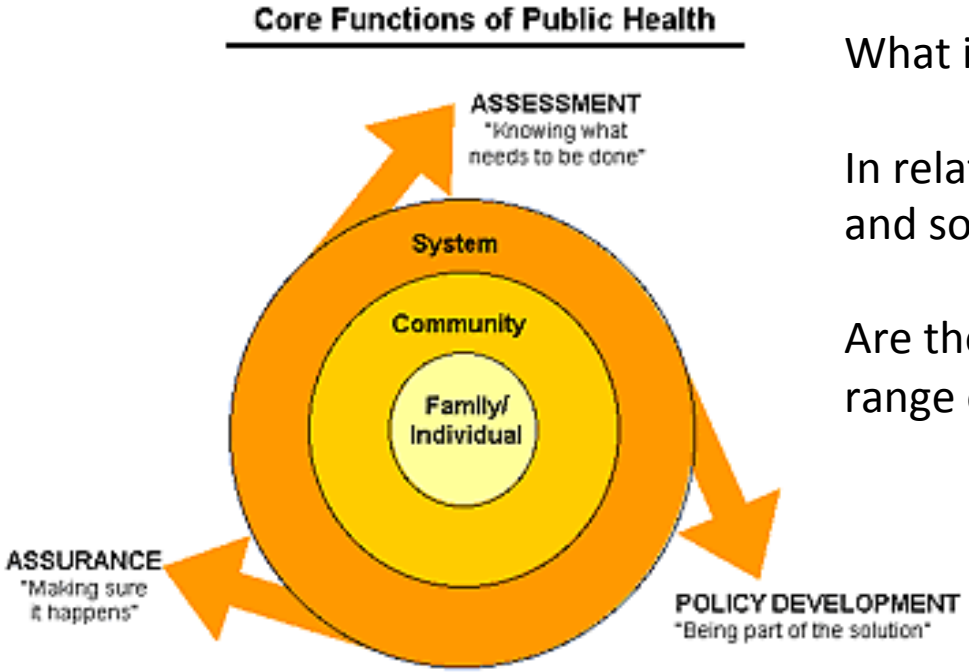
Impact of Rare Diseases

Caron Molster
Deputy Director, Office of Population Health Genomics,
Department of Health,
Government of Western Australia


Looking at the impact of rare diseases from a public health & government point of view



What is the local impact of RD?

In relation to what aspect of health and social care?

Are there common impacts across a range of areas?

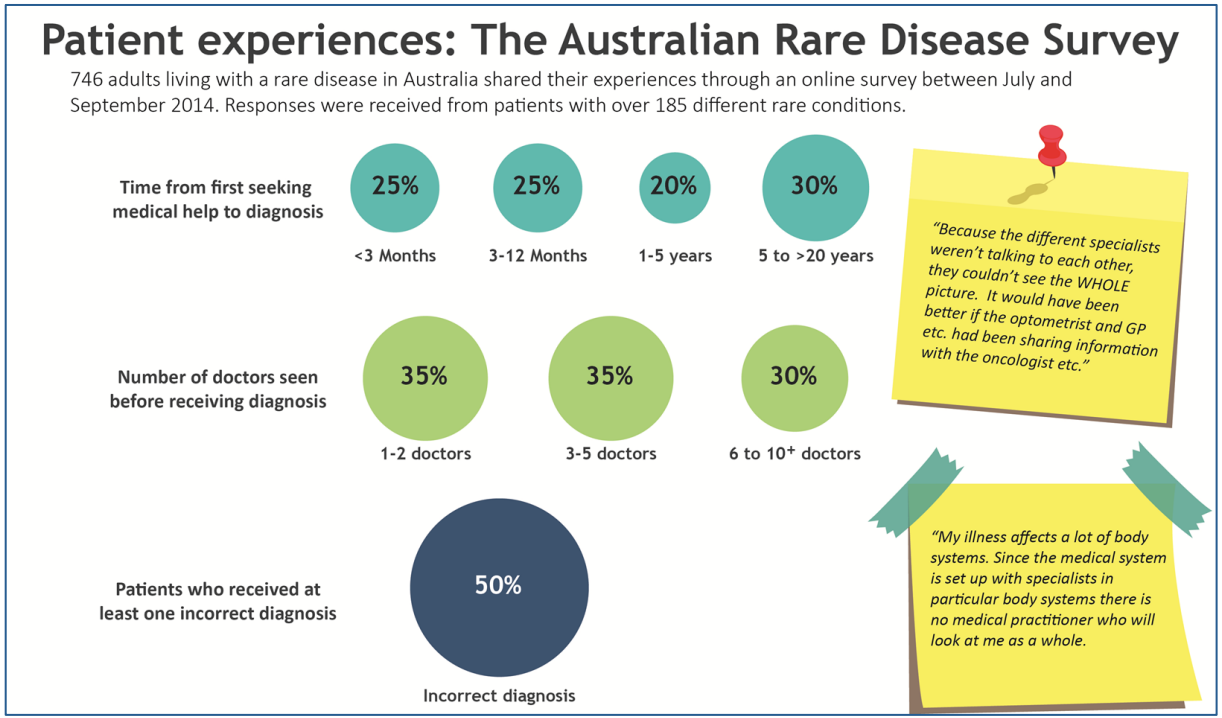
Are services and interventions addressing the impact?

