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

Advances in Rare Diseases - Australia

a continuing journey and new beginnings

Professor **Gareth Baynam**
Head, Western Australian Register of Developmental Anomalies (congenital anomalies and cerebral palsy registers)
Consultant Geneticist, Genetic Services of Western Australia
Adjunct Policy Advisor, Office of Population Health Genomics, Public and Aboriginal Health Division
Angela Wright Bennett Fellow for Rare and Undiagnosed Diseases
Department of Health
Western Australia

Australasian Rare Disease Landscape

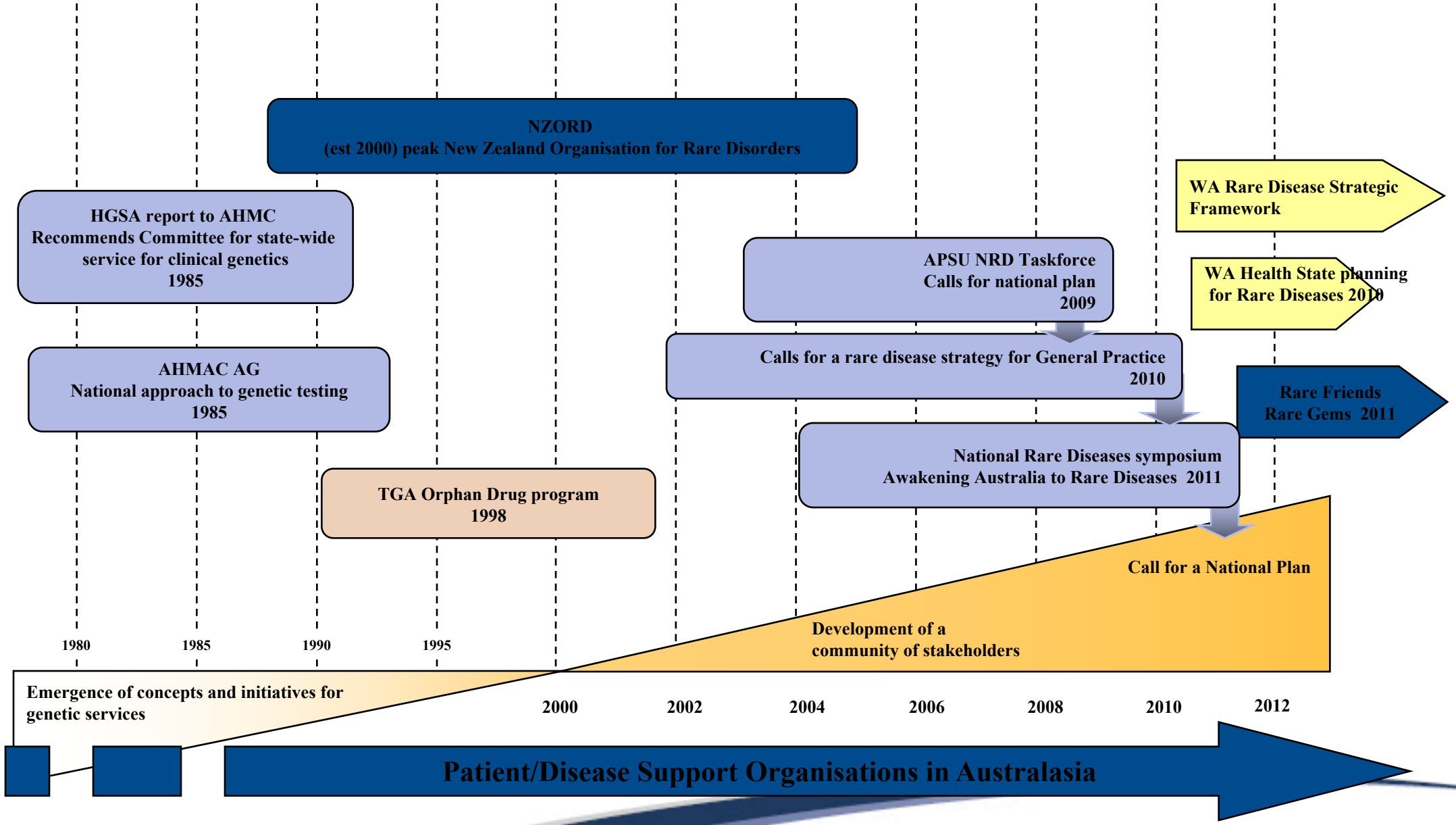
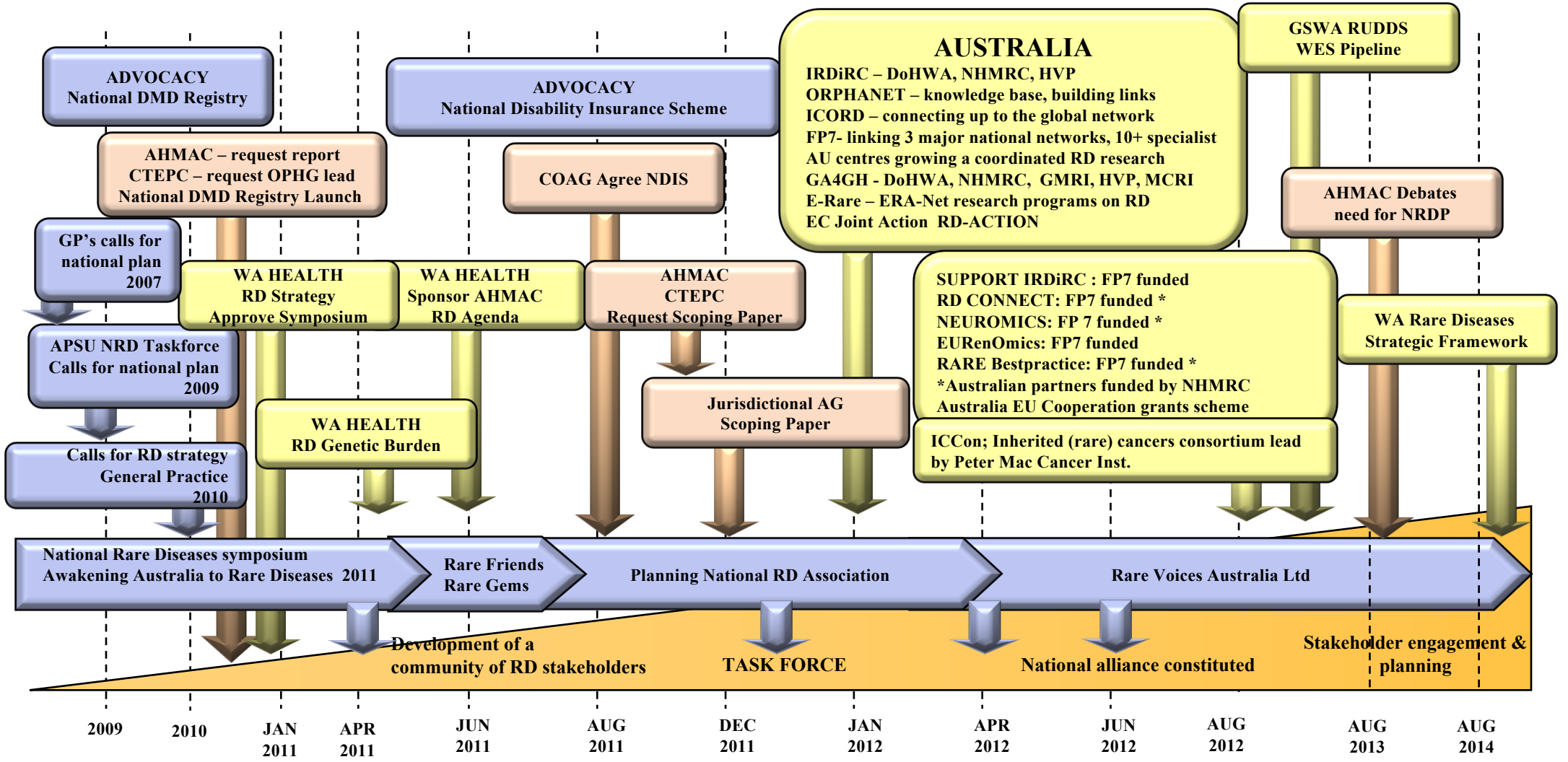


