Government of **Western Australia**
Department of **Health**

How to Make Rare Diseases an Asian Health priority

Professor Tarun Weeramanthri
Chief Health Officer &
Assistant Director General
Public Health Division
Department of Health
Western Australia

2nd Rare Disease Asia Conference
17-19 November, 2016
Kuala Lumpur, Malaysia

Outline of talk

- We are at a tipping point – from recognising the problem to acting on the problem
- Are you ready for that change?
- Can you advocate effectively in the new world?
- How government might view this, and what you need to ask of government
- Draw on experience of recent meetings in Thailand and Malaysia

Helen Clark: Administrator of the United Nations Development Programme

Written statement at the 11th annual International Conference on Rare Diseases and Orphan Drugs 2016

- More than 300 million people around the world live with at least one rare disease.
- Ill health and the costs associated with it are major factors which push people into poverty
- 2030 Agenda for Sustainable Development fundamental principle ‘to leave no one behind’
- No country can claim to have achieved universal healthcare if it has not met the needs of those with rare diseases
- Governments need to look at legal and regulatory incentives for development of drugs and technology
- Collaboration between all sectors is critical

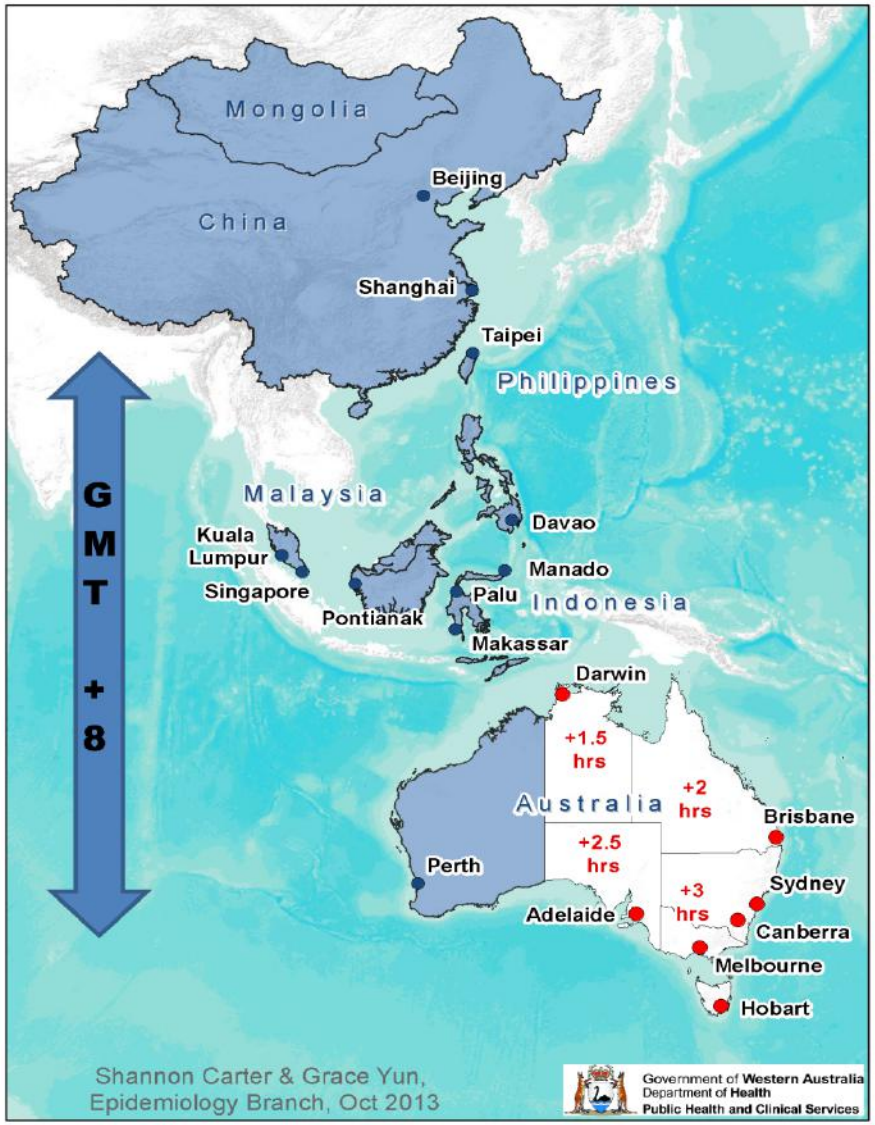
Rare diseases: key policy questions

- Is this really a problem compared to other issues?
- What can we done about it, and what is government's role?
- Is it affordable?
- What stakeholder resistance will be encountered?
- Is there a clear path forward and what barriers will there be?
- Who will thank us?

Rare diseases: key policy answers

- It’s as big as diabetes, it’s hidden, and you’re already paying for it
- Better coordination of existing resources will save money and lives
- Government has a key role but does not have to do and fund everything - a small expert team can make a big difference
- There will be resistance to change from some professional groups and some patient organisations
- But the benefits in terms of individual patient stories (as well as at the group level) are immediate and compelling
- If you build on all strengths, you will be able to describe a clear path forward

In the Zone


Mongolia
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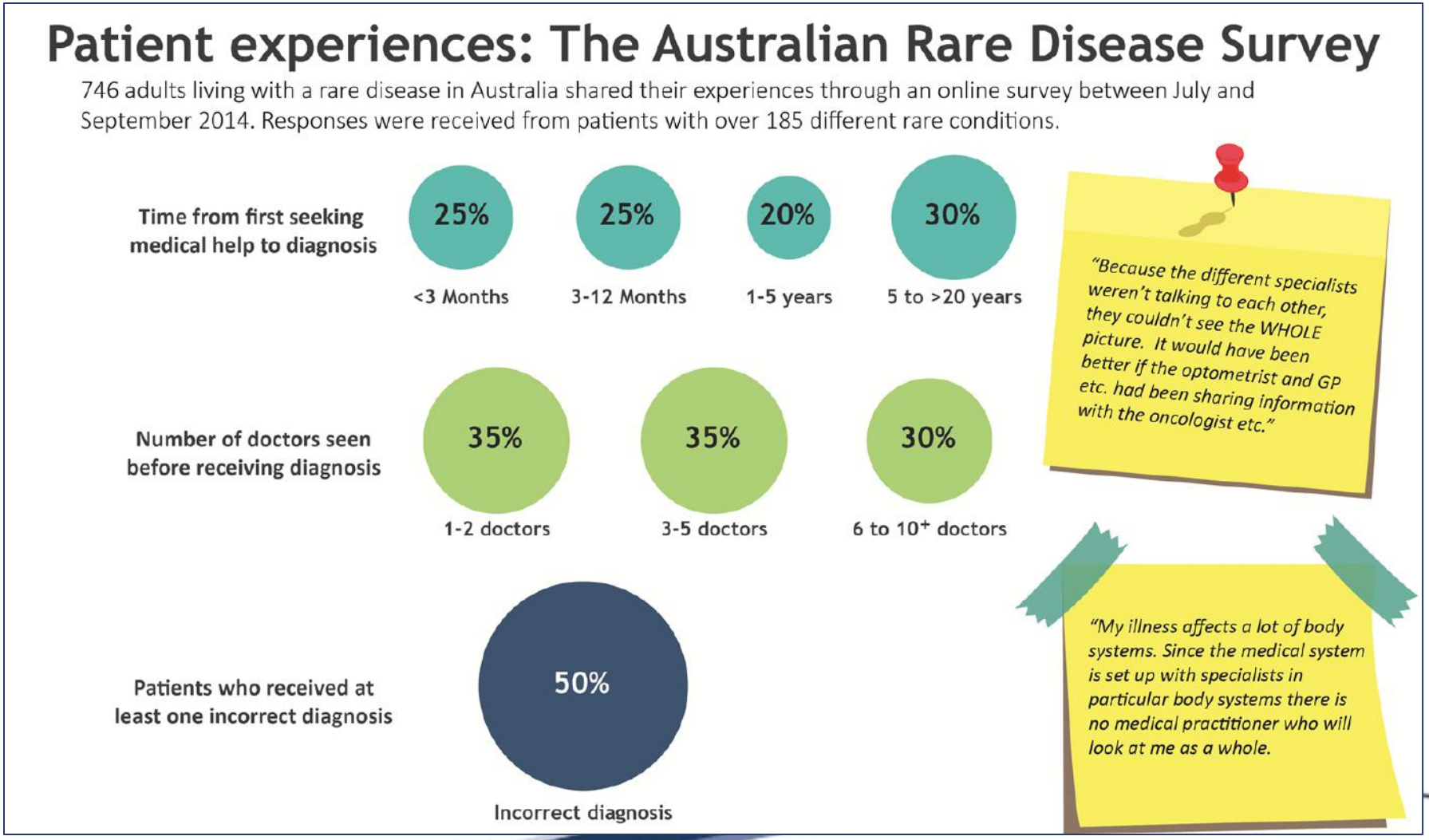
Darwin
+1.5 hrs
+2 hrs
Australia
Brisbane
+2.5 hrs
Perth
+3 hrs
Sydney
Adelaide
Canberra
Melbourne
Hobart
GMT + 8
Shannon Carter & Grace Yun,
Epidemiology Branch, Oct 2013
Government of Western Australia
Department of Health
Public Health and Clinical Services