What can be done about the impact?

Which perspective on impact?

- Individuals
- Families/carers
- Communities
- Health system

Assessing the impact – the diagnostic journey



Molster et al. 2016 Survey of healthcare experiences of Australian adults living with rare diseases, Orphanet Journal of Rare Diseases

Assessing the impact – the diagnostic journey

"Apart from the financial cost, the time, energy, emotional and psychological resources required to persist in this process…are unsustainable".

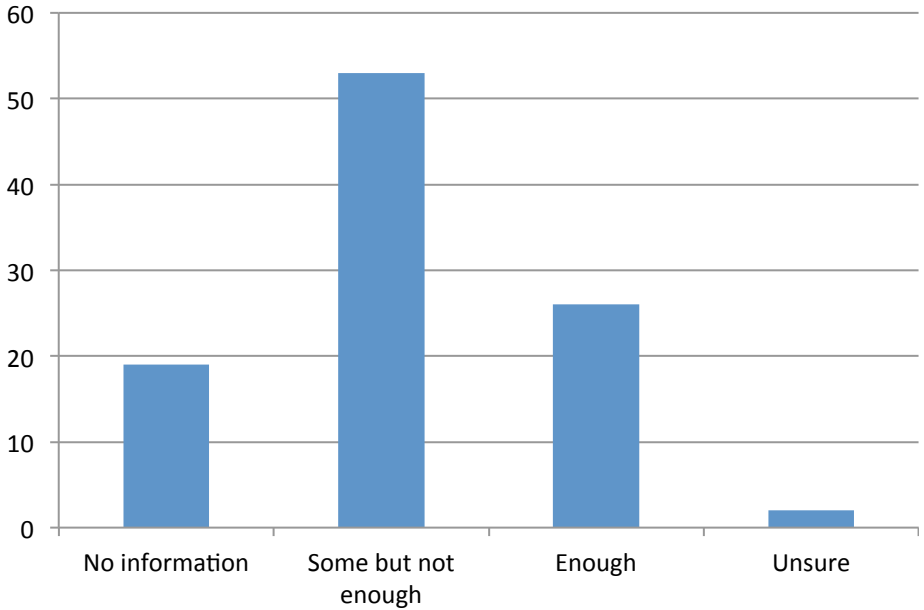
Assessing the impact – the diagnostic journey

The undiagnosed

‘ . . . we spent 27 weeks in hospital, we couldn't access any support at all . . . we’re sat there going “right, you can't help us but you can help the family across the way…because they've got a diagnosis . . . they can access everything” and it’s so frustrating.’
[Parent][1]

1 Genetic Alliance UK 2016 The hidden costs of rare diseases – a feasibility study

Information at time of diagnosis



"There was a lot I wasn't told. The diagnosis was given in a 10-minute appointment which is insufficient time to advise someone about diagnosis of a rare, life-changing illness".

50% didn't understand all information given

Assessing the impact – care coordination

Need multiple specialists and other health professionals to manage care
Vast majority did not have a health professional to coordinate their care

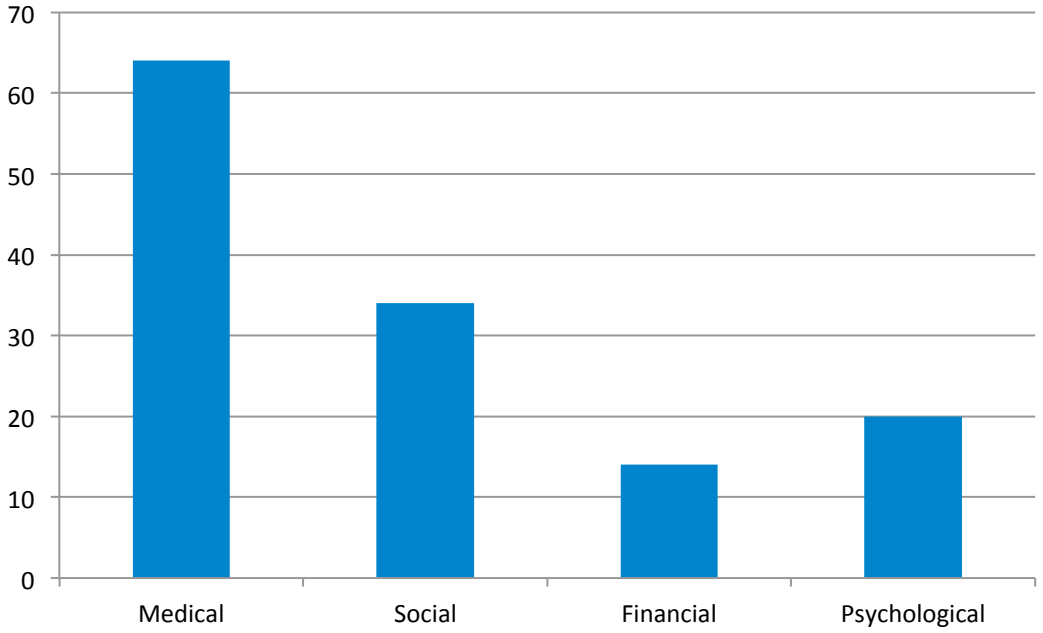
Patients and families face significant ('hidden') costs associated with the way that their care is managed[1]