Figure adapted from Orphanet

Australasian Rare Disease Landscape: People are the Catalyst



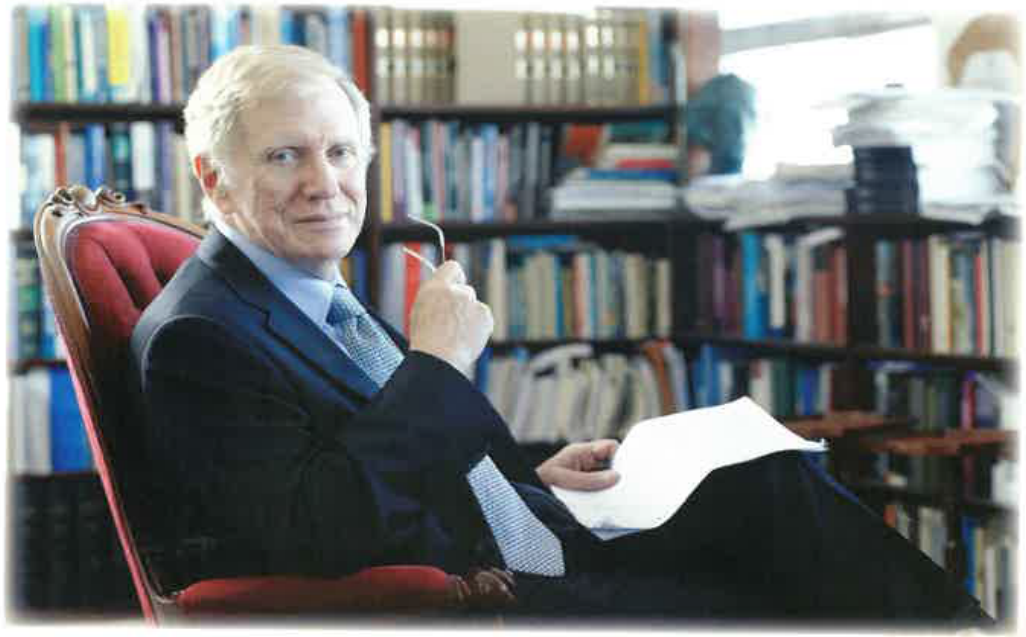
GLOBAL LANDSCAPE

NIH Office of Rare Disease Research (ORDR), NORD, Genetic Alliance,
ORPHANET, EURORDIS, TREAT-NMD, EUROPLAN, EUROPLAN II
IRDiRC, EU Framework Program 7(FP7), Horizon 2020 (FP8); EC JA RD-Action