What special experience do we have in Western Australia?

- Small expert group - only dedicated genomic health policy unit in Australia, set up in 2001
- Developed a State Strategic Framework for Rare Diseases
- Promoting a National Plan for Rare Diseases
- Helped establish Rare Voices Australia
- Leading way in data collection, registry development and biobanks in Australia
- Membership of many international collaborations in this area

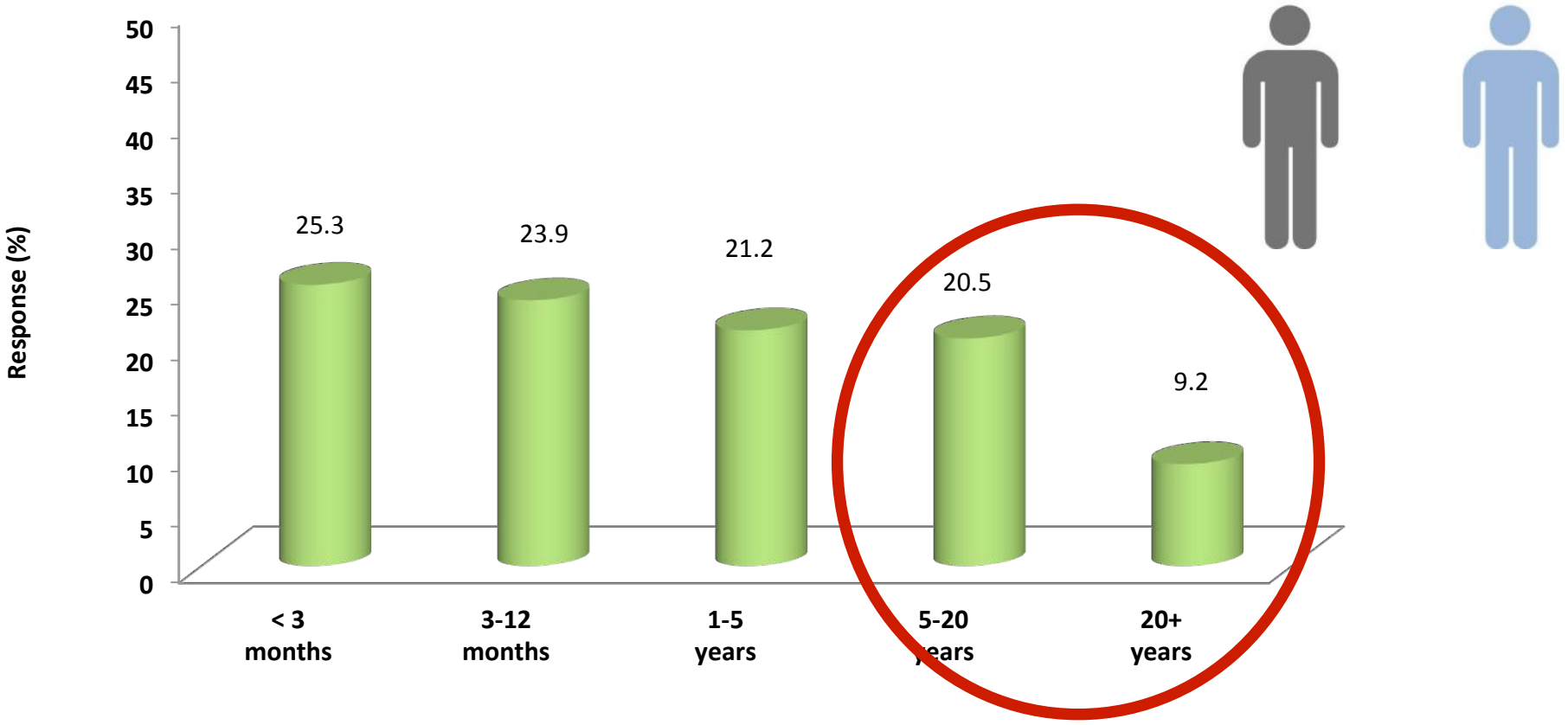
Is this really a problem? The importance of local data

Identifying inefficiencies



The diagnostic journey

1 in 2 people were initially misdiagnosed



Time to confirmed diagnosis after first seeking medical help

- All respondents with a confirmed diagnosis
- n=746

The power of a diagnosis – the foundation of quality healthcare

- Certainty
- Reduced isolation
- Reduce unnecessary investigations
- Access to improved care (social and medical)
- Clarify recurrence risk
- Even without new treatments, quality of life will improve