Financial	Other
Costs associated with appointments: Time off work and reduced income; childcare; travel including petrol, public transport and taxis; parking; food and refreshments; accommodation; sundries; accessible vehicles and transport options.	**Time:** Time off work; time spent coordinating ('project managing') care and the various agencies and appointments involved; time spent fighting to access care and support.
Other financial costs associated with wider condition management: Private healthcare; childcare and respite; specialist activities and equipment; IT, internet and telephone costs (including paper and printing cost); prescriptions; fees for informal helpers and carers; disruption to employment and income.	**Psychosocial, health and well-being:** Disruption to schooling, employment and personal time; impact on relationships and social life; isolation; impact on identity and sense of self; living with uncertainty; mental health; fatigue; confidence and self esteem; anxiety and stress associated with appointments.
Wider family: Costs identified above related to patients, parents and grandparents; siblings and wider support networks.	

1 Genetic Alliance UK 2016 The hidden costs of rare diseases – a feasibility study

Assessing the impact – lack of support

"It's exhausting to need to keep pushing and I think the mental health support needs to be increased".



Received sufficient support

Collective Impact of Rare Diseases on the WA Health System

A **data linkage study** aimed at identifying and describing a cohort of people:

- who were admitted to WA hospital between July 1999 and December 2010
- with one of 467 RD recorded in their hospital records

The utilisation of inpatient hospital services by our study cohort was compared to the general WA population.

There is a **marked disparity** between the proportion of the population with a **rare disease** and the **combined cost** to the **state health system**

In 2010 the study cohort accounted for:

2.0%
of the WA population

4.6%
of the people admitted to hospital

9.9%
of WA hospital admissions

10.5%
of WA hospital expenditure

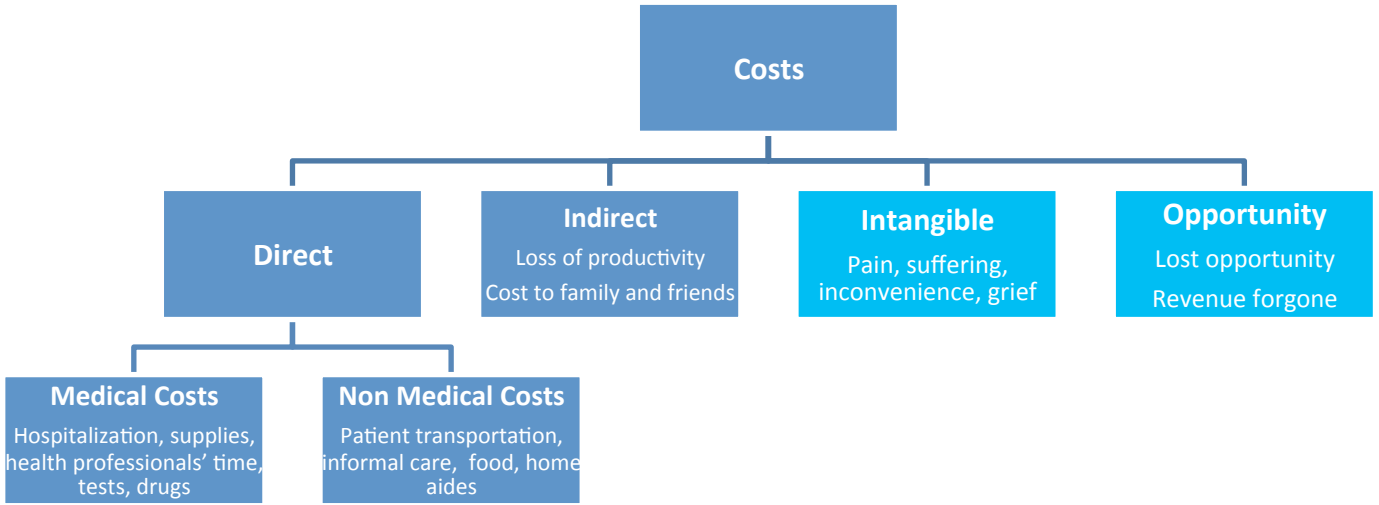
BURQOL-RD project

- Socio-economic burden of 10 RD
- Health-related quality of life
- Cost-of-illness (COI) studies
- Literature review
- Direct costs – medical, non-medical
  - E.g. time invested in appointments, drugs, equipment, hospital care, home care, transport, specialist education
- Indirect costs – productivity loss
  - E.g. time lost from paid labour, unpaid labour, lost leisure time, employer costs of paid sick leave, loss of income, unemployment