Figure adapted from ORPHANET

2014: RARE VOICES AUSTRALIA

The Australian Experience of Living with a Rare Disease



The Hon Michael Kirby AC, CMG
Founding Patron, Rare Voices Australia

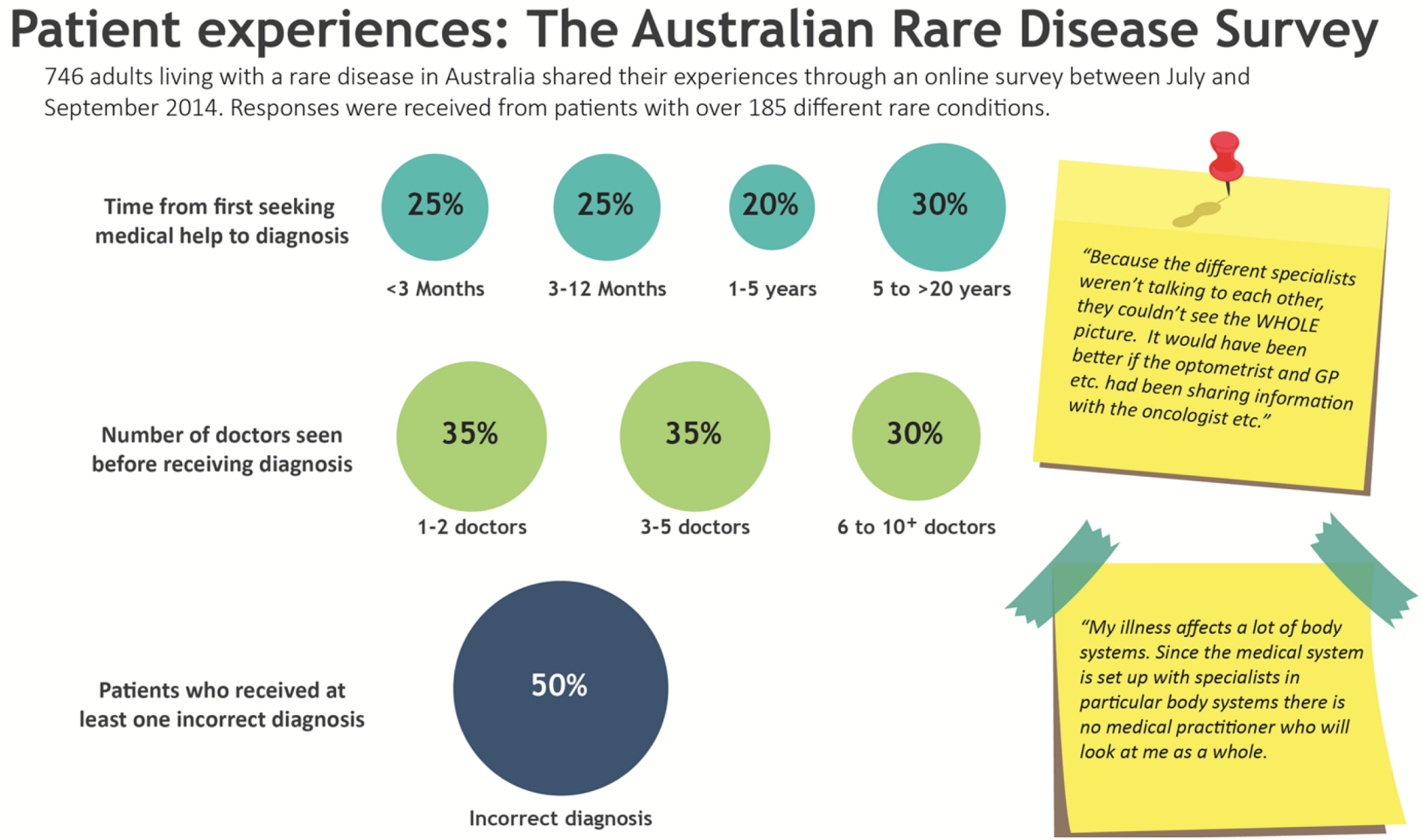
"Every voice has a right to be heard by our leaders. The voices of people with rare diseases are no exception. These people are our fellow citizens and deserve equality in healthcare and research. These voices appeal to human love for one another. We should respond"

Policy development:

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

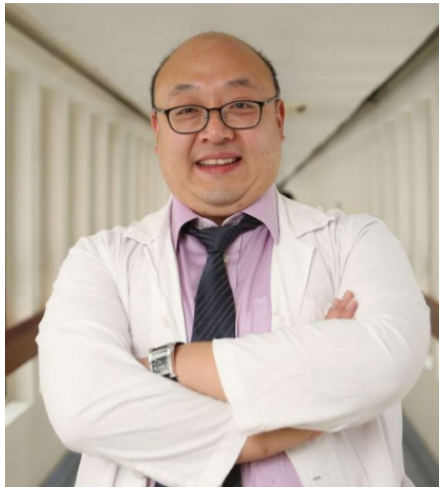
- 12 objectives
  - Facilitate access to support networks and information
  - Build on existing services for screening and diagnosis
  - Promote care coordination
  - Champion integration and partnerships in the delivery of healthcare
  - Facilitate health professionals' access to information
  - Build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

Assessing the impact: diagnostic journey



Molster et al. 2016 Survey of healthcare experiences of Australian adults living with rare diseases, Orphanet Journal of Rare Diseases 11:30 DOI: 10.1186/s13023-016-0409-z

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:

