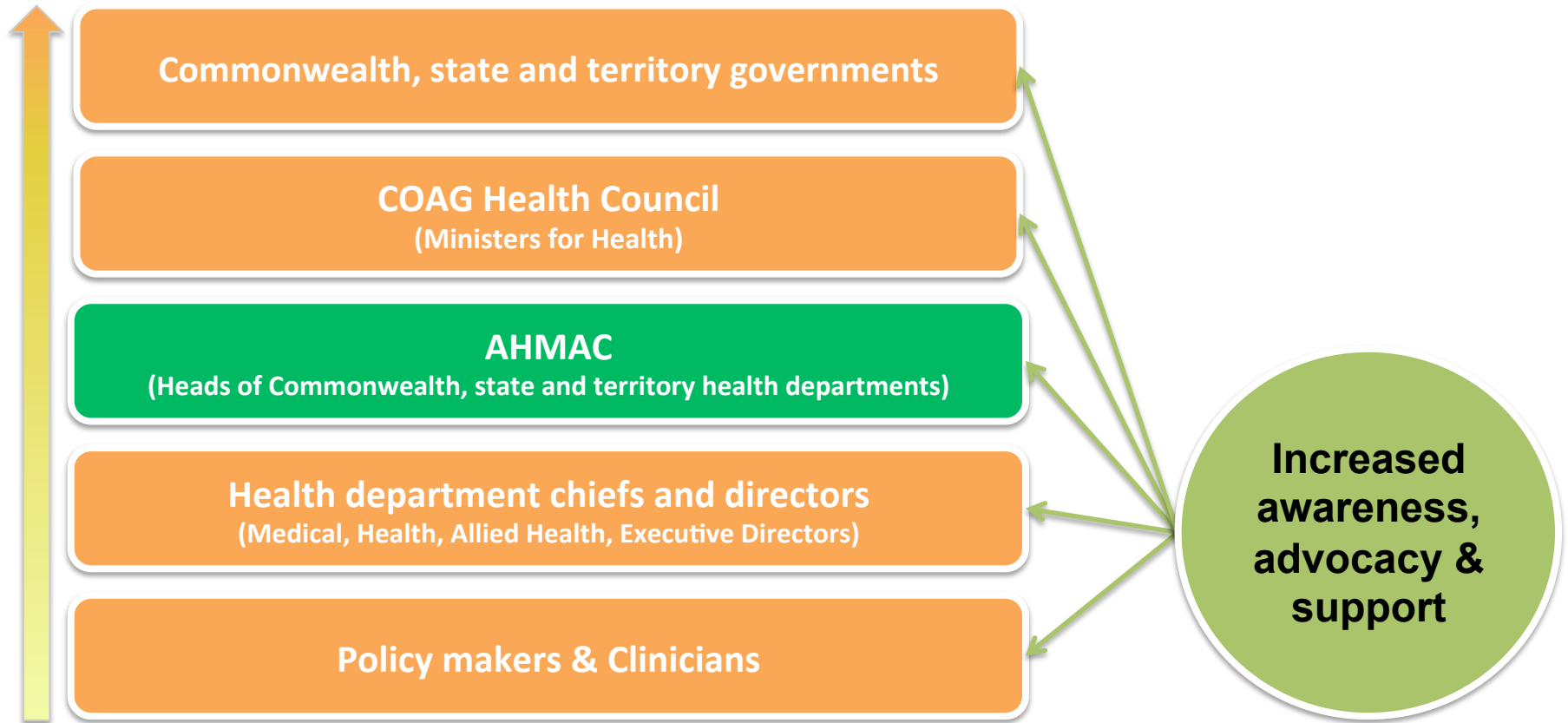
Need for local evidence

Roadmap to a national plan



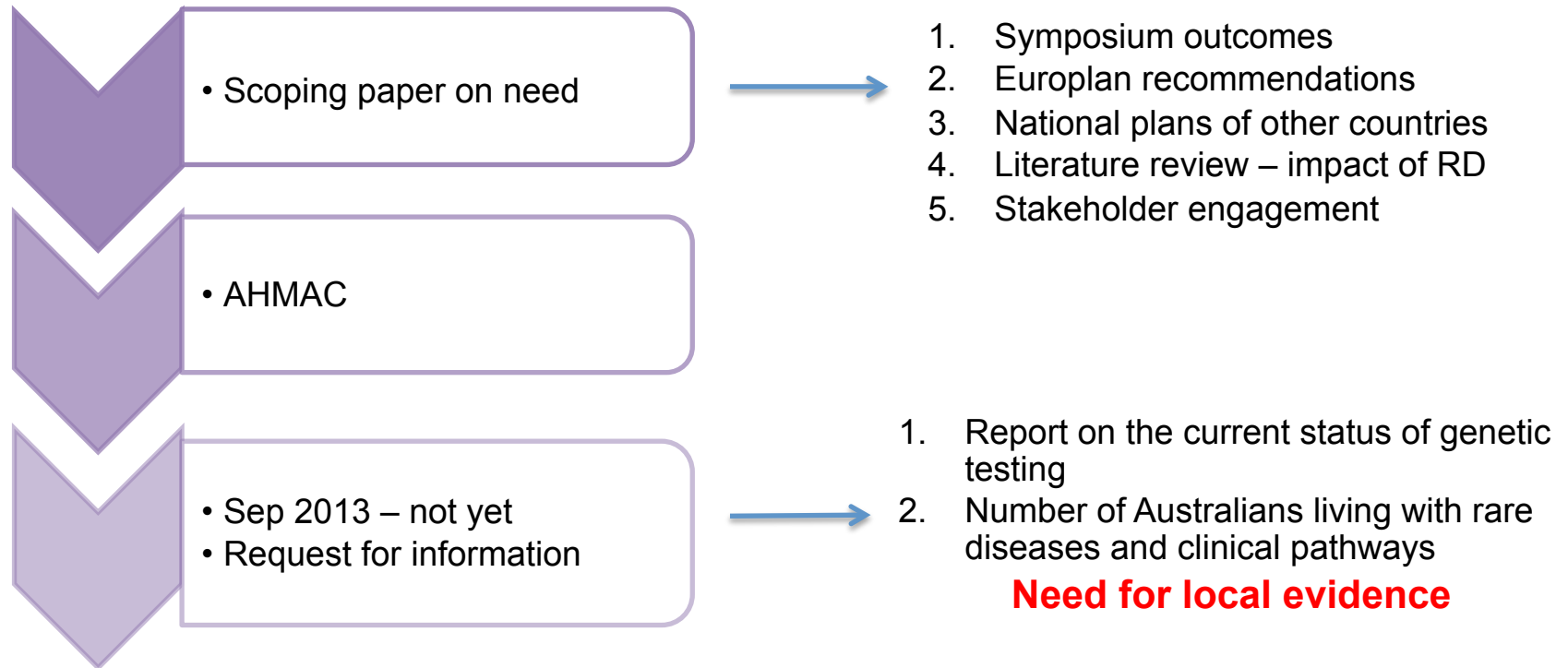
Adapted from Taruscio et al 2010

Advocating for a national plan





Advocating a national plan to AHMAC

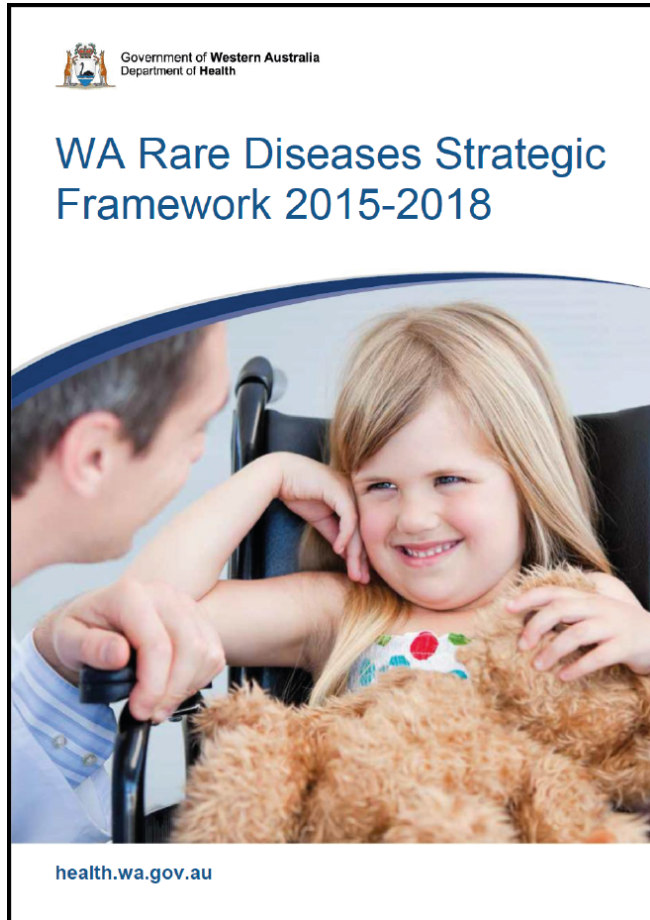




Proposed elements of a national plan



- Improve collection and sharing of data.
- Collective view of rare diseases.
- Map existing resources and services.
- Undertake rare disease surveillance, monitoring and reporting.
- Improve access and case coordination.
- Develop national partnerships for R&D of diagnostics and therapeutics.
- Investigate improving service pathways.
- Recognise the need for a rare disease peak alliance.
- Improve public access to information about rare diseases.
- Align the national plan for rare diseases with other national strategies.