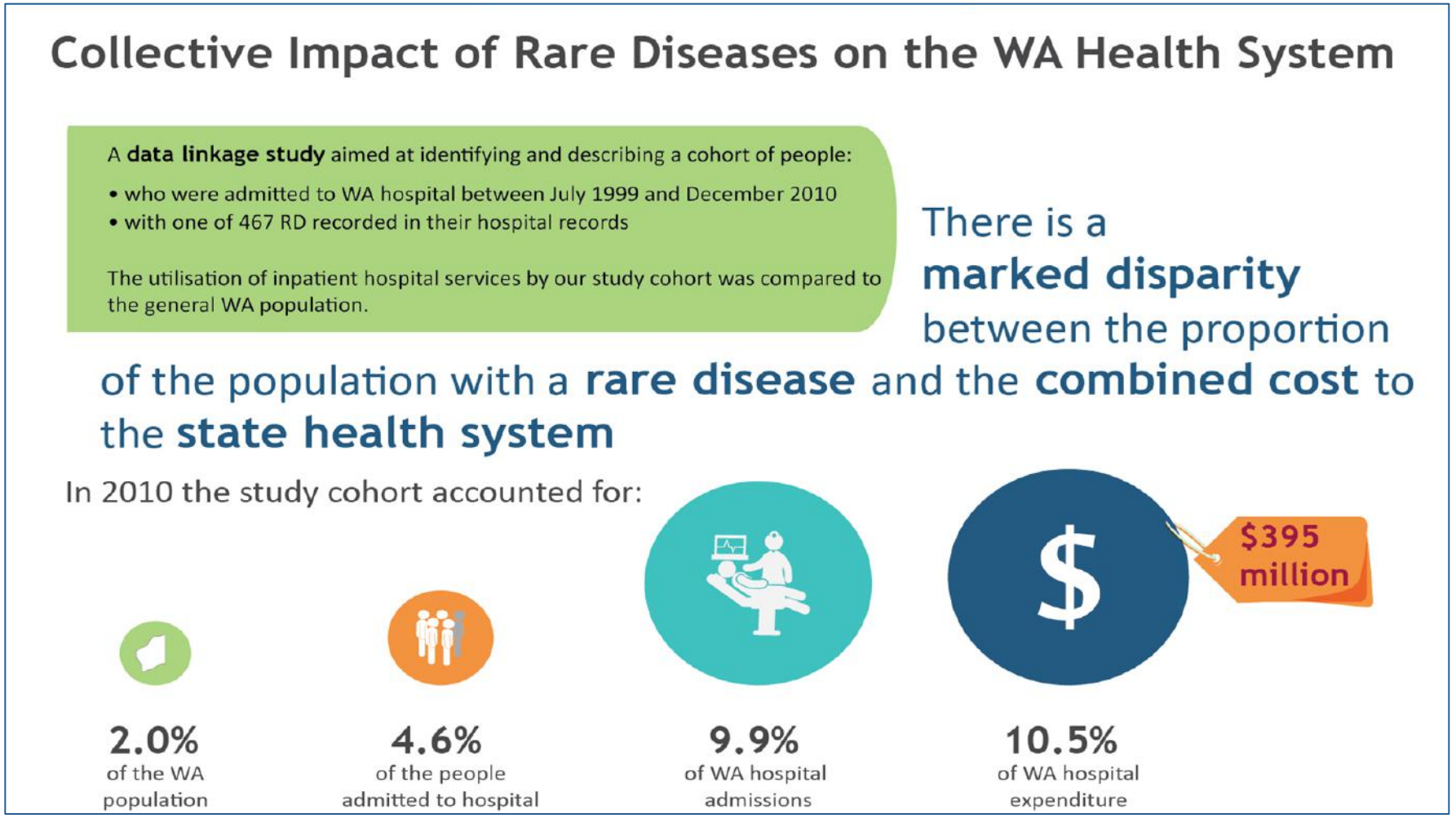
Official journal of the American College of Medical Genetics and Genomics **ORIGINAL RESEARCH ARTICLE** Genetics inMedicine

Open

The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort

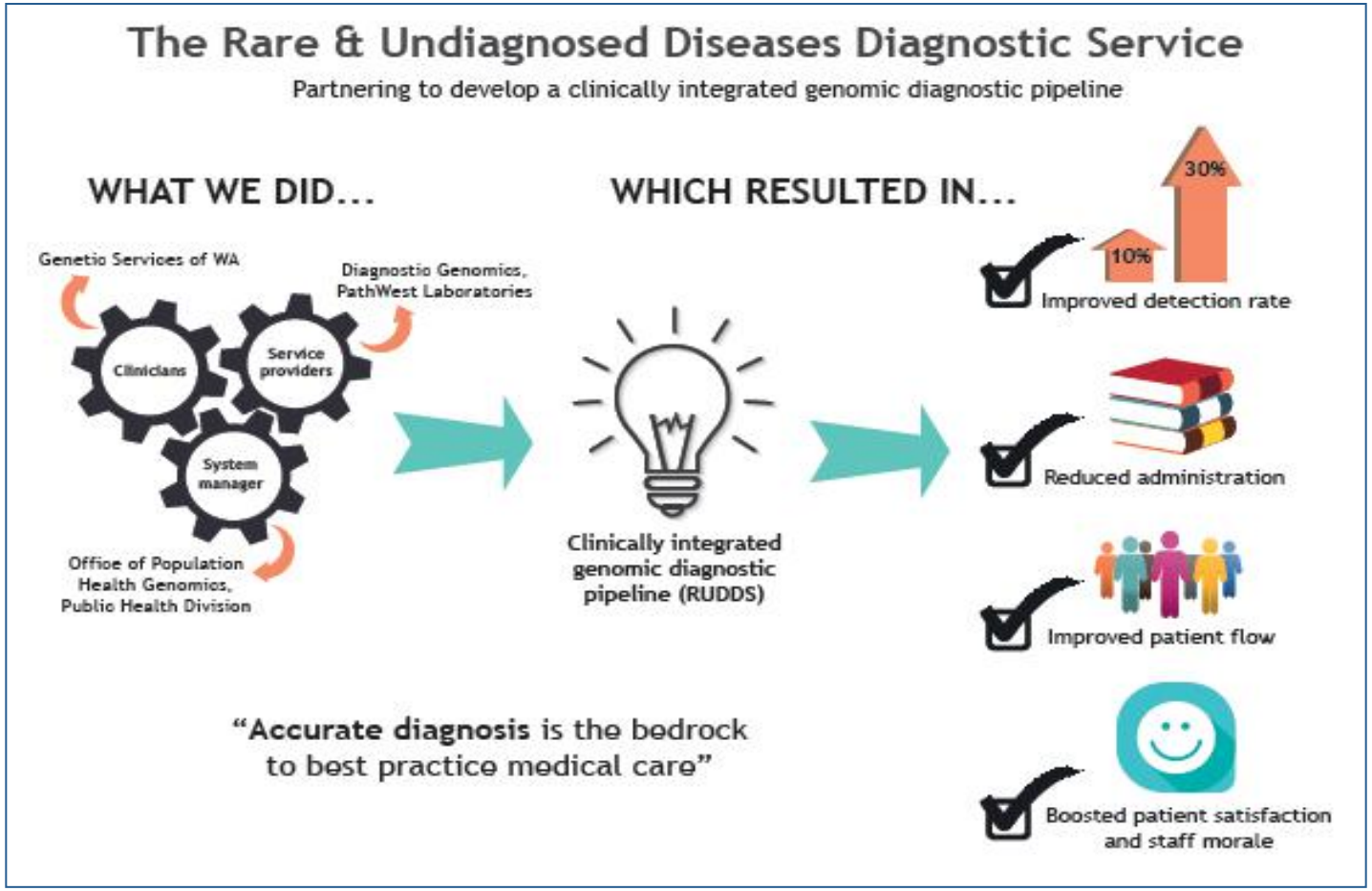
Caroline E. Walker, BSc(Hons), PhD[1], Trinity Mahede, BSc(Hons), MPH[1], Geoff Davis, BA(Hons), PGDipPH[2], Laura J. Miller, MA, PhD[3], Jennifer Girschik, PhD[3], Kate Brameld, BSc(Hons), PhD[4], Wenxing Sun, BHlthSc, GDipBiostats[3], Ana Rath, MD[5], Ségolène Aymé, MD, PhD[5], Stephen R. Zubrick, MSc, PhD[6,7], Gareth S. Baynam, FRACP, PhD[1,7–11], Caron Molster, BBus(Hons)[1], Hugh J.S. Dawkins, BSc(Hons), PhD[1,4,12,13] and Tarun S. Weeramanthri, PhD, FRACP[14]