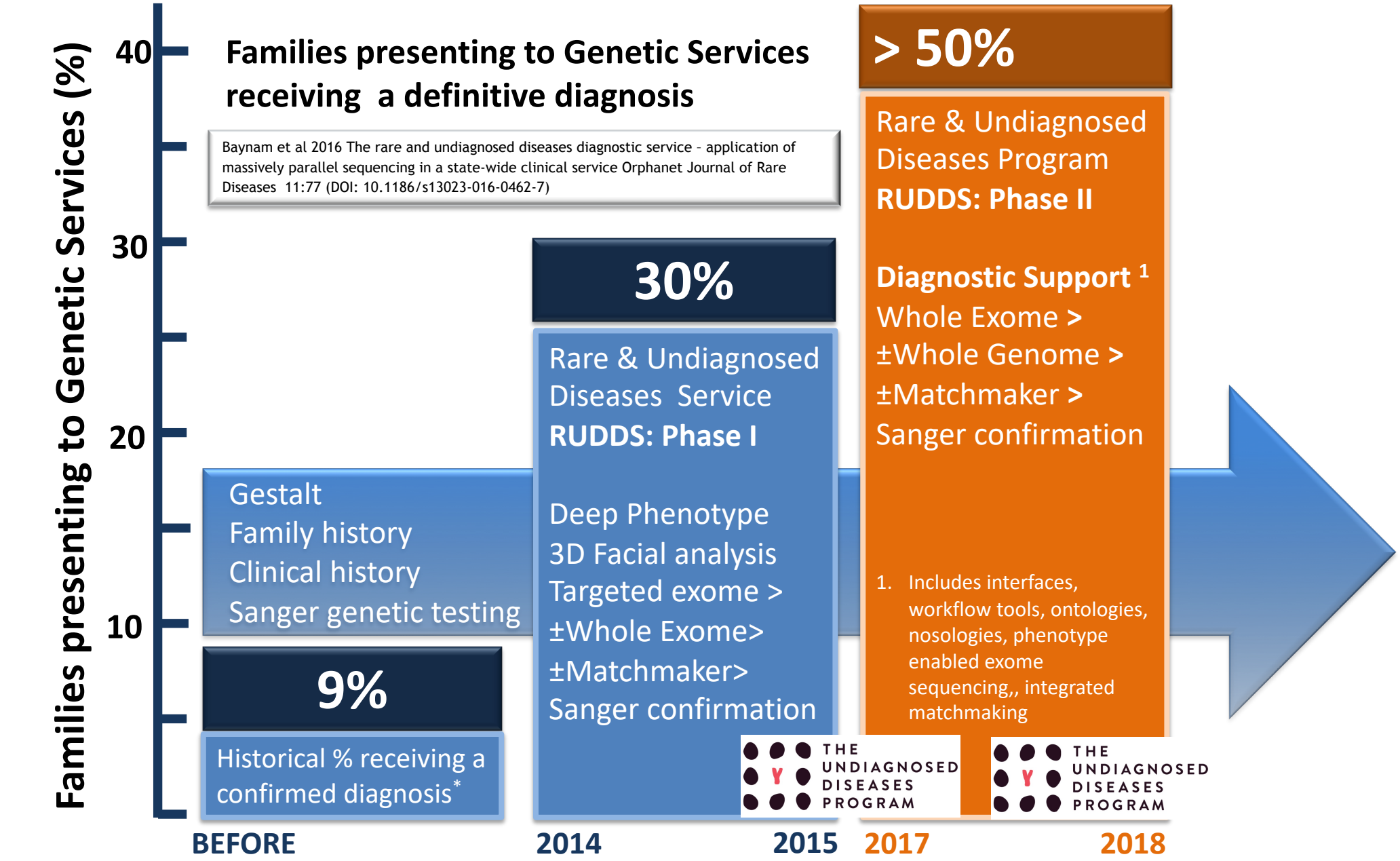
2.0% of the WA population

4.6% of the people admitted to hospital

9.9% of WA hospital admissions

10.5% of WA hospital expenditure

Walker et al. 2016 The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort. Genetics in Medicine : doi:10.1038/gim.2016.143



* Diagnosis in this instance meaning a definitive diagnosis with a high level of certainty with or without a molecular confirmation (pathogenic mutation)