Late 2013 – began development

Support Assistant Director-
General → Director-General

Updated literature review

X-referenced with relevant
existing state and national policies

Developed initial list of strategic
areas



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1. Need for planning
2. Evidence-base
3. Consumer engagement
4. Diagnosis & referral
5. Multidisciplinary coordinated care
6. Best practices - care and treatment
7. Support, access to information
8. Research & development

Stakeholder consultation



Appreciative eye



In every organisation
something works



SOAR methodology





SOAR

Strengths

OPHG; models of care;
international collaborations;
diagnostic services

Opportunities

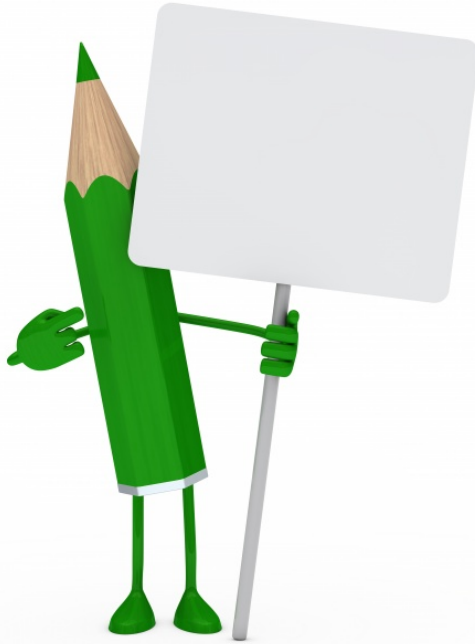
Referral pathways; bridge
state/national divide;
epidemiology; RD clinic

Results

RD introduced to health data
collections; complex case
coordination; increased
clinician awareness

Aspirations

Equity of access; quick, early
diagnosis; acknowledgement
of RD as a collective group



Evidence

Drafting content

1. Coordinated planning
2. Consumer engagement
3. Active participation in healthcare
4. Care coordination
5. Access to support and information
6. Integration of healthcare
7. Screening and diagnosis
8. Best-practice care guidelines
9. Emerging technologies
10. HPs access to information on RD
11. Epidemiology and other evidence
12. Clinical and translational research



What a Rare Diseases Strategy would do for WA

What is a rare disease?

A life-threatening or chronically debilitating disease which is statistically rare, with an estimated prevalence of less than 1 in 2000, and has a high level of complexity such that special combined efforts are needed to address the disorder or condition. Rare diseases include muscular dystrophy, cystic fibrosis and fabry disease.

Why are rare diseases important?

Estimates suggest there are between 5,000 to 8,000 rare diseases, which collectively affect 6-8% of the population. Extrapolating this to the Australian population suggests that more than 1.2 million Australians and 140,000 Western Australians are currently living with a rare disease.

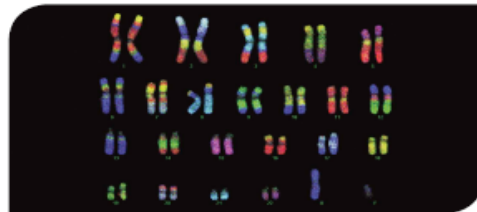
Reports from consumers suggest that care for rare diseases is fragmented and uncoordinated and thus there is an opportunity to improve efficiency and effectiveness of the health system.

Compared with other disease groups, patients with rare diseases account for a disproportionately high cost to the health system. Data is needed to quantify the collective impact of rare diseases in order to inform policies and service planning.

WA Rare Disease Strategy

The WA Rare Disease Strategy will aim to provide:

- Official recognition of the healthcare needs of people living with rare diseases
- A cohesive framework of WA Health's initiatives in relation to rare diseases
- Opportunity to benefit 190,000 Western Australians, their carers and families
- Support for clinicians, researchers and policy-makers.



What a Rare Diseases Strategy would do...

Carers would be able to say...