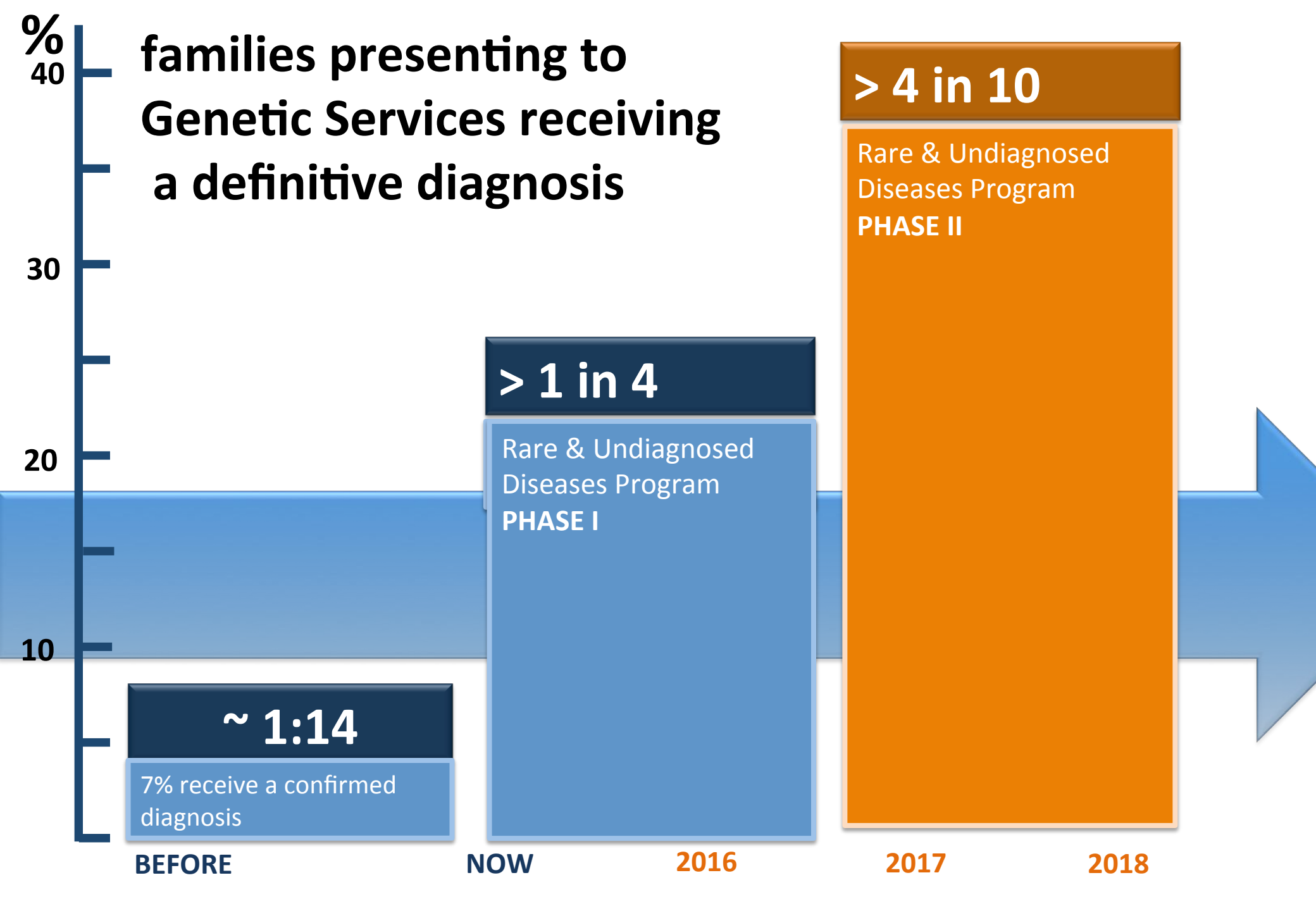
Identifying costs to health system



What can be done about it?
Clinical Services Redesign

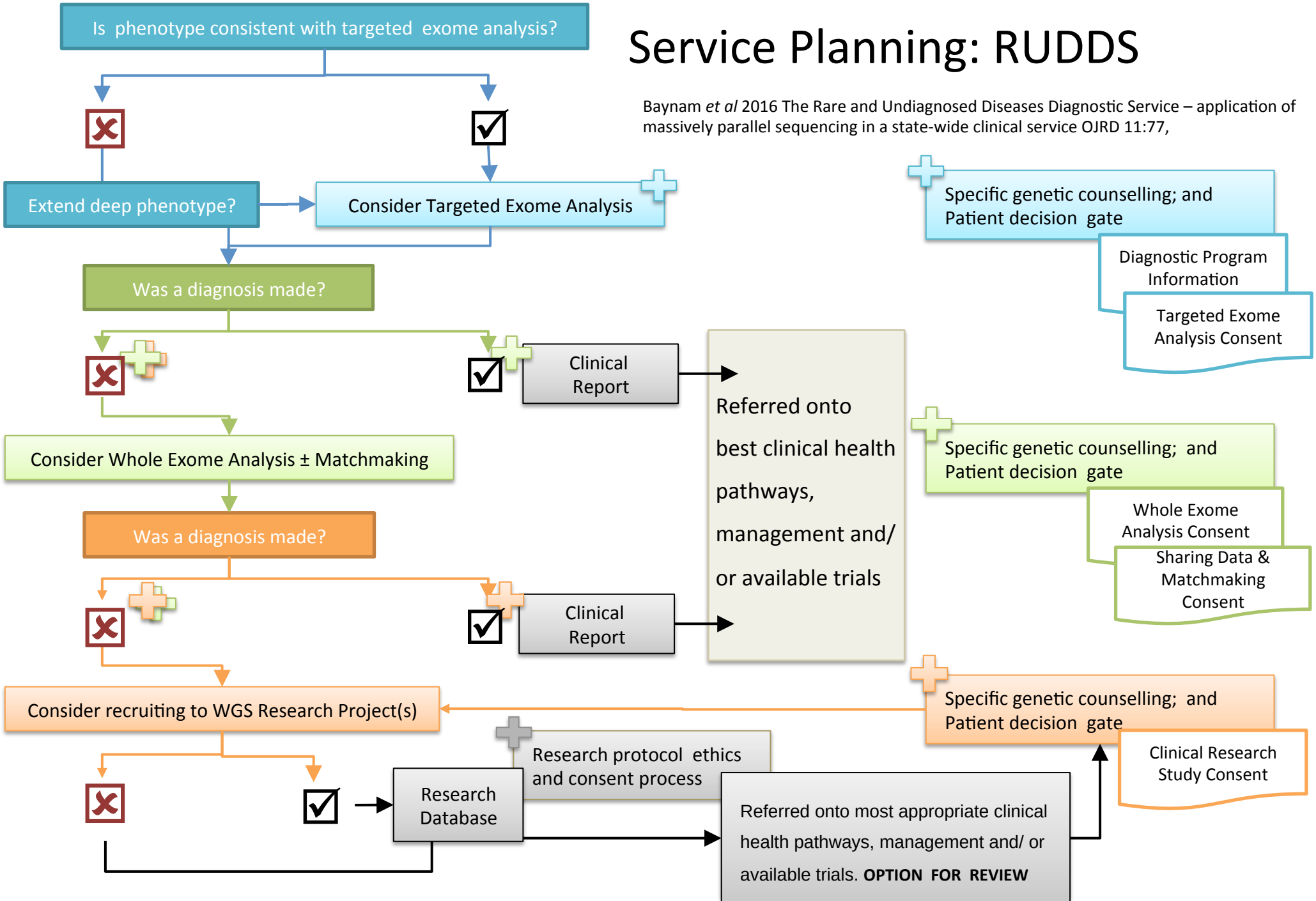
Improving service outcomes




%
40
30
20
10
families presenting to Genetic Services receiving a definitive diagnosis
~ 1:14
7% receive a confirmed diagnosis
> 1 in 4
Rare & Undiagnosed Diseases Program
PHASE I
> 4 in 10
Rare & Undiagnosed Diseases Program
PHASE II
BEFORE
NOW
2016
2017
2018

Service Planning: RUDDS

Baynam *et al* 2016 The Rare and Undiagnosed Diseases Diagnostic Service – application of massively parallel sequencing in a state-wide clinical service OJRD 11:77,



Government of Western Australia
Department of Health

What an Undiagnosed Diseases Program would do for WA

Rare diseases and the diagnostic odyssey

Rare diseases (RD) are a health priority. They are estimated to affect up to 6–8% of the population which in WA is up to 190,000 people, including more than 60,000 children. Many RD have their onset in childhood, continue for life, and are disabling and burdensome to individuals, families and the healthcare system.