"It is clear that the evidence base for the COI at individual, health system and societal level is fairly poor when it comes to understanding the pressures faced by both individuals, families and society in the context of RDs"[1]

1 Angelis et al. 2015 Socio-economic burden of rare diseases: a systematic review of cost of illness evidence

Other important costs



Assessing the impact – more local evidence

The economic and social impact of rare diseases on individuals, the health system and society

Costs of treating and managing RD

Identify gaps in existing evidence

Recommendations for future research

Literature review

Methodology

Policy development – the problem and solution

- What is the local impact?
- Why is it an important issue?
- What needs are to be addressed?
- What can be done about it?
- What will be the benefit?
- To who?
  - Patients/families/carers
  - Health professionals
  - Health system

Policy development – what can be done?

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

- 12 objectives
  - Facilitate access to support networks and information
  - Build on existing services for screening and diagnosis
  - Promote care coordination
  - Champion integration and partnerships in the delivery of healthcare
  - Facilitate health professionals' access to information
  - Build epidemiology and health system evidence
- Foundation: more local evidence required!

Assurance – linking people to services

"We connect key stakeholders and service providers with people affected by genetic and rare diseases"

GENETIC AND RARE DISEASE NETWORK

Building capacity of community service organisations

Provide increased access to resources, support and information on health and other services

To link people living with RD, their carers and families to existing healthcare and other services

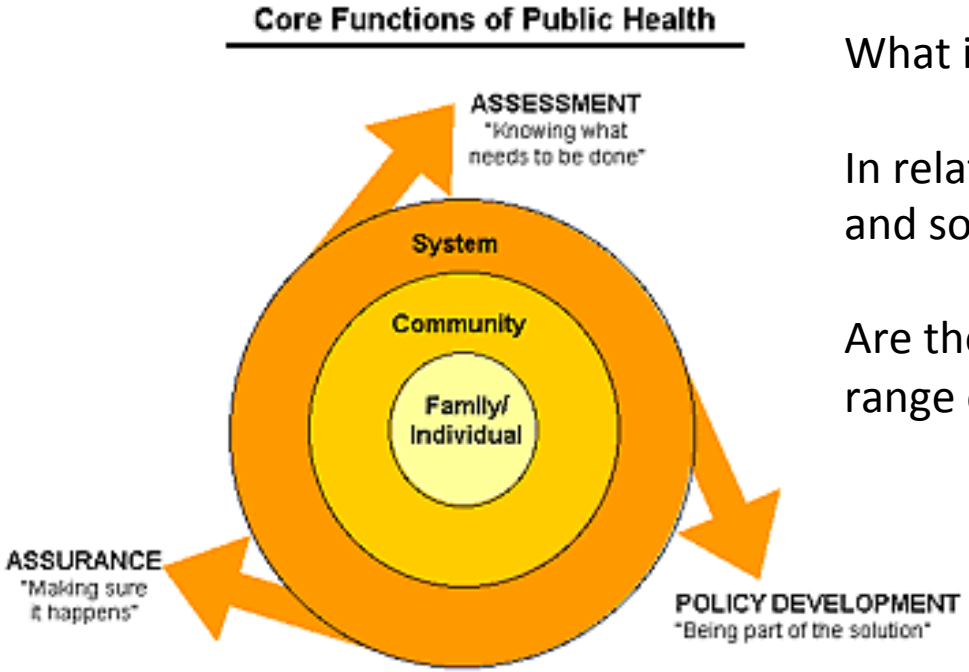
Assurance – linking people to services

Undiagnosed Diseases Program WA (UDP-WA)

Target group: Children who remain undiagnosed despite numerous hospital admissions and specialist assessments across multiple disciplines.

Program steps

1. Case referred to program. Program Director invites parents or carers to take part.
2. A multi-disciplinary Expert Panel reviews existing medical history and makes recommendations.
3. Patient attends a day facility at children's hospital for up to five days for tests and examinations.
4. With patient consent data is shared with national and international partners.
5. The UDP-WA team determines if a definitive diagnosis can be made.
6. Parent/caregiver attends a meeting with the Program Director to discuss the findings and receives a written report.



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