- My journey, in supporting my loved one, is easier and less stressful due to greater awareness, by clinicians and government, of the issues we face living with a rare disease
- The importance of my knowledge and the information I can share, as a carer, is acknowledged in guidelines developed for health professionals
- I am considered as a partner with other care providers in the provision of care, and the unique knowledge and experiences I have are acknowledged
- I have better support through access to information about patient groups across Australia.

Health care professionals would be able to say...

- I have a greater awareness of rare diseases and the issues and challenges they pose for patients, their carers and families, and the clinicians supporting them
- I know to question whether the people I see, whose symptoms I can't explain, have a rare disease
- I have access to information about rare diseases, to support me to provide early and accurate diagnoses to people with rare diseases
- I have a better understanding of the services and specialists to which I may refer patients
- I have access to best practice guidelines on rare diseases to support me to provide evidence-based care
- National clinical networks exist to support the development and distribution of these guidelines, which means that I may contribute my experiences and learn from those of other clinicians.

Consumers would be able to say...

- Efforts are being made to ensure I receive a timely and accurate diagnosis, and appropriate care
- I can access international clinical trials, and therefore new treatments, through local level registries
- There are opportunities for me to work with governments to share my experiences and inform policies and programs for rare diseases
- My data is being recorded by hospitals in a meaningful way to enable policy makers, service providers and researchers a clear view of the services I, and other people with rare diseases, need
- I have better support; I know how to connect with other patients, and find the services and specialists that can help me manage my disease.

WA Health would be able to say...

- We have a clearer understanding of the services accessed by people living with rare diseases in our state, which may inform the services we offer
- We have data on rare diseases and can: consider trends over time; identify the costs associated with rare diseases; and use this data to inform service planning and policy development
- We are supporting our clinicians and care providers by contributing to a plan that provides them with tools to better manage rare diseases
- We have an understanding of the level of coordination of care across the services accessed by people with rare diseases
- We have in place policies and services to support a vulnerable and isolated population of healthcare users.



Produced by the Office of Population Health Genomics
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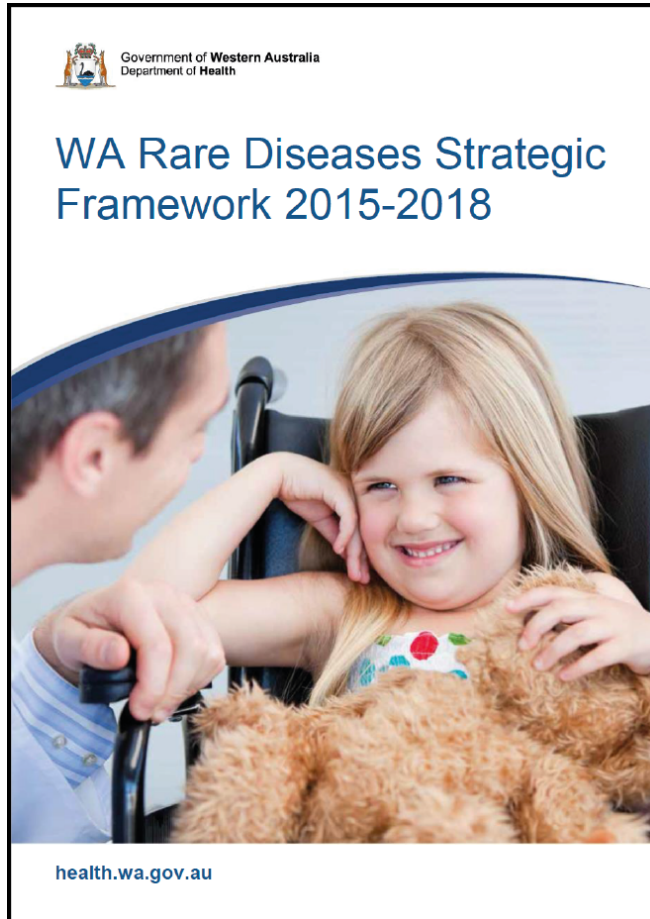
Delivering a Healthy WA



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Drafting

Stakeholder review

Final document

Executive sign off

Released June 2015



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Vislon

The best possible health and wellbeing for Western Australians living with rare diseases (RD)

Aim of the WA Rare Diseases Strategic Framework

Provide a framework for the coordination of WA Health initiatives for rare diseases for 2015–2018

Priority 1

To advance RD planning in WA and Australia

Objectives

1. Adopt a coordinated, collaborative approach to RD planning.

Priority 2

To promote a person-centred approach throughout WA Health for people living with RD

Objectives

2. Engage with people living with RD, their carers and families
3. Promote active participation of people living with RD in their healthcare
4. Promote care coordination for people living with RD
5. Facilitate access for people living with RD, their carers and families to support networks and information on RD.