An accurate diagnosis is the bedrock of best practice medical care. For RD achieving a diagnosis is particularly challenging. There are 5,000–8,000 known RD and most are complex with multisystem dysfunction.

Many patients experience a diagnostic odyssey. In a European study, 25% of individuals waited 5-30 years for a diagnosis and in 40% of instances the initial diagnosis was wrong (2). A recent WA lead study showed similar findings (3).

What is an undiagnosed disease?

An undiagnosed disease is a long-standing medical condition for which the health system has been unable to provide a diagnosis.

An Undiagnosed Disease Program for Western Australia

GENETIC SERVICES OF WESTERN AUSTRALIA

The health system could say...

- We have more comprehensively addressed the needs of individuals and families living with undiagnosed diseases.
- We can further partner with patients in the development of new management approaches.
- We can benefit from health savings.
- We can be lead partners in global health networks.
- We can further support clinical training and clinical translational research.

Families could say...

- We have closure.
- We are less isolated.
- We better understand what the future might (or might not) hold.
- We have avenues for better treatment, disorder specific medicines or best practice medical care.
- We have improved engagement with the health system.
- We can make financial savings.
- We have improved emotional well being.

Those not receiving a definitive diagnosis could say...

- We have closure for our family.
- The avenues to pursue a diagnosis have been further and more cohesively explored.
- We are less isolated, through connection with the community of undiagnosed individuals e.g. through UDP-related resources and relevant organisations such as Syndromes Without A Name (SWAN) and the Genetic and Rare Diseases Network WA (GaRDN).
- We have improved medical care by integration with relevant services and/ or specialists.
- We can give insight into better management and contribute to the development of new therapies.
- We have improved engagement with the health system.

Undiagnosed Diseases Network

The Undiagnosed Diseases Program and Network

The Undiagnosed Diseases Program was established within the USA National Institutes of Health (NIH) in 2008. It has become a global network of clinical genetics centres, using multidisciplinary teams, to provide diagnoses for patients with severe undiagnosed diseases.

Many UDN patients have previously visited multiple specialists, have had many hospital admissions and a myriad of investigations.

Diagnosis for those who had none

As a consequence of having no diagnosis, patients, and their families experience anxiety, uncertainty and sometimes inappropriate management of their condition.

In these most diagnostically intractable cases 25% have received a definitive diagnosis. This figure is higher when in children.

Health System Savings

For adult patients, direct costs accrued within the health system prior to assessment by the UDN was estimated to be a minimum of US$ 36,000 (AU$ 49,000) per patient.

With an early, accurate diagnosis much of this cost would have been averted. Future savings will also accrue along with an individuals life span

Preliminary assessments by the UDN suggest that the cost per patient diagnosed is less than a single admission in a tertiary hospital.

Paediatric costings are begin finalised and are anticipated to reveal similar high pre-existing costs and savings opportunities.

References

1. Department of Health Western Australia, *WA Rare Diseases Strategic Framework 2015-2018*, 2015.
2. EURORDIS, *Survey of the delay in diagnosis for 8 rare diseases in Europe*, EurordisCare2, 2007.
3. Molster et al., *Health care experiences of adults living with a rare disease in Australia*, In preparation, 2015.
4. Lacbawan et al., Costing the Diagnostic Odyssey: The UDP-NIH Experience

What an Undiagnosed Diseases Program would do

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- We can further support clinical training and clinical translational research.

Undiagnosed Diseases Program UDP-WA:
Synopsis of 1st Case

- A 7 year old girl with more than 40 hospital visits since a baby.
- Seen multiple specialists
- The cross-disciplinary expert review process converged on a class of conditions, which on further exploration by the team focused to one condition
- A definitive diagnosis of a complex messenger RNA disorder with a prevalence of about one in 1 million people.
- The child has now been referred into the appropriate management pathways and carries a diagnosis, outcomes for which the family are greatly appreciative.

Photo: Case files of 1st UDP-WA case (right stack) but missing 3 current files still in circulation

STUDENTS 4 RARE DISEASES

SOLVED

GENETIC SERVICES OF WESTERN AUSTRALIA

Phenotate

Together, we will build a curated library of accurate, machine-readable disease annotations for differential diagnosis software.


The Weekend W

April 30-May 1, 2016 thewest.com.au

GUIDE TO BALI
TRAVEL SPECIAL
LIFTOUT INSIDE

LITTLE GIRL IS ONE IN A MILLION
NEWS PS


Rare syndrome: Seven-year-old Lily Fretwell, right, is not much bigger than her sister Indigo. 2. Picture: Ian Munro

Tiny Lily has a one-in-a-million conditior

Cathy O'Leary

Seven-year-old Lily Fretwell is not much bigger than her two-year-old sister Indigo because of baffling slow growth since she was in her mother's womb.

Now a WA program investigating medical mysteries has solved its first case, discovering that Lily is one-in-a-million who has an extremely rare genetic syndrome.

After hundreds of tests since her birth, Lily's parents Dorota and Adrian finally know why she has an array of unusual health problems and is the size of a three-year-old.

Although there is no cure for trichohepatoenteric syndrome, doctors can better monitor her health.

The syndrome affects the liver, immune system and intestines and can cause skin spots and brittle hair.

"Lily was tiny from seven weeks into my pregnancy and had to be tube-fed for the first few years, and had lots of infections, but we never knew why," Mrs Fretwell said.

"We got used to lots of results which said 'negative' or 'inconclusive'.

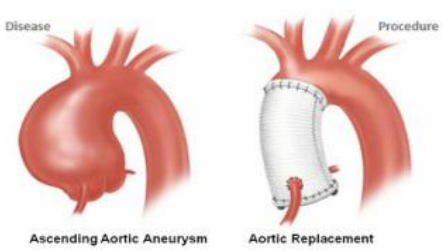
"While she is still very small, her health is relatively OK now and she's at school in Year 2 doing well."

It is the first diagnosis made by WA's Undiagnosed Diseases Program, an Australia-first project.

Director Gareth Baynam said Lily would no longe have costly and invasiv tests to determine the caus of her condition.

"This is a child who ha undergone more tests an has spent a longer time i hospital in her short life than many adults would in a lifetime," Dr Baynam said.

"We hope having a diag nosis means spending less time in hospital so she and her family have more time enjoying life."



