From research translation to transformation in a public health system

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• A simple narrative about what Australia has been doing since it began to wake up to rare diseases
• Recognising that 6-8% of our population was invisible in the health system
• Rare disease families wanting to be visible, too be able to live the best life possible
  – A speedy accurate diagnosis
  – Equitable place in the health system
  – Assurance of receiving best practice care
  – Gaining visibility and a voice
• What changes have we witnessed in Australia as a result of translating global initiatives locally
Closing Slide

• What will the rare diseases landscape be in your country in 2027?

• What new knowledge will you take home to help improve the lives of people living with rare diseases; and

• What will be your starting point/strategy for effecting transformative change
Technology Adoption: Starting points

• Technology (Steel processing technologies; oil & gas discoveries; combustion engine; computing; informatics; genomics …….)

• Innovation and technology adoption occurs in waves
• Resources and society organise around the technology
  – people; investment ; markets, and resources aggregate
• First wave of knowledge translation and benefits accrue in resource rich countries based on conventional investment risk - reward paradigm
• Creates inequities – initially those with the most benefit the most
• Subsequent waves of knowledge translation and benefits are linked to start (entry) point and local resource leverage…equity
Rare Disease

- Recognised public health issue; unmet need
  - Orphan drugs policies
  - Convergent Technologies: molecular & computing
- Catalyst two people agree to organise EC and US NIH resources help better coordinate efforts and aggregate their respective rare disease efforts
- Immediate reaction - a global vision created
- How could Australia participate & meaningfully contribute in this powerhouse initiative?
Demographics

AREA
Australia: 7,691,951 km²
Western Australia: 2,529,875 km²

POPULATION
Australia: 25 million
Western Australia: 2.5 million

POPULATION SPREAD
Australia: 70% live in capital cities
Western Australia: 75% live in Perth

POPULATION DENSITY
Northern Territory: 0.15 people / km²
Western Australia: 0.9 people/ km²
New South Wales: 8 people/ km²
Victoria: 22 people/ km²

Western Australia’s public health system is the largest geographical area in the world covered by a single health authority.
Big Country Small Population

Unmet Needs and issues faced
- At need population within our community
- Rare diseases invisible to the health system
- Dispersed population (urban to remote communities)
- Dispersed resources
- Limited research funds

Outcomes required
- Equitable access
- Optimised outcomes
- Sustainable
- Better use of public health system resources
- Empower rare diseases patients
Our Starting Point

• Be pragmatic: use local strengths & be agile
• Use the public health system, moving towards person-centred care (Equity)
• Access clinical records (Data)
• Health system executive looking for efficiencies and improved patient outcomes (Optimise)
• Empower patient voices (Champions)
• Engage clinical (Champions)
• Create policy frameworks (Sustainable)
What did we do?

• Patient organisations - Surveys – who are our stakeholders and local champions; what can be done
• Clinical services – what is in place, identified champions in the system; what can be done
• Local information – what data are collected, identified champions; what can be done
• Developed a plan – appreciative approach; empowered by stakeholders; what can be done
• Needed to be sustainable – we took a clinical service start point
• Used genomics policy makers
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!
Impact to policy – why did we succeed?

- Built on available evidence
- Have not tried to change the whole system – built on strengths and opportunities
- Political support and champions
  - Linked policy-makers and stakeholders, including patients and patient organisations who gave personal stories
- Recognises more evidence of impact required
Assessing the impact: diagnostic journey

Patient experiences: The Australian Rare Disease Survey

746 adults living with a rare disease in Australia shared their experiences through an online survey between July and September 2014. Responses were received from patients with over 185 different rare conditions.

- **Time from first seeking medical help to diagnosis**
  - 25% <3 Months
  - 25% 3-12 Months
  - 20% 1-5 years
  - 30% 5 to >20 years

- **Number of doctors seen before receiving diagnosis**
  - 35% 1-2 doctors
  - 35% 3-5 doctors
  - 30% 6 to 10+ doctors

- **Patients who received at least one incorrect diagnosis**
  - 50% Incorrect diagnosis

"Because the different specialists weren’t talking to each other, they couldn’t see the WHOLE picture. It would have been better if the optometrist and GP etc. had been sharing information with the oncologist etc."

"My illness affects a lot of body systems. Since the medical system is set up with specialists in particular body systems there is no medical practitioner who will look at me as a whole."

Assessing the impact: health system

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

$395 million


health.wa.gov.au
Needs to be addressed?

- Early, accurate diagnosis
- Information at time of diagnosis
- Multi-disciplinary care
- Integrated, coordinated health services
- Psychological, financial and social support
Assurance – linking people to services

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

- 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
Families presenting to Genetic Services receiving a definitive diagnosis

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,
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
• Case referred to program. Program Director invites parents or carers to take part.
• A cross-disciplinary Expert Panel reviews existing medical history and makes recommendations.
• Patient attends a day facility at children’s hospital for up to five days for tests and examinations.
• With patient consent data is shared with national and international partners.
• The UDP-WA team determines if a definitive diagnosis can be made.
• Parent/caregiver attends a meeting with the Program Director to discuss the findings and receives a written report.
Matchmaker Exchange
All people living with rare disease

• Timely and accurate diagnosis
• Aboriginal genomics
• Enabling optimised best care
What next – pathways to diagnosis

- Ensuring timely accurate diagnosis
  - IRDiRC task forces
    - Solving the unsolved, UDP, UDN International, MME
    - RD-Connect platforms and 3D facial
- Patient Archive – Human Phenotype Ontologies (HPO)
- Orphanet disease coding system
- Developing best practice guidelines
- Primary healthcare pathways
- Patient centred outcomes
- Integrating new knowledge
  - Care4Rare, Phenome Central, Patient Archive, ERN, RDCRN…
What next – health policy

• Ensuring increased visibility
• Ensuring sustainability
• Ensuring Equity and optimised outcomes
• Policy frameworks
  – Standardised disease classification and coding
  – Standardised phenotype language
  – Data sharing
  – Indigenous genomic (reference) data
  – Timely accurate diagnosis
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