Priority 3

To contribute to a high-quality health system for people living with RD

Objectives

6. Champion integration and partnerships in the delivery of healthcare for RD
7. Build on existing WA Health services for screening and diagnosis of RD
8. Encourage the use of evidence-based, best-practice guidelines to deliver healthcare for RD
9. Identify emerging technologies to enhance the delivery of health care for RD
10. Facilitate access by health professionals to information, education and training on RD.

Priority 4

To foster world-class research on RD

Objectives

11. Build epidemiology and health system evidence for RD
12. Strengthen clinical and translational research in RD.

Build the case for a collective view of RD

Build the compelling case for addressing needs

Build on existing initiatives and services

Build the evidence

Objective 1: Adopt a coordinated, collaborative approach

This strategic framework brings a range of WA Health initiatives for RD together into a single framework. It aligns with other WA Health documents which have elements relevant to RD planning. These include the *WA Chronic Conditions Framework 2011–2016*, *Our Children Our Future – A Framework for Child and Youth Health Services in Western Australia 2008–2012* (currently being updated) and *Palliative Model of Care*. It is important to encourage future WA Health policies, strategies, frameworks and models of care to consider RD and align with this strategic framework.

Coordinated planning for RD begins by taking a collective view of RD. This recognises that RD have commonalities and in total affect up to 6–8% of the population, thus representing a significant proportion of Western Australians who use WA Health services. A collective view should raise the profile of RD in a health system that is oriented towards more common diseases and is more efficient than if planning efforts were duplicated many times over for the 5,000–8,000 individual RD.

Effective implementation of the initiatives in this strategic framework must involve collaboration, networking and partnerships with local, national and international RD stakeholders. This recognises that links between people living with RD, their carers and families, healthcare providers, researchers, industry and policy makers are vital to facilitate the best possible health and wellbeing for people living with RD. National and international collaborations and partnerships will build on strong local networks. Extending beyond local borders enables WA Health to draw on the international experiences of countries that are further progressed with RD planning and facilitates the coordination of local access to national and international resources for RD.

In addition to state initiatives, it is imperative that coordinated planning for RD occurs at a national level. This will bring Australia in line with other countries which have adopted national plans and initiatives to respond to the needs of people living with RD. WA Health has already made significant contributions at a national level and will continue to engage in and sponsor initiatives that promote the need for RD planning at the national level.

Initiatives

1. Encourage the consideration of RD in WA Health policies, strategies, frameworks and models of care.
2. Represent RD on relevant WA Health Networks.
3. Promote a collective view of rare diseases and the adoption by WA Health of a standard definition of RD.
4. Promote sustainability for RD planning in WA.
5. Foster collaboration, networking and partnerships with local, national and international stakeholders.
6. Build the capacity of community service organisations in the rare diseases sector to develop and contribute to networks, collaborations and partnerships, including those with WA Health.
7. Establish a WA RD advisory group.
8. Support initiatives to advance RD planning at the national level.



Objective 2: Engage with people living with RD, their carers and families

Engagement is a process by which the aspirations, concerns, needs and values of people living with RD, their carers and families can be incorporated in WA Health decision-making [30]. Engagement recognises that people have the right to be included in the decision-making processes that affect their healthcare and that people living with RD, their carers and families have unique knowledge and experiences that can contribute to health system decision-making. Engagement with people living with RD, their carers and families should occur in all areas of the health system including: the development of policies and strategies; service planning, design/redesign, delivery and evaluation; quality improvement; and the development of education resources [21, 31–33]. The *WA Health Consumer Carer and Community Engagement Framework 2011–2016* [30] establishes engagement as an integral part of core business for WA Health. The Framework must be accompanied by the development of a culture of engagement so that people living with RD, carers and families are genuinely involved in decision-making.

This strategic framework commits WA Health to engaging people living with RD, carers, families and community service organisations in the RD sector by way of: their involvement in key organisational committees and appointments to advisory and governing structures (e.g. the RD advisory group); partnering to implement the initiatives of this strategic framework; and adopting mechanisms to ensure their views and needs are understood by WA Health. Throughout these processes consideration will be given to ways of involving and addressing the needs of harder-to-reach groups such as Aboriginal Australians, people from culturally and linguistically diverse (CALD) backgrounds and people living in rural/remote areas.