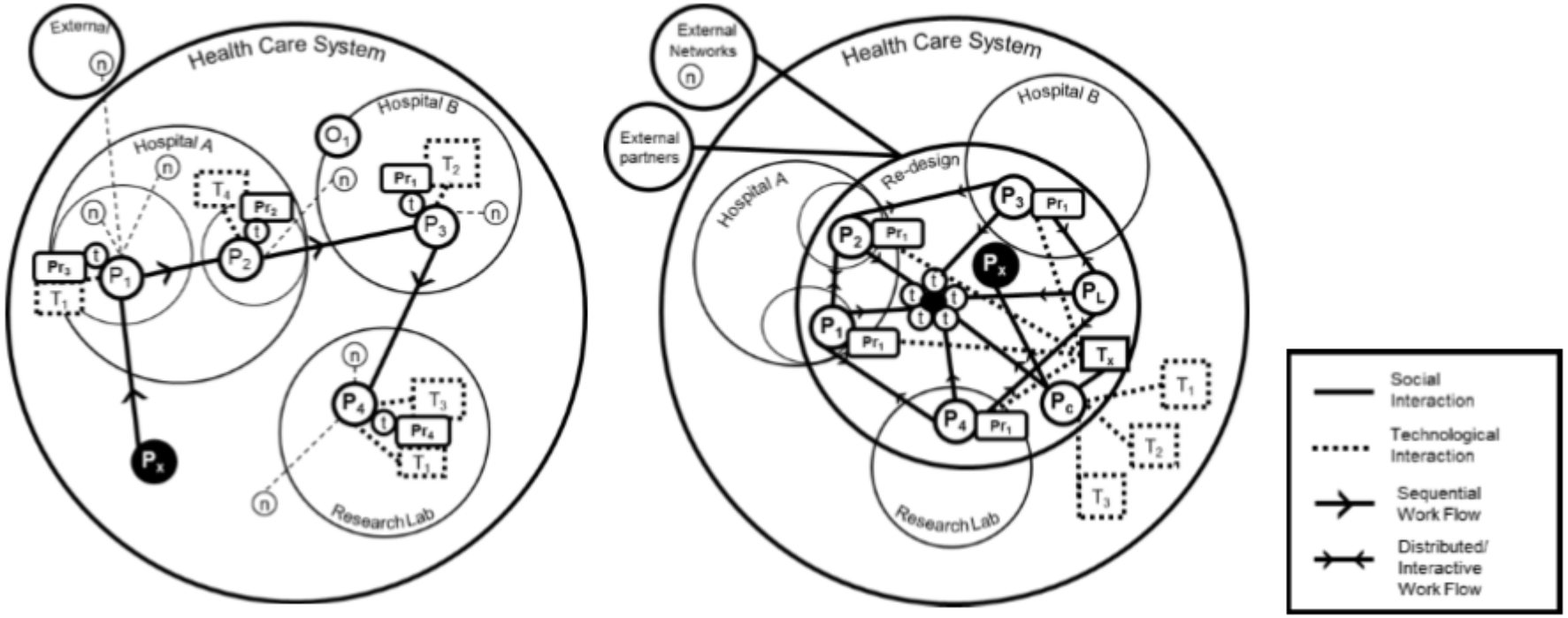
THE UNDIAGNOSED DISEASES PROGRAM

Undiagnosed Diseases Network INTERNATIONAL

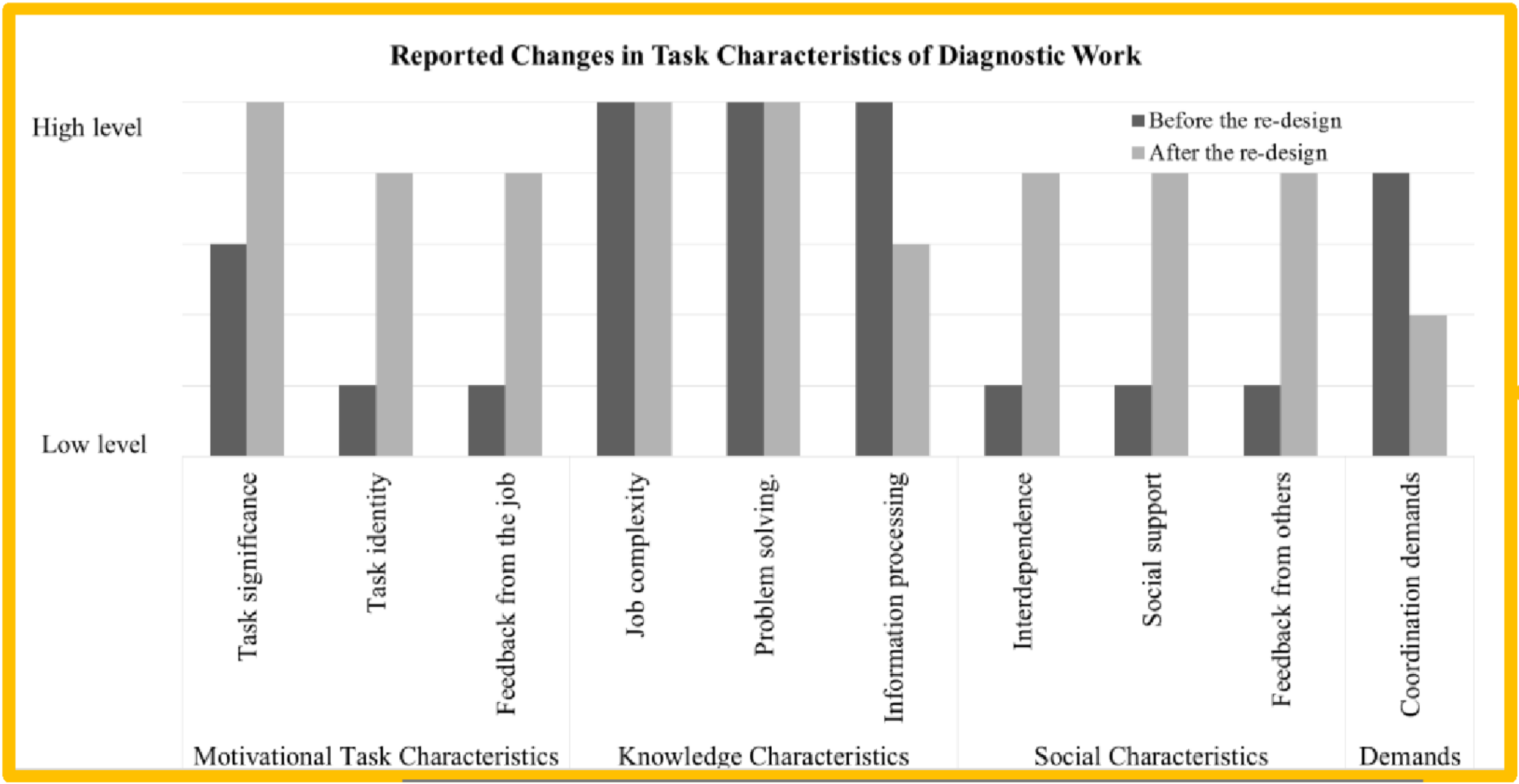
Sketch by Lynne Chapman

Transformative Work Design

Undiagnosed Diseases Program

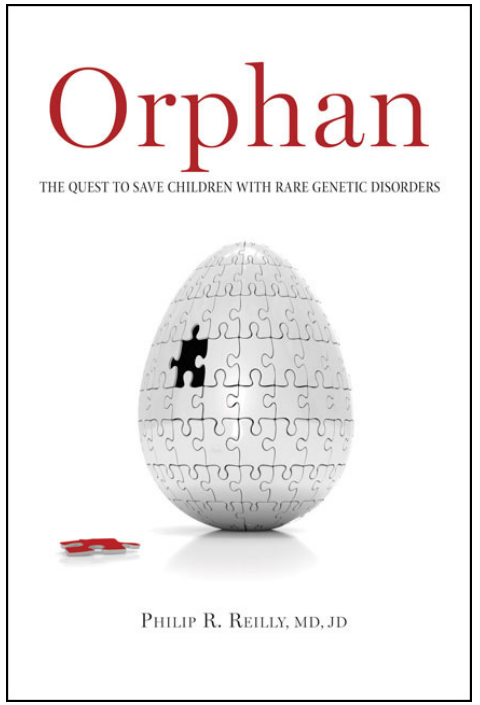


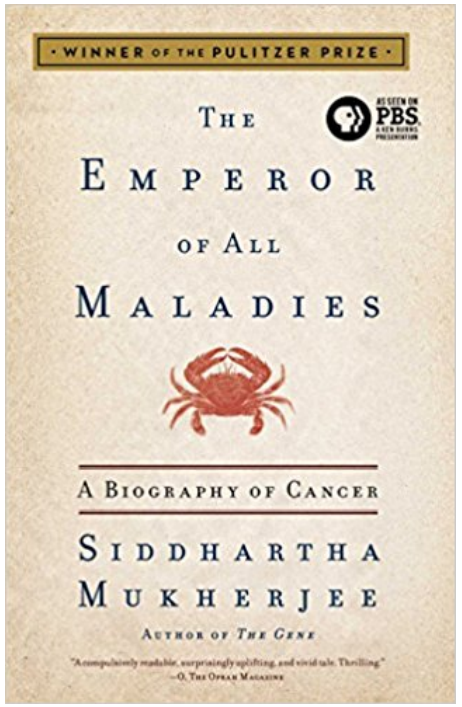
Courtesy of the Centre for Transformative Work Design, Sharon Parker, Florian Klonek, Georgia Hay. Sketch by Lynne Chapman


Reported Changes in Task Characteristics of Diagnostic Work
High level
Low level
Before the re-design
After the re-design
Task significance
Task identity
Feedback from the job
Job complexity
Problem solving.
Information processing
Interdependence
Social support
Feedback from others
Coordination demands
Motivational Task Characteristics
Knowledge Characteristics
Social Characteristics
Demands

National Health and Medical Research Council

- “Preparing Australia for the Genomics Revolution in Health Care”
- $ 25 Million
- commenced 2016
- Includes rare diseases and cancer flagships
  - RD flagships
    - Organ based
    - Acute care in newborns
    - Intellectual disability
    - Stillbirth/ neonatal death
  - Australian Functional Genomics Network





Australian Genomics Health Alliance

Medical Research Future Fund (MRFF)

- $20 billion investment in the health of Australians, our economy and the sustainability of our health system.
- $1.4 billion spend in the first 5 years
- Includes $500 Million Genomics Health Futures Mission, est 2018

Genomics Health Futures Mission

- involves:

1. **new and expanded clinical flagship studies to tackle rare diseases, rare cancers** and complex conditions;
2. **new clinical trials** and technology applications;
3. increased academic and researcher collaboration and new career pathways;
4. co-investment with philanthropy and business to support new industries;
5. community dialogue to better understand the value of genomics and gain appreciation of the privacy, legal, social and familial impact; and
6. analytical power backed by national standards and protocols that ensure secure data holdings, access, analysis and sharing to benefit Australians.

GHFM: Rare Diseases Specific Calls

- 2018
  - ‘Mackenzie’s Mission’ $20 million trial in **pre-conception screening** for rare and debilitating genetic birth disorders
  - **Rare Diseases and Rare Cancers Program**. $38.6 million supporting 23 new clinical trials to improve treatments and discover cures for debilitating and deadly rare cancers and rare diseases.
  - **Rare Cancers, Rare Diseases and Unmet Need Clinical Trials Program.** Further $33 million

Ministerial Council for **Precision Health**, WA

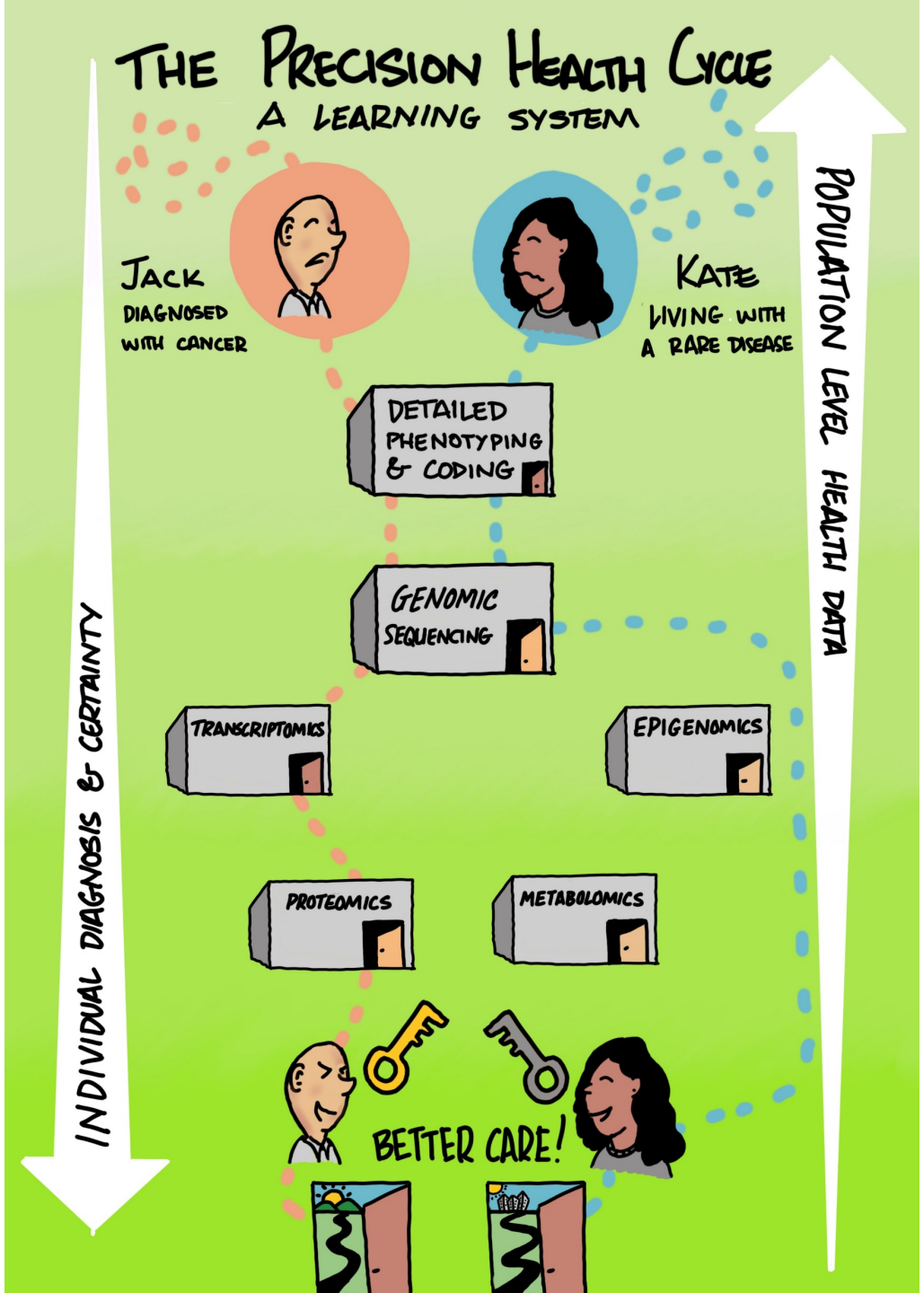
Hon Roger Cook BA GradDipBus MBA MLA

Deputy Premier; Minister for Health; Mental Health

- Est April 2019
- **Precision Health** = Precision Medicine + Plus Precision Public Health
- **Precision Public Health** = combining old and new approaches to do the most for the most, and the most for those at highest need
  - New = (gen)omics, spatial technologies, big data and AI etc
- representatives from medicine, science, industry, Aboriginal health, patient organisations, medical research and commercialisation, as well as experts from key precision health-related technologies of genomics, phenomics, informatics and geographical information systems.

"Western Australia already benefits from emerging precision health initiatives such as the Undiagnosed Diseases Program of WA (which provides earlier diagnosis for people with rare and baffling medical conditions), and the

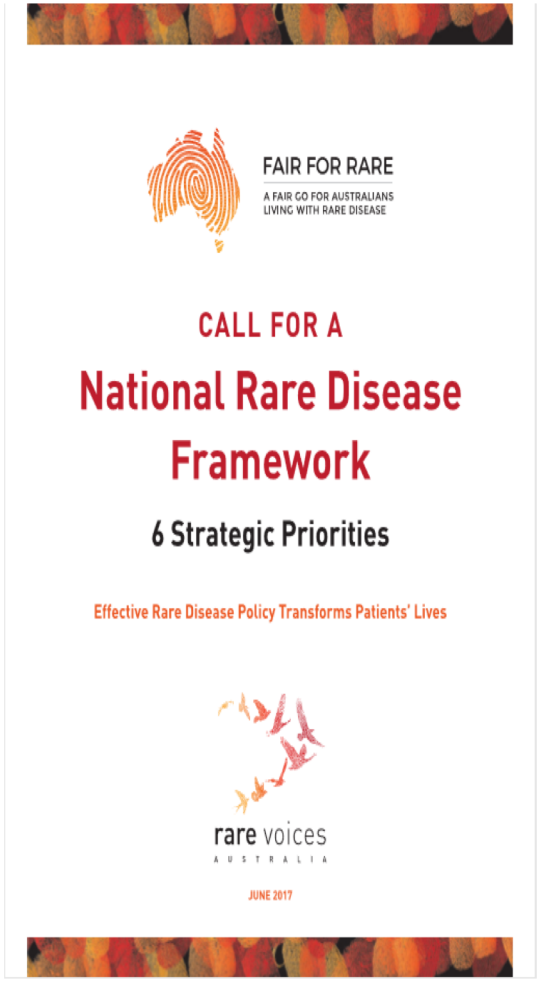
SPATIAL
EPIGENOMICS
CLINICAL PHENOTYPE
OLD KNOWLEDGE
DIFFERENT LANGUAGES
SIMILAR CONVERSATIONS
NEW KNOWLEDGE
CLINICAL GENOTYPE
EXPOSOMICS
PHENOMICS



Call for a National Rare Disease Framework, 2018

Strategic Priorities –

1. Diagnosis
2. Access to Treatments
3. Data Collection
4. Coordinated Care
5. Access to Services
6. Coordinated Research



rare voices
AUSTRALIA

NATIONAL STRATEGIC ACTION PLAN FOR RARE DISEASES

The Hon. Greg Hunt MP - Minister for Health

MEDIA RELEASE - 16 November, 2018

<u>Australia's first National Rare Diseases Framework</u>

… Government is taking more action to support people battling rare conditions by developing the ***Australia's first National Rare Diseases Framework and Action Plan****. Funding … will be provided to* ***Rare Voices Australia*** *through the Morrison Government's* ***Public Health and Chronic Disease Program*** *to enable the* ***collaborative development*** *of the action plan and framework …*

…recognises the fact that these diseases are statistically rare … ***therefore special and concerted efforts are needed to address them****… rare diseases which are life-threatening or chronically debilitating.*

Slides Courtesy Nicole Millis

rare voices AUSTRALIA

ACTION PLAN CONSULTATION PROCESS

- **Stakeholder Consultation**
  - National Rare Disease Summit (Nov 2018 - Melb)
  - Round Table Consultation (26 Feb 2019 – Syd)
  - RD Patient Org Consultation (26 Feb 2019 – Syd)
  - Targeted Stakeholder Consultation – Access to Services (12 Mar 2019 – Melb) Stakeholder Consultation: (25 March 2019- Perth)
  - Targeted Consultation: Industry – 8 April 2019 Stakeholder Consultation: (29 April 2019 – Brisbane)
  - Online Survey/s (April/May 2019)

- **Writing & Steering Committee Feedback Draft Report (May)**

- **Draft Report DoH – mid June 2019**

rare voices AUSTRALIA

SOME EMERGING PRIORITIES…

- Coding – fundamental
- Specialised Diagnostic response (access/inequity)
- Improving pathways/ rare disease journey
- Centres of Excellence (what does ‘excellence’ look like in RD?) What sort of model?
- Integrated Care: interdisciplinary, smooths patient journeys, optimal, continuous and effective whole of life care
- Clinical Care /Research relationship
  (what is special about RD research?)

rare voices AUSTRALIA

Thanks !


RARE DISEASE
DIAGNOSTIC ODYSSEY
UNCERTAINTY
CANCER
LATE SCREENING
ABORIGINAL
BARRIERS TO SCREENING
OBESITY
BROAD MESSAGING
INFECTIOUS DISEASES
LIMITED MONITORING
SILOED
REACTIVE DISEASECARE
PROACTIVE HEALTHCARE
HEALTH SYSTEM CHANGE
A PERSONAL, PRECISION HEALTH SYSTEM
PERSONAL LIFESTYLE CHANGE
FAMILY
GENOMIC TEST
DIAGNOSIS
TARGET CARE
LIQUID BIOPSY
EARLY DETECT
PRECISE CARE
COMMUNITY
REMOTE SCREEN
REFERENCE DATA & CLEAR PATH
PRECISE SCREEN
TAILORED INTERVENTION
RAPID IDENTIFICATION
CULTURE
TIGHT CONTROL
-ONE-
SIZE FITS ALL
BLUNT instruments
TOLD YOU SHOULDN'T
PERSONALISED
PRECISE
screening
intervention
treatment
LEARN WHAT'S RELEVANT TO YOU
ENABLES
COLLABORATION

WHAT WE ARE LEARNING…

Strong sense of collaboration and generosity in the sector

Action Plan – seen as real opportunity for change

rare voices AUSTRALIA

Australian National Congenital Anomaly Register

- Committee est 2019 under the Australian Institute of Health and Welfare
- Rare Diseases **Coding** – an item of likely importance
  - Congenital Anomalies are a large class of mainly rare diseases
- UK approach – **National Congenital Anomaly and Rare Diseases Registration Service**
  - combined ICD-10 coding and Orphanet coding
  - Build on a foundation of congenital anomaly register infrastructure and supplemented through learnings from and approaches to rare diseases and cancer
- New **WHO Advisory Group on Birth Defects** (aka congenital anomalies) est 2018
  - Note existing approaches/ infrastructure for congenital anomalies in many jurisidictions

NATIONAL STRATEGIC ACTION PLAN FOR RARE DISEASES

The Hon. Greg Hunt MP - Minister for Health

MEDIA RELEASE - 16 November, 2018

Australia's first National Rare Diseases Framework

Specific priorities, actions and activities *will be identified through* ***extensive consultation*** *including people with a rare disease, clinical and academic experts, policy makers and state and territory governments.*

The framework and all recommendations for action will be supported by ***robust evidence*** *so that people with rare diseases can receive timely diagnosis, treatment and the best care possible.*

This plan will benefit people with rare diseases, their families and the wider community ….

rare voices AUSTRALIA