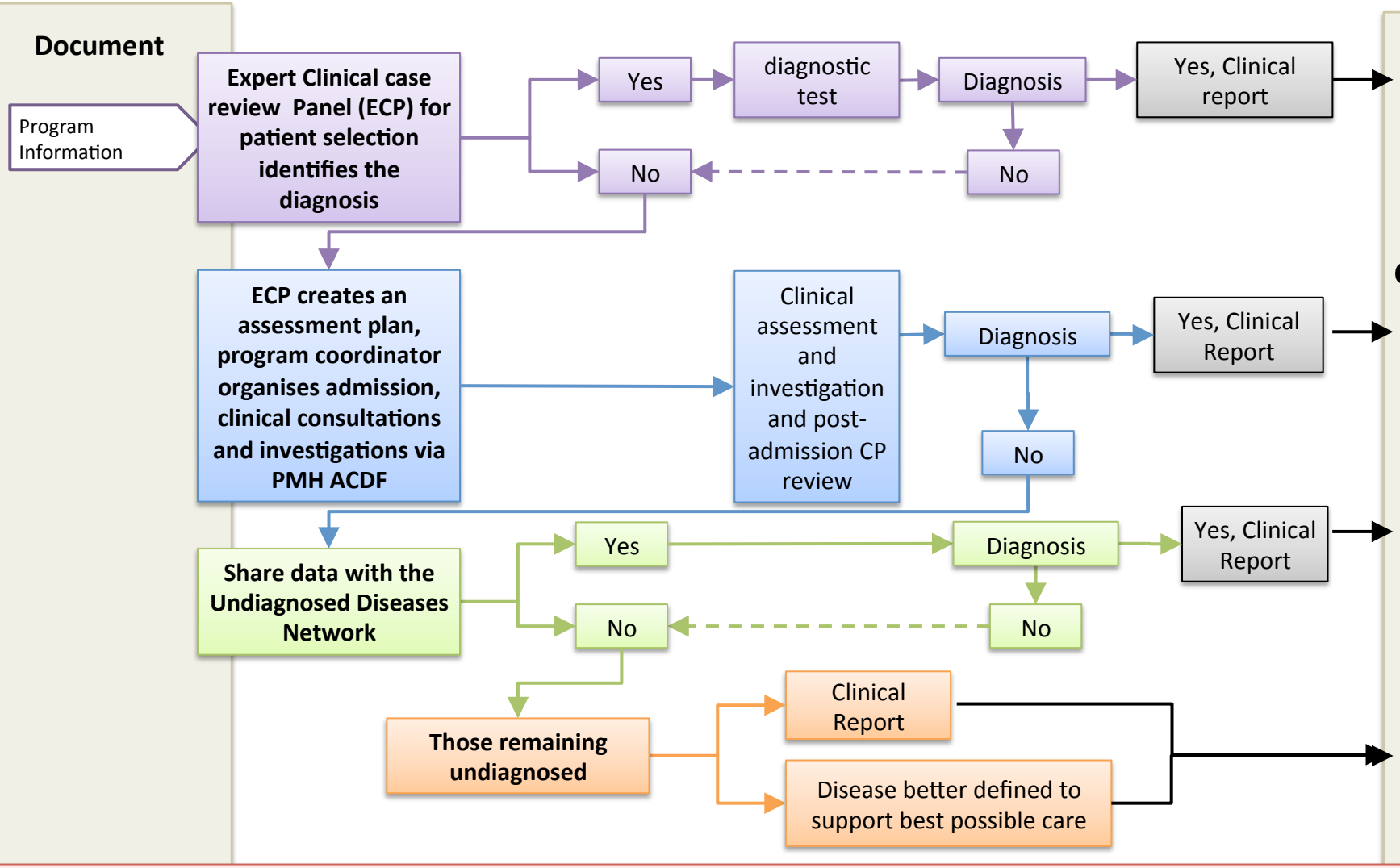
GARVAN INSTITUTE OF MEDICAL RESEARCH

TELETHON KIDS INSTITUTE

Undiagnosed Diseases Program

Government of **Western Australia**
Department of **Health**
Child and Adolescent Health Service

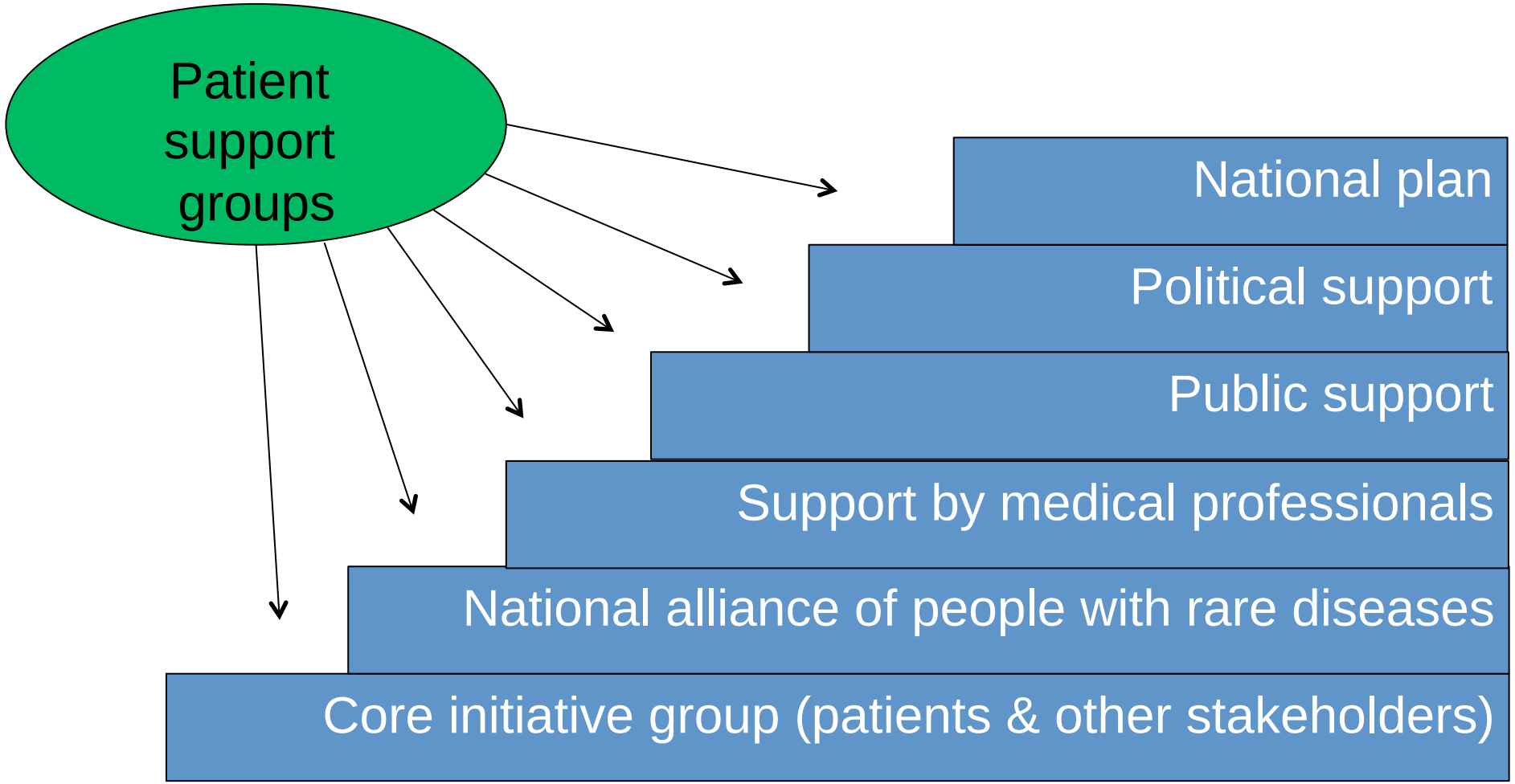
Cases selected by a multidisciplinary consultant expert team on the basis of clinical need and indicators in favour of achieving a diagnosis
Genetic Counselling offered throughout the Program | Option to leave the Program at any time



ALL CASES/FAMILIES RECEIVE AN OFFER TO PARTICIPATE IN RELVANT STUDIES AND FOR THEIR DATA TO BE USED IN ETHICALLY APPROVED STUDIES
ALL CASES/FAMILIES HAVE THE OPTION TO LEAVE THE PROGRAM AT ANY TIME