Emerging evidence indicates that engagement contributes to improved health outcomes at the individual, service, network and systems level [22, 24]. It results in a more responsive and integrated health system and helps to achieve long term sustainability [34] and equitable and effective decision-making for health services planning [35]. In general a service designed with an understanding of the views and needs of those who use it is more likely to effectively target these needs.

Initiatives

1. Include people living with RD, carers, families and their representatives on the state RD advisory group and other committees, advisory groups and working groups that are established to implement this strategic framework.
2. Consider ways to involve and address the needs of harder-to-reach populations, such as Aboriginal, CALD and rural/remote populations.
3. Build the capacity of community service organisations in the rare diseases sector to investigate and represent to WA Health the views and needs of people living with RD, their carers and families.



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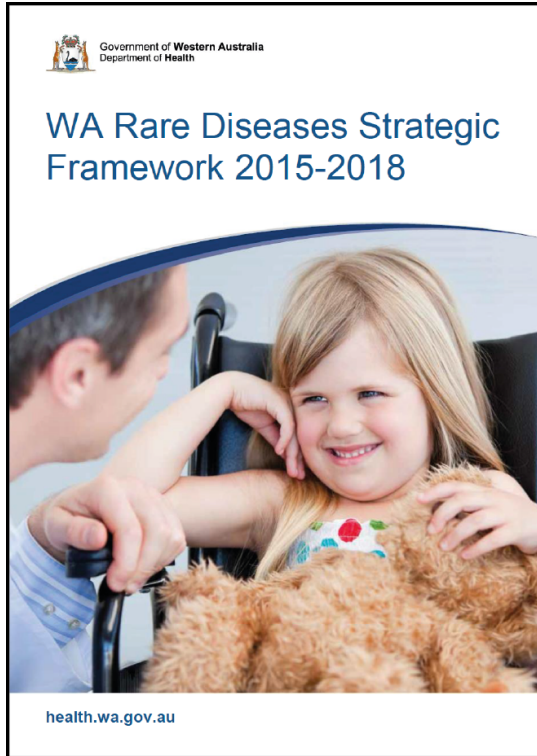


Implementation Plan



- Governance – roles, accountabilities and responsibilities
- Critical success factors – commitment, engagement, planning, communication
- Monitoring, evaluation and review
- Outcomes for each objective
- Deliverables and expected completion date for every initiative

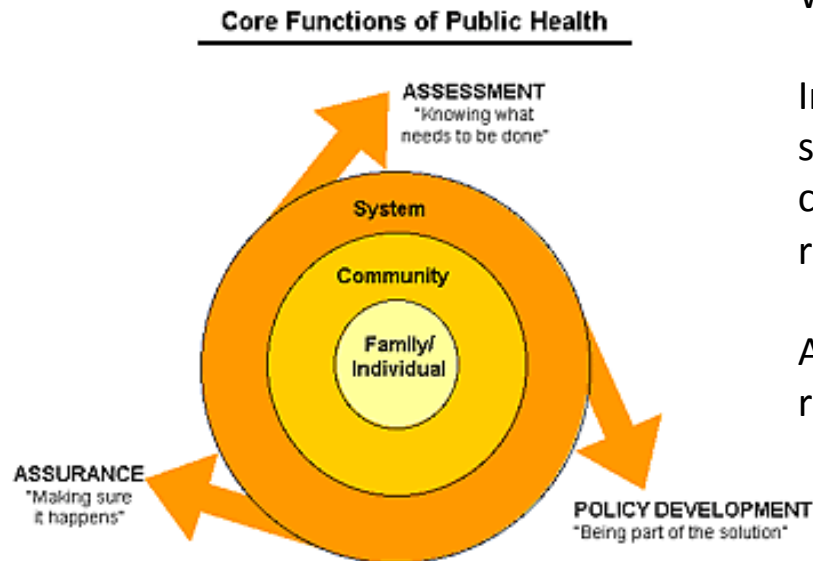
Why did we succeed?



- Haven't tried to change the whole system – built on strengths and opportunities
- Political support and champions
- Links between policy-makers and stakeholders, including patients and patient organisations who gave personal stories
- Communication – internal and external
- Recognises more evidence of impact required



Where are we now?



What is the local impact of RD?

In relation to what aspect of health and social care? (e.g. diagnosis, care coordination, multi-disciplinary care, research)

Are there common impacts across a range of areas?

Are services and interventions addressing the impact?

What can be done about the impact?