



Evaluating Care for DMD: The Australian perspective CARE-NMD

Budapest , 18-19 April 2013

Professor Hugh Dawkins

Director

Office of Population Health Genomics

Public Health & Clinical Services Division

Department of Health, Western Australia

A/Professor Monique Ryan

Head, Neuromuscular Clinical and Neuromuscular Research Unit

Royal Children's Hospital , Melbourne

Victoria, Australia

A/Professor Joshua Burns

Research Fellow

National Health and Medical Research Council

Children's Hospital at Westmead

University of Sydney, New South Wales, Australia

Lesley Murphy

Director

Rare Voices Australia

National alliance representing the voice of all Australians living
with a rare disease

PERSPECTIVES

- Vignettes from Australia [models]
- The need for implementation in each jurisdiction [national consistency]
- Compelling reason for translation [policy] and national consistency [Plan]



DEMOGRAPHICS

AREA

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

POPULATION

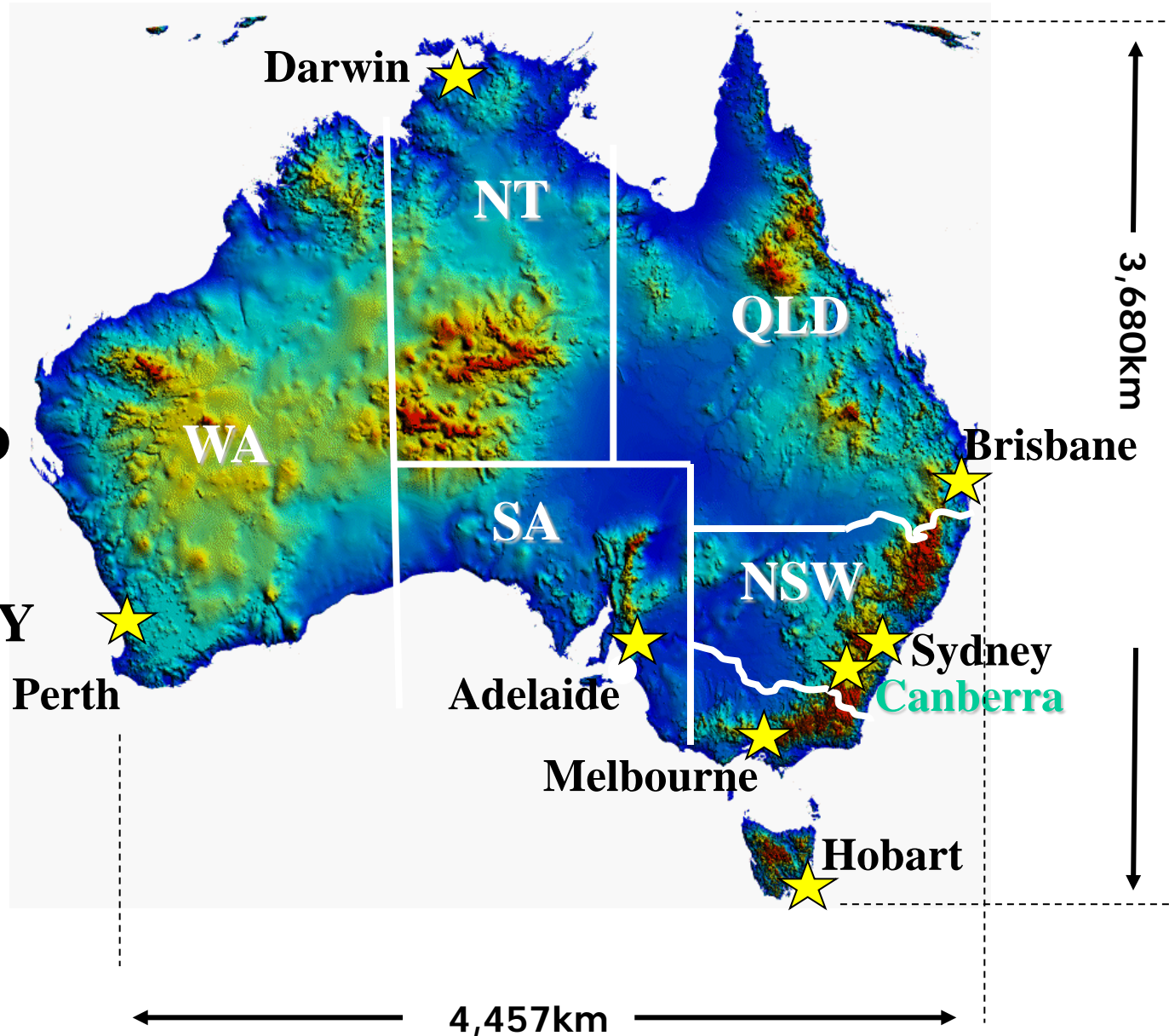
Australia: 22,000,000
Western Australia: 2,200,000

POPULATION SPREAD

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

POPULATION DENSITY

Northern Territory: 0.15 people / km²
Western Australia: 0.75 people/ km²
New South Wales: 8 people/ km²
Victoria: 22 people/ km²



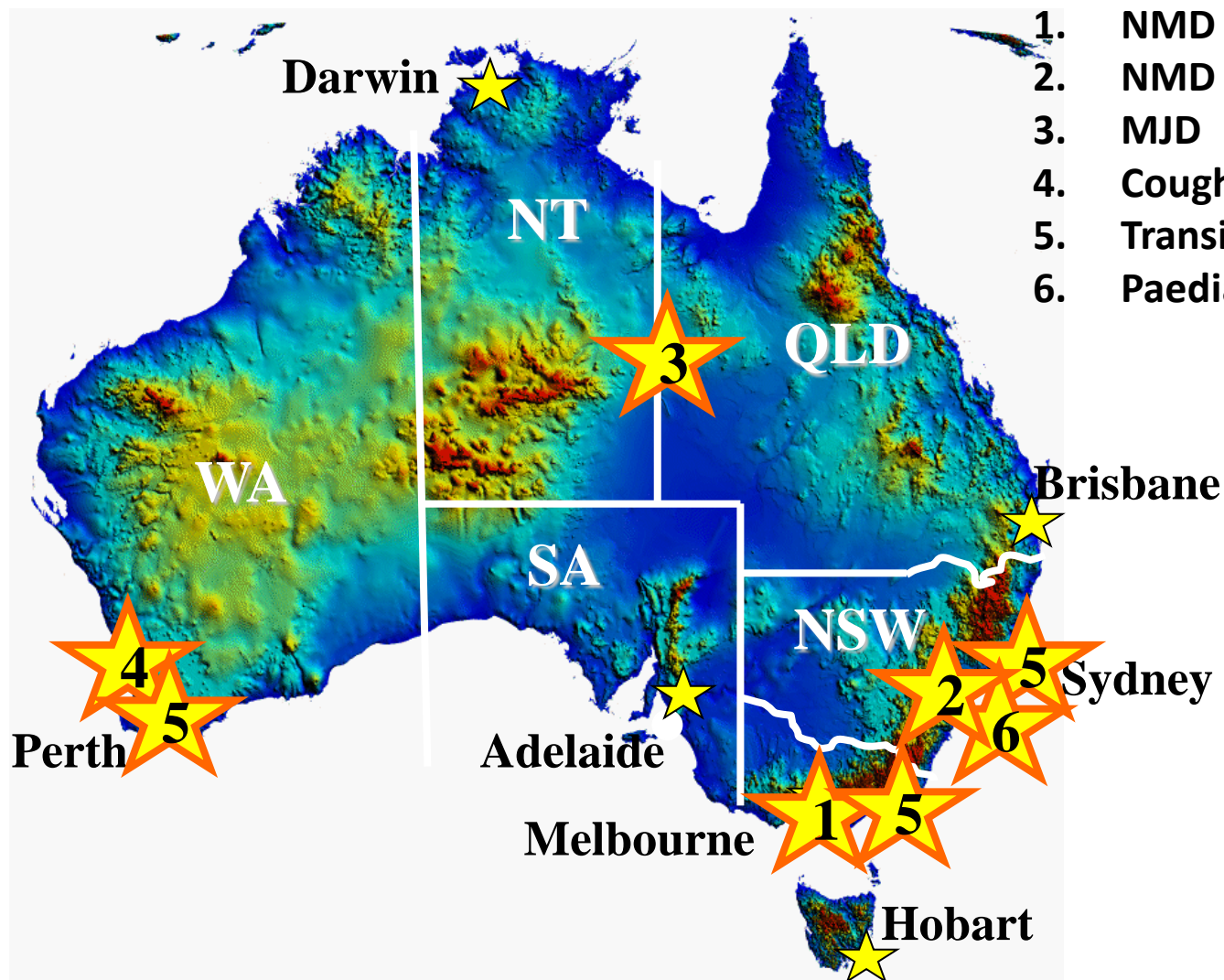
MODELS

VIGNETTES FROM AUSTRALIA



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

SNAPSHOT OF RD CARE PROGRAMS



1. NMD Clinic
2. NMD Centre of Research Excellence
3. MJD
4. CoughAssist Evaluation
5. Transition engagement
6. Paediatric CMT Clinic

Australian Neuromuscular Network [National Network]

- AIMS
 - to ensure excellence in diagnostic methods and clinical management, and equal access to clinical trials and new therapies, for all individuals in Australia and New Zealand affected by neuromuscular disorders.
 - proposes to achieve our vision by establishing a cohesive, integrated neuromuscular network which enables people to work together across Australia and New Zealand, for the well-being of patients.
- STEERING COMMITTEES TO HELP ACHIEVE AIMS
 1. Patient Diagnostic Network
 - To coordinate a national collaborative diagnostic service and research network for neuromuscular disorders that is cost-effective, maximises availability and minimises duplication of services. This will include introduction of new diagnostic methods into Australia and New Zealand, as well as introduction of screening for newly identified genetic disorders.
 2. Patient Registries
 - To develop nationwide disease registers, based on accurate molecular diagnosis for patients with neuromuscular disorders, aligned with international registries such as the TREAT-NMD registries.
 3. Clinical Trials and Clinical Network
 - To establish a clinical trials framework for neuromuscular disorders accessible to patients around Australia and New Zealand.
- ENABLING : training and face to face meetings to inform and help consolidate effort and align efforts and more importantly develop national best practice.
- The clinic structures in Melbourne and Sydney differ slightly but all work towards the same best practice guidelines. Some of the stumbling blocks are distance and community support. this is

1 . Paediatric NMD Clinics

- Two states [NSW & VIC] have multidisciplinary clinics for patients with NMD's
- These centres are linked to international and national NMD initiatives including:
 - Australian Neuromuscular Disease Network [ANN] practices for management of rare diseases part of RARE Bestpractice [Global CI-A: Domenica Taruscio; Australia: Kathryn North & Hugh Dawkins]
 - NHMRC National Centre for Research Excellence in Neuromuscular Disorders: Transforming the management of neuromuscular disorders from compassionate assistance to targeted therapy and prevention [CI-A Kathryn North]
 - One primary aim of the CRE is to assist with setting up multidisciplinary clinics in the other states.
- TREAT-NMD and CARE-NMD
 - Leading the CARE-NMD survey in Australia

New South Wales

- Children's Hospital at Westmead :
 - Paediatric NMD weekly neurogenetics clinic and a monthly neuromuscular management clinic.
 - a transition clinic with Royal North Shore Hospital, whereby the team from RNSH attend a clinic at the Children's Hospital at Westmead to meet the patients and explain the process of transition from paediatric care to adult care.
 - facilitates rapport and trust building which have been identified as challenges to successful transition to adult care. The ability for the medical team and allied health teams to liaise and handover aims to facilitate a smooth transition.
 - a monthly peripheral neuropathy clinic, which is the only one in Australia to provide this level of care for patients with peripheral neuropathy (primarily Charcot-Marie-Tooth disease).
- Sydney Children's Hospital :
 - weekly multidisciplinary clinic for patients with a muscle disease (NMD), as well as outreach clinics where they visit Wagga Wagga and Coffs Harbour twice a year to conduct clinics for rural families.
- The multidisciplinary clinics work to incorporate research and best practice in the multidisciplinary framework. This includes patients accessing a range of health professionals, including medical, allied health and other ancillary services as required; in keeping with current best practice guidelines.

THE ROYAL CHILDREN'S HOSPITAL

Holistic & multidisciplinary care,
management and research for children
with Neuromuscular Disorders at

Daniella Villano

MDA Neuromuscular Nurse Coordinator

Advanced Practice Nurse

MDA Neuromuscular Clinic



- Commenced in February 2008
- Multidisciplinary clinic which brings together relevant medical specialties, allied health therapists and other health professionals.
- “*One-stop-shop*” specifically designed for children with neuromuscular disorders and their families.
- Platform for neuromuscular research

MDA Neuromuscular Clinic Team

- Neurology
- Respiratory
- Cardiology
- Orthopaedics
- Genetic Counseling
- Physiotherapy
- Occupational Therapy
- Orthotics
- Social Work
- Nurse Coordinator
- Mental Health
- Teacher Consultant
- MDA Staff Support
- Diagnostic testing as required
- Transition program



Why come to clinic?

- Each child's overall physical function, pulmonary function, cardiac function, muscle strength and bone health are monitored
- A regular review of medical management
- Any side effects of treatment are assessed and monitored
- To receive up-to-date information and support
- Ongoing documentation of each child's condition & progress, that can be communicated with others involved in the child's care
- Planning for current and future issues e.g. surgery, equipment, preparation for schooling, transition to adult services

WHAT DO WE DO?

Equipment

Education

Improved Services

Research

Advocacy

Improving the lives of Indigenous Australians with Machado Joseph Disease in remote and rural areas



MACHADO JOSEPH DISEASE HOW IT AFFECTS MY BODY

Loss of balance

Sad/depressed

Slurred speech

Sleep disturbance

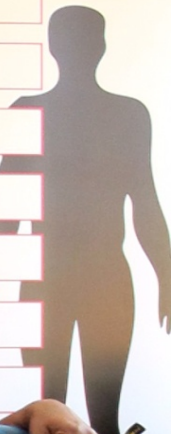
Blurry/shaky vision

Swallowing difficulties

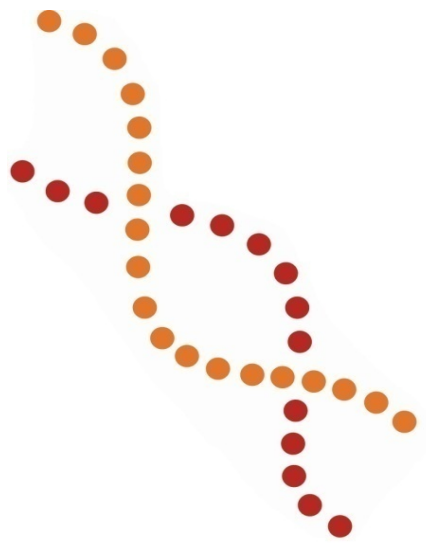
Bowel/bladder problems

Muscle cramps

Falling over



Machado Joseph Disease and the MJD Foundation



Machado Joseph Disease

Spinocerebellar Ataxia 3



Genetic:

- Autosomal dominant (50%)
- Not sex linked
- Anticipation Effect

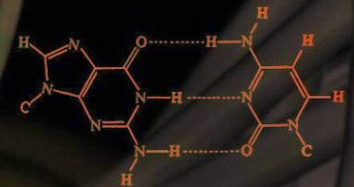
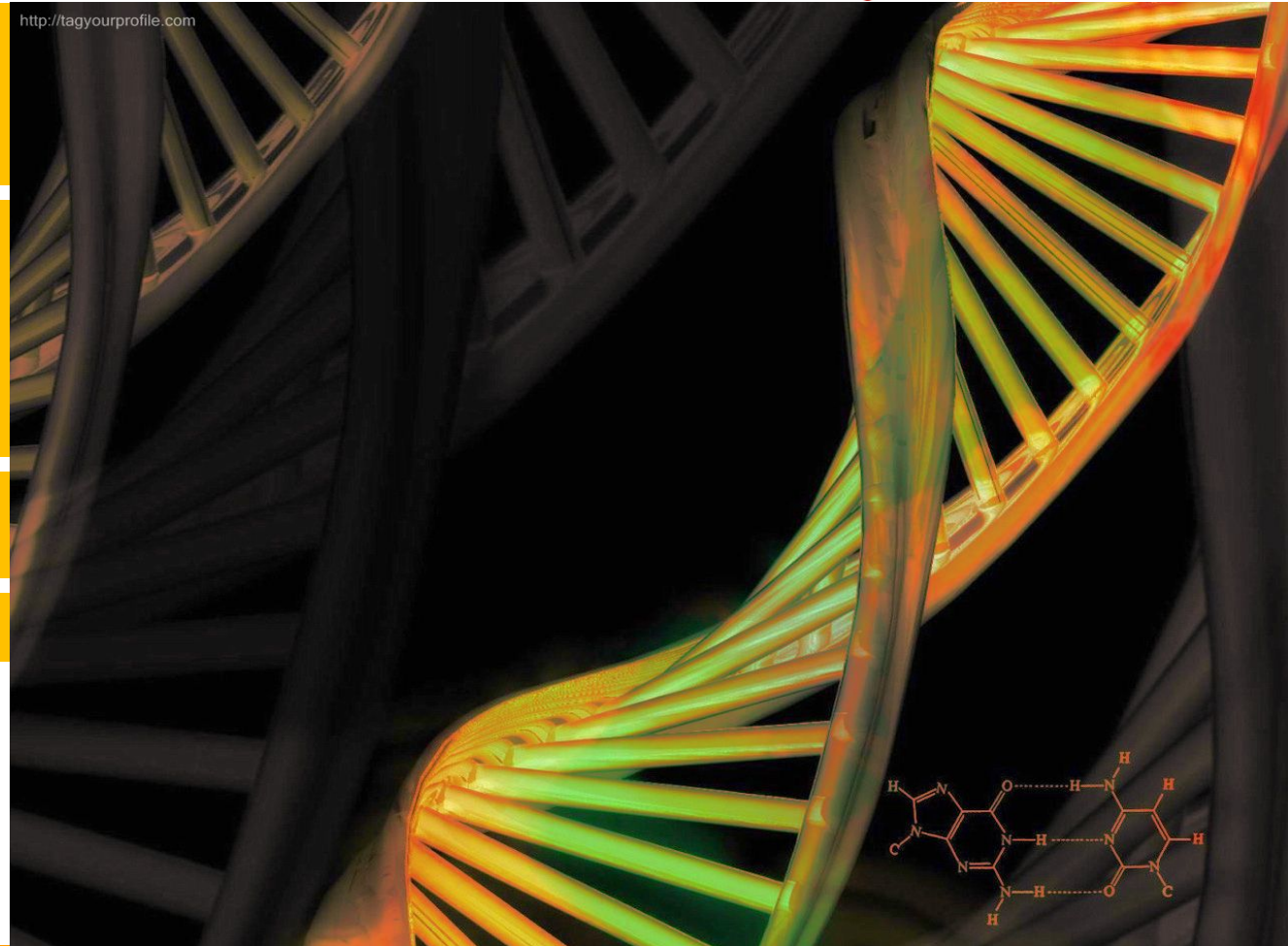
Neurodegenerative:

- Premature death of nerves in the cerebellum
- Causes abnormal protein buildup
- Slows motor pathways
- No loss of intellectual capacity

Dependence 5-10 years
Generationally determined

Incurable

<http://tagyourprofile.com>



Improving Quality of Life for Indigenous Australians living with MJD

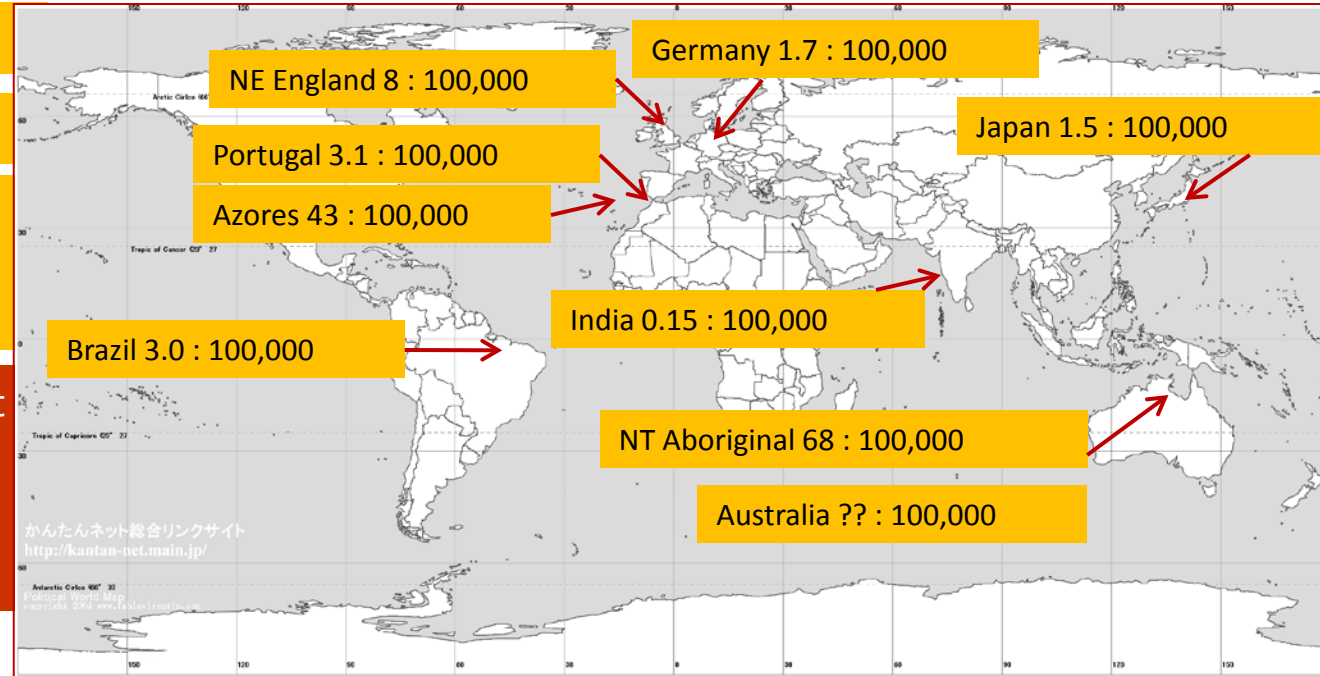
MJD Prevalence

Most common SCA

Worldwide ~0.6 : 100,000

NT Indigenous Australians have the highest prevalence in the world ~68: 100,000

Even higher in estimates for east Arnhem land communities
303-909 : 100,000
(MacMillan, 2011, prediction figures based on a validated Huntington's Disease model)



Improving Quality of Life for Indigenous Australians living with MJD

MJD Foundation



Mission: The MJD Foundation seeks to improve the quality of life of Indigenous Australians and their families living with Machado Joseph Disease in Arnhem Land and beyond.

Charity

- Established in May 2008
- Public company limited by guarantee under the Australia Corporations Act

Board of Governance

- local leaders, field experts

Staffing

- Administrative, therapy, community workers – Indigenous and non Indigenous (some with MJD)

Funding

- Investments, Corporate and Individual donations. Project Grants

Integral Relationships

- Partners and Sponsors



Improving Quality of Life for Indigenous Australians living with MJD

MJD FOUNDATION WHAT DO WE DO?

Engagement

Equipment



Work at a grass roots level to build relationships, and understand what families/communities need/want.

Education



MJD Foundation organise programs and projects under core activities:

Improved Services

- EQUIPMENT
- EDUCATION
- IMPROVED SERVICES
- RESEARCH
- ADVOCACY

Research

Advocacy

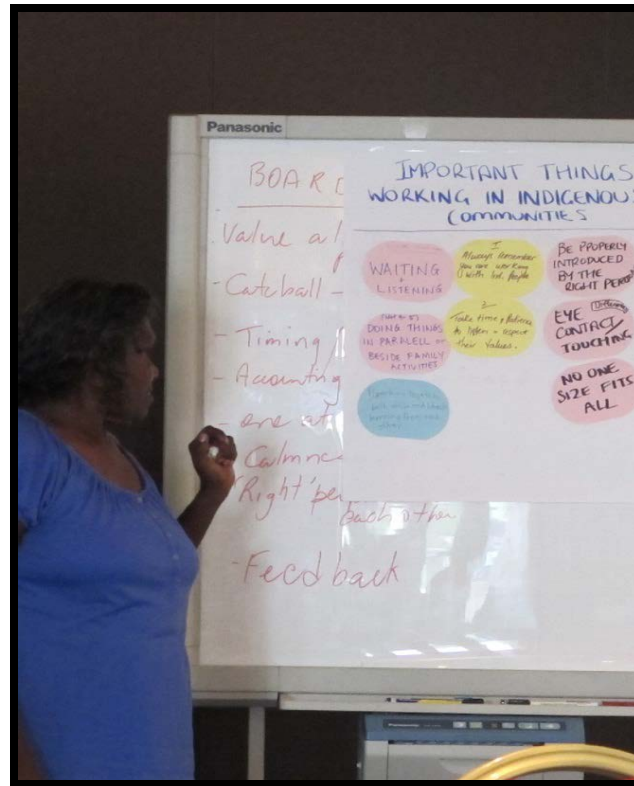


Collaborate with other service providers, and meet gaps that are not funded.

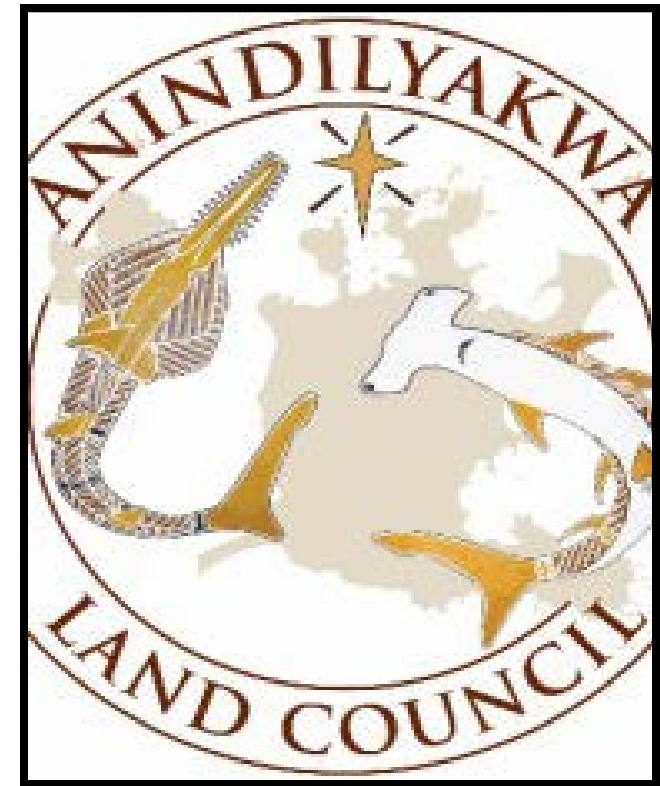
How do we operate?



Meeting needs where they happen -can be a little challenging...



Advice from the experts

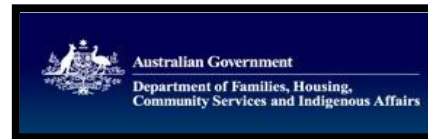
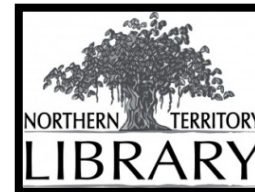


Cultural integrity and community engagement is vital.

How are we funded?



A combination of managed investment from a large Aboriginals Benefit Account grant, philanthropic and project grants, corporate sponsorship and partnership arrangements.



Paediatric Transition and Engagement

- National Clinical Network
 - Federal Department of Health and Aging initiative
- Australian Paediatric Surveillance Unit [APSU, Sydney]
- Office of Population Health Genomics [WA Health]
- Department of Health Services [Victoria]

- APSU and OPHG engagement with young people living with RD involved in transition to better inform transition planning
- OPHG engaging with clinical and allied health specialists to inform transition planning of complex disorders from a service delivery perspective



Cough-Assist Service Evaluation

- Muscular Dystrophy WA initiative: >47 Cough Assist [insufflation-exsufflation] machines to children and adults with NMD and service and care under a NMD program
- WA Health perspective; collect evidence of the Cough-Assist program to determine if this should be expanded to a state wide service
- Intervention [Case] matched to Control study using a controlled before and after design [Cochrane effective practice and organisation of care 2007]
- Survey of participants
- Service evaluation and cost benefit analysis using data-linkage to hospital health data collections
- Data from 12 months prior to intervention for 12 months of intervention
- Cohort of 36 cases with matched controls
 - 20 [55%] with Duchenne, 3[8%] with SMA, 2 [5%] and single cases of other MD diagnosed disorders
- Analysis of data and health measures from WA health data collections



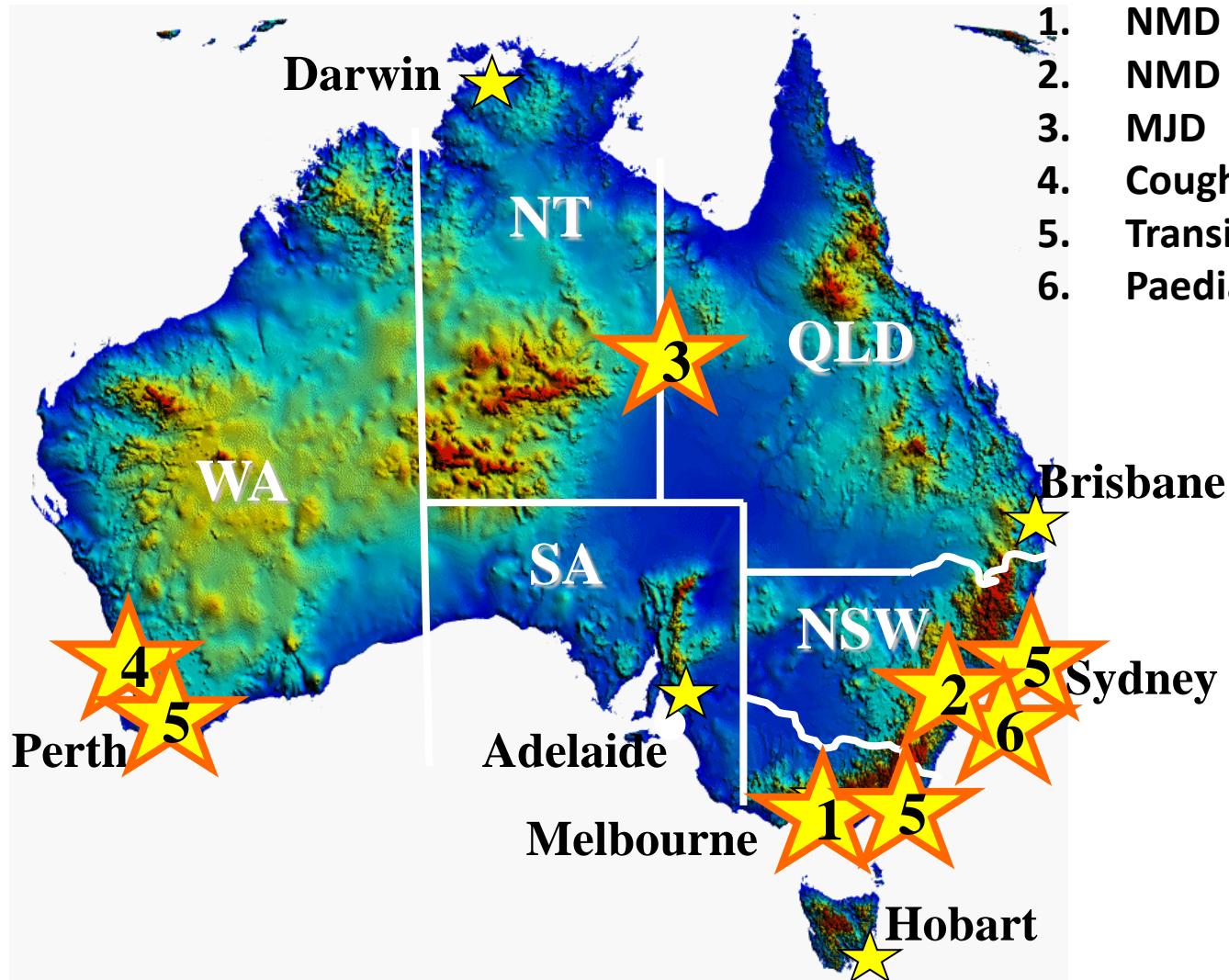
NATIONAL CONSISTENCY

THE COMPELLING CASE FOR A NATIONAL PLAN



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

What's wrong with this picture



1. NMD Clinic
2. NMD Centre of Research Excellence
3. MJD
4. CoughAssist Evaluation
5. Transition engagement
6. Paediatric CMT Clinic



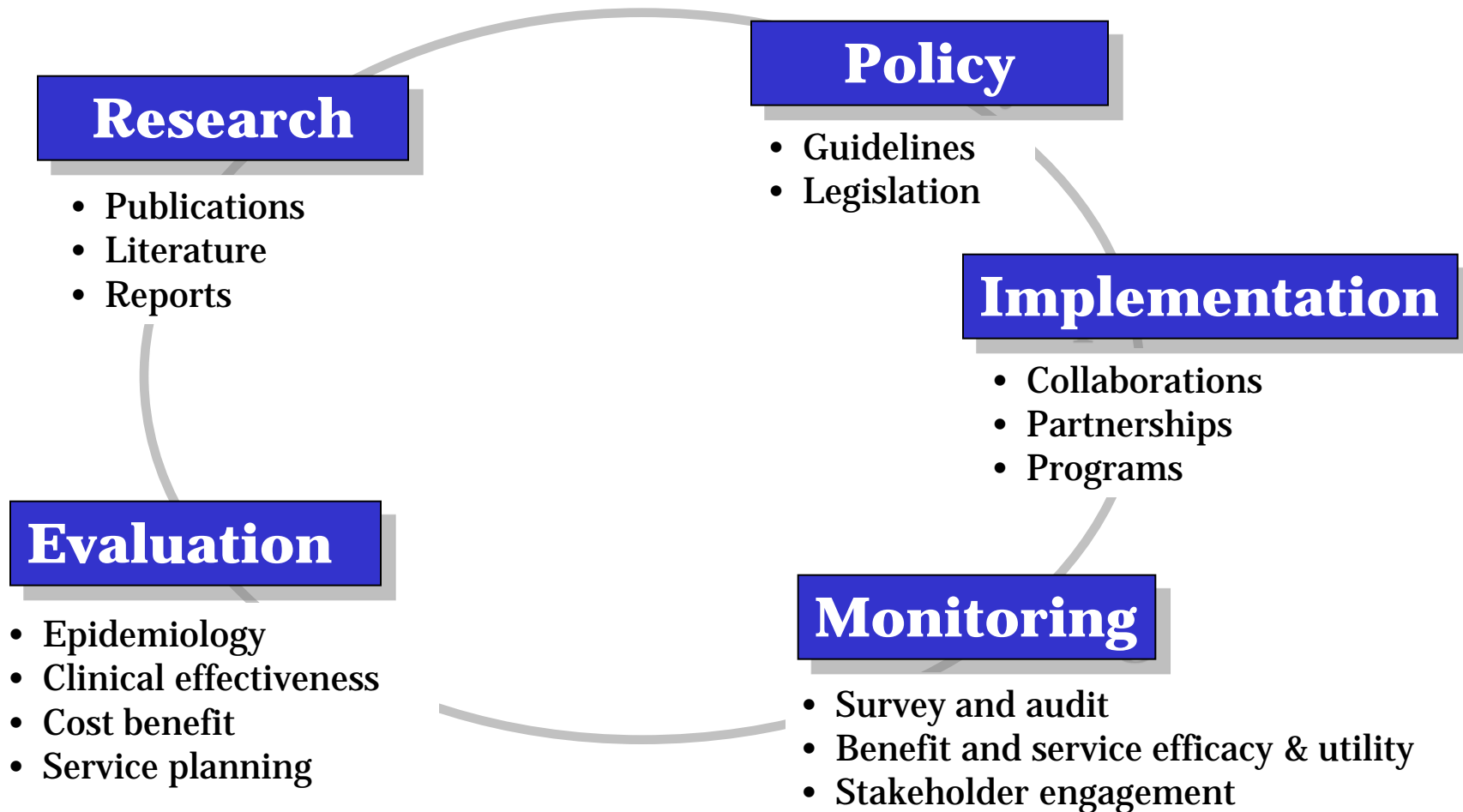
POLICY

THE NEED FOR KNOWLEDGE TRANSLATION

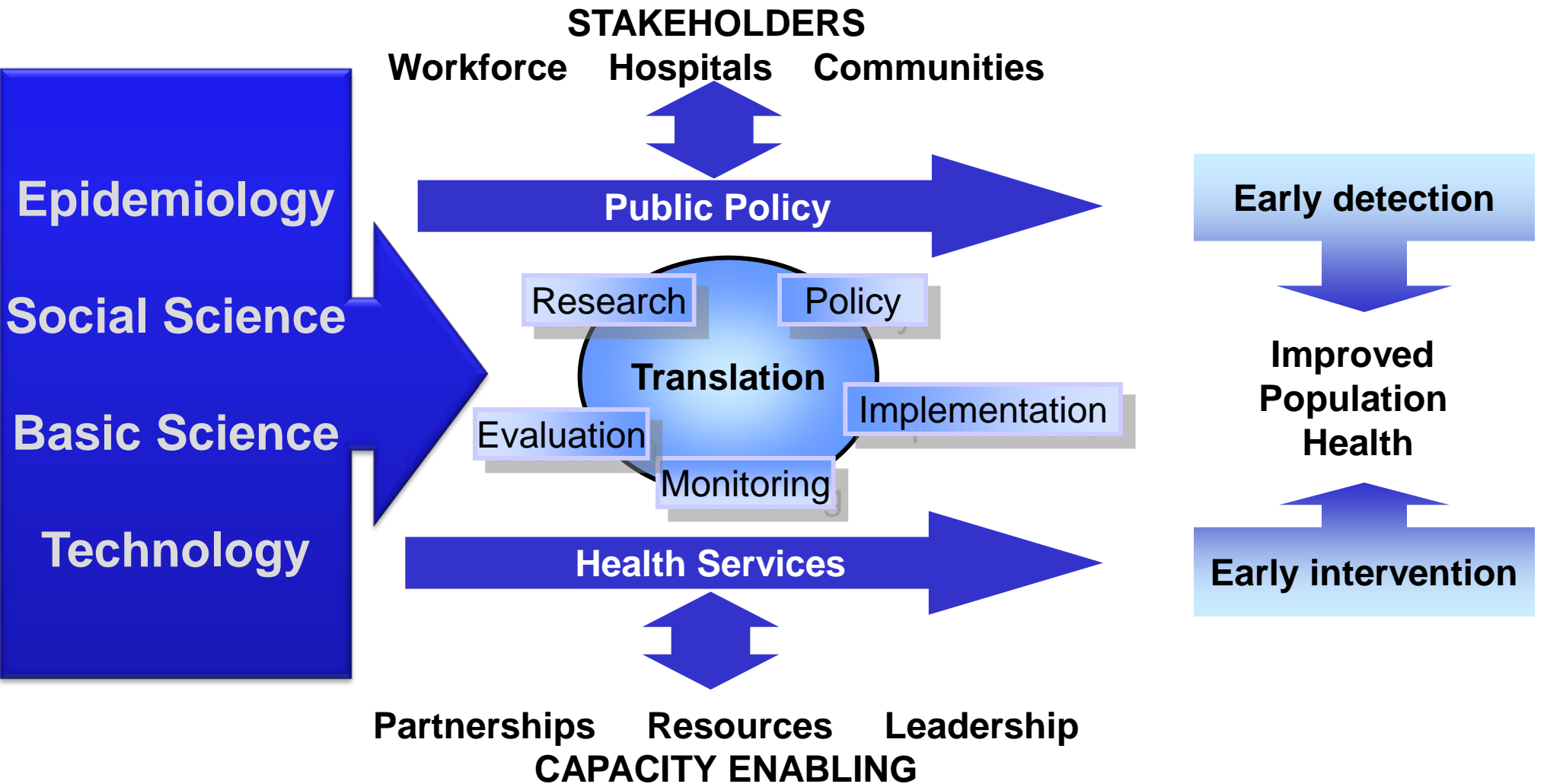


Government of **Western Australia**
Department of **Health**
Public Health Office of Population Health Genomics

The virtuous cycle of public health



TRANSLATION OF KNOWLEDGE INTO HEALTH BENEFIT









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



OFFICE OF POPULATION HEALTH GENOMICS

Established 2001

PURPOSE

- To lead the translation of genomics and new knowledge into health benefits

AIMS

- To maximise the health of the population through implementation of genetic and -omic technologies;
- To translate the benefits of genomics into improved community knowledge and health; and
- To ensure that strategies are consistent with community needs and ethical, legal and cost-effective.



WA HEALTH

- Delivering health services to nearly 2.3 million Western Australians