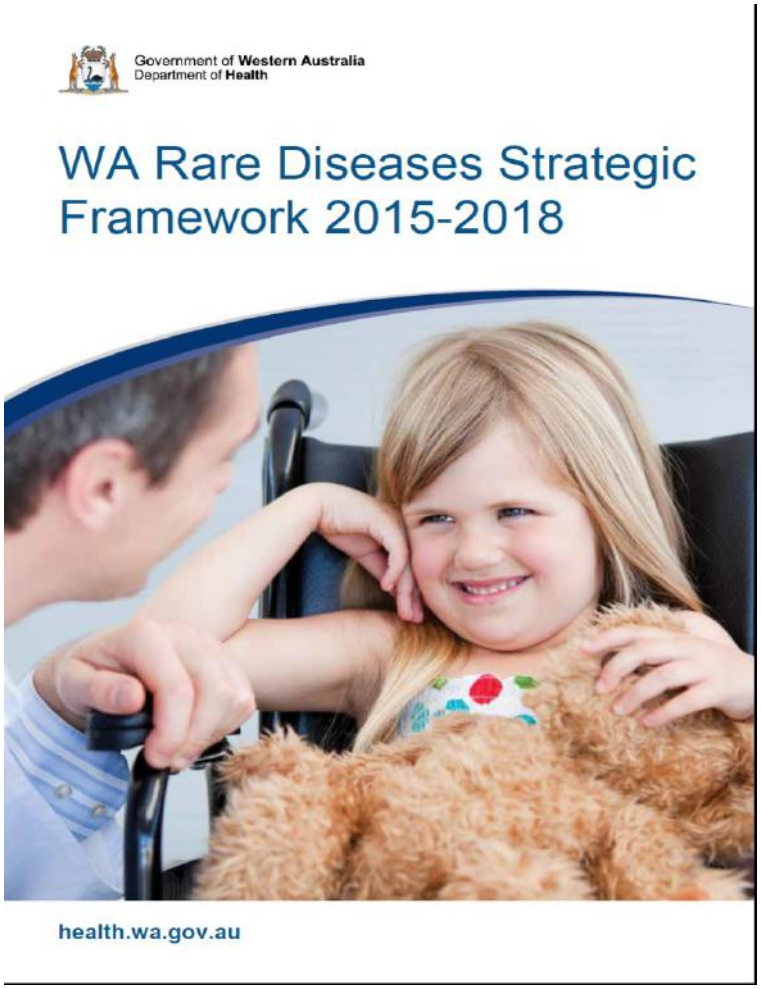
What can be done about it?
Policy and planning

Roadmap for policy and planning



Taruscio et al 2010

Developing statewide policy



VISION: The best possible health and wellbeing for Western Australians living with rare diseases (80% of which are genetic)

- Recognises rare diseases as a public health priority
- Provides a framework for the coordination of initiatives
- A focus on identifying gains in efficiency and effectiveness of services provided within the WA public health system

Developing national policy

- Newborn Bloodspot Screening (NBS) National Policy Framework

- A nationally consistent approach to NBS that:
  - ✓Supports best practice
  - ✓Increases transparency
  - ✓Strengthens governance
  - ✓Supports decision-making
  - ✓Offers guidance for monitoring and evaluation

What can be learnt from past experience?

Recommended content for the Australian National Rare Diseases Plan (2011)

- high level policy direction, that includes a collective definition of rare diseases;
- guidance to adopt ICD-11 classification of rare diseases and the collection of much needed data (surveillance);
- information for all stakeholders, to guide referral, diagnoses, treatment and access to support services;
- a map of the services available to people with rare diseases;
- best practice guidelines;
- a coordinated research approach; and
- opportunities for consumers to engage in rare disease planning and policy development.

Putting the argument – learning from our 'failure' in 2013 to gather support for an Australian Rare Diseases Plan

1. Concern about resources needed in an already stretched system (duplication of services, extra services, cost benefit of services, where would money come from?)
2. Professionals – lukewarm support from some existing disease specific groups, contributed to lack of other states supporting us
3. Patient support groups – lack of long-term experience with a combined advocacy group. Rare Voices Australia set up in 2012
4. Many useful learnings however – a setback only. Still advocating and moving forward in 2016!

Government of **Western Australia**
Department of **Health**

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.

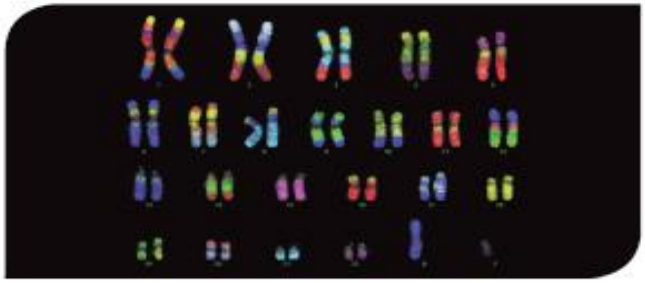
Reports from consumers suggest that care for rare diseases is fragmented and uncoordinated and thus there is an opportunity to improve efficiency and effectiveness of the health system.

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.

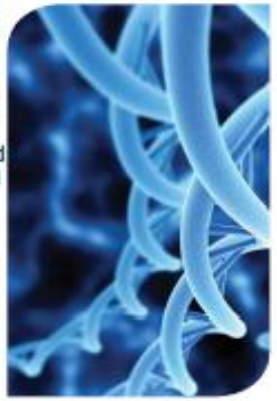
Health care professionals would be able to say…

- 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.

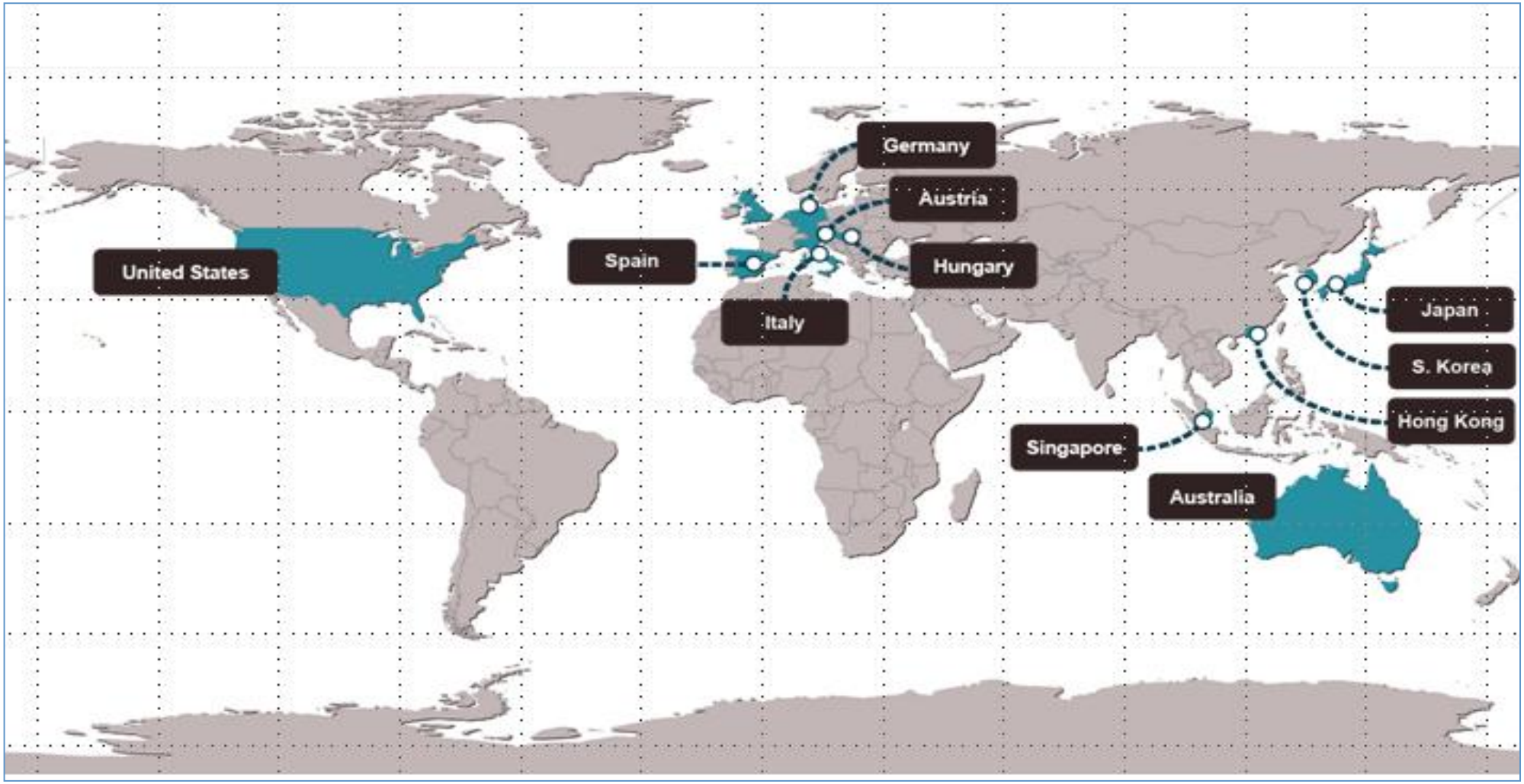
Produced by the Office of Population Health Genomics

E: genomics@health.wa.gov.au
T: (08) 9222 6888

Delivering a **Healthy WA**

International experience – how to copy and paste

Undiagnosed Diseases Network International, UDN-I, 2015 - 2016



ORPHANET USERS:
20,000 user accesses daily from over 200 countries

2/3 Professionals
1/3 Patients and relatives

International partnerships

1. International Rare Diseases Research Consortium – means to diagnose most rare diseases by 2020, and 200 new therapies by 2020
2. Undiagnosed Diseases Network International
3. Orphanet – portal for rare diseases and orphan drugs - 20,000 users daily from all countries, 2/3 professional, 1/3 patients and relatives
4. Global Alliance on Genetics and Health – resource for ethics, data sharing and policy
5. Rare Diseases International – global alliance of people living with a rare disease
6. UN NGO Committee for Rare Diseases

NEW PATIENT ORGANISATION INITIATIVES

Rare Diseases International (RDI)

EURORDIS-led initiative (E**urope**), in partnership with National Organization for Rare Disorders (**USA**), **Canadian** Organization for Rare Disorders, **Japanese** Patient Association, **Chinese** Organization for Rare Disorders, **Indian** Organization for Rare Diseases, **Ibero-American Alliance** for Rare Diseases (ALIBER), **French Alliance** for Rare Diseases (Alliance Maladies Rares), International Patient Organization for Primary Immunodeficiencies (IPOPI), Dystrophic Epidermolysis Bullosa Research Association International (DEBRA International), among other groups.

- to create the global alliance of rare disease patients and families.
- to be a strong common voice on behalf of the people living with a rare disease around the world,
- to advocate for rare diseases as an international public health priority, and
- to represent/ enhance the capacities of its members

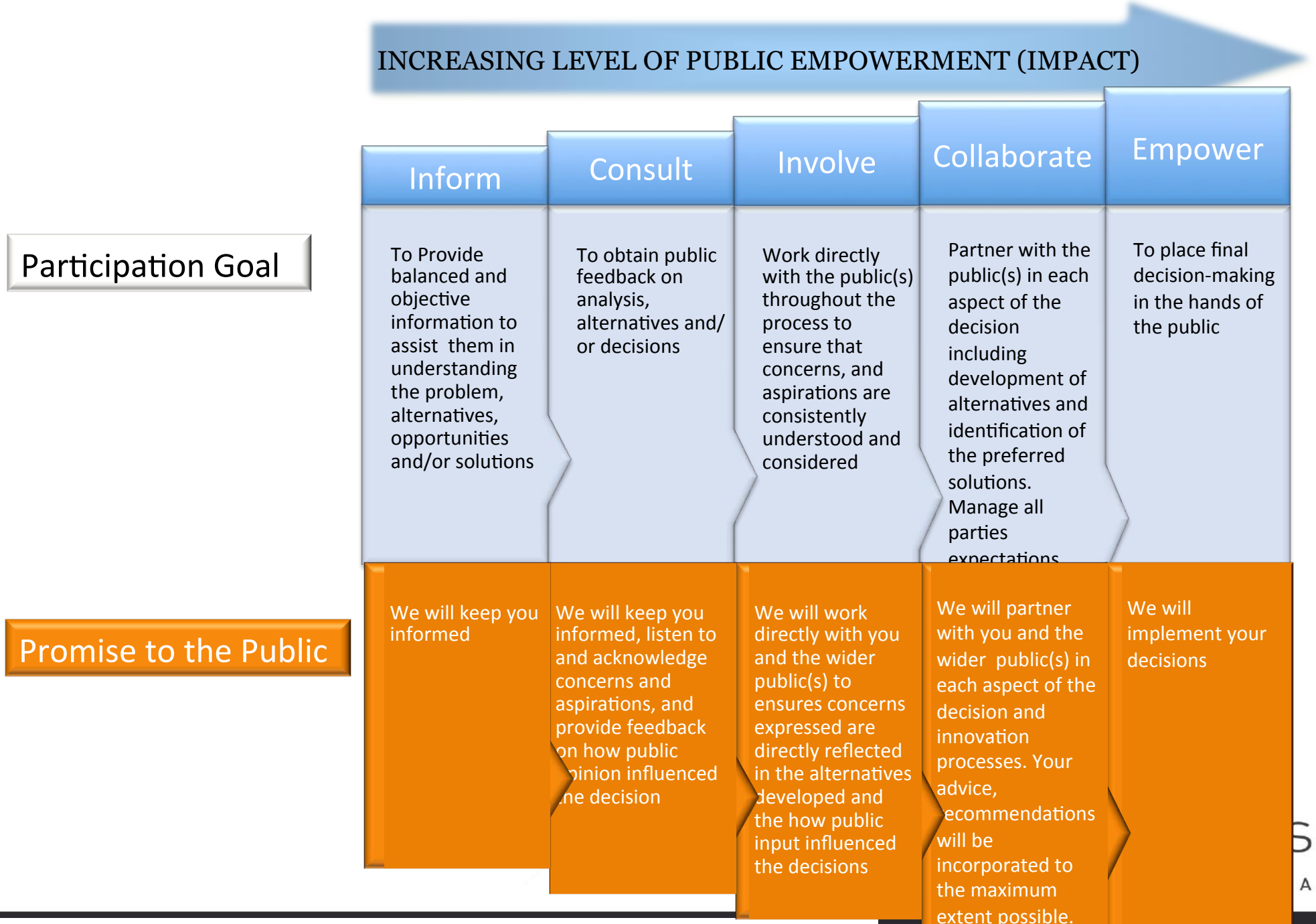
UN NGO Committee for Rare Diseases

a multi-stakeholder, inclusive, global ecosystem focused on rare diseases, which aims:

- To increase visibility of rare diseases at the global level
- To extend and share knowledge about rare diseases and their unmet needs
- To connect NGOs interested in rare diseases and their partners within a global platform
- To promote international, multi-stakeholder collaboration and actions for rare diseases
- To align rare diseases as a global priority in public health, research and medical and social care policies

The formal inauguration of the Committee is due to take place at the UN headquarters in New York in November 2016

rare voices AUSTRALIA



Adapted from the International Association for Public Participation : IAP2 Spectrum of Public Participation

Collaboration and partnerships – government & peak rare and genetic diseases groups

Government of **Western Australia**
Department of **Health**
Office of Population Health Genomics

- A small group in government can catalyse a large amount of energy and networks
- National and international partnerships and networks are available and *share freely* – you just have to join
- You can choose the path forward that fits you and the timeframe – there are plenty of ‘small wins’ at the beginning

International Partners

Rare diseases: what will you ask for?

- Draw on your own experience and words – ‘a difficult diagnosis, lost in the dark, left behind and running low’
- Ask for something
- Don’t ask for more than three things
- Ask for advice (politicians, policy makers etc.)
- Don’t ask for money or specific treatments - ask for an open transparent system of resource allocation
- Reassure them about your ongoing role and support
- Know your friends! Insiders and outsiders together achieve great things, but have different roles
- Show them that you are not ‘conflicted’

Terimah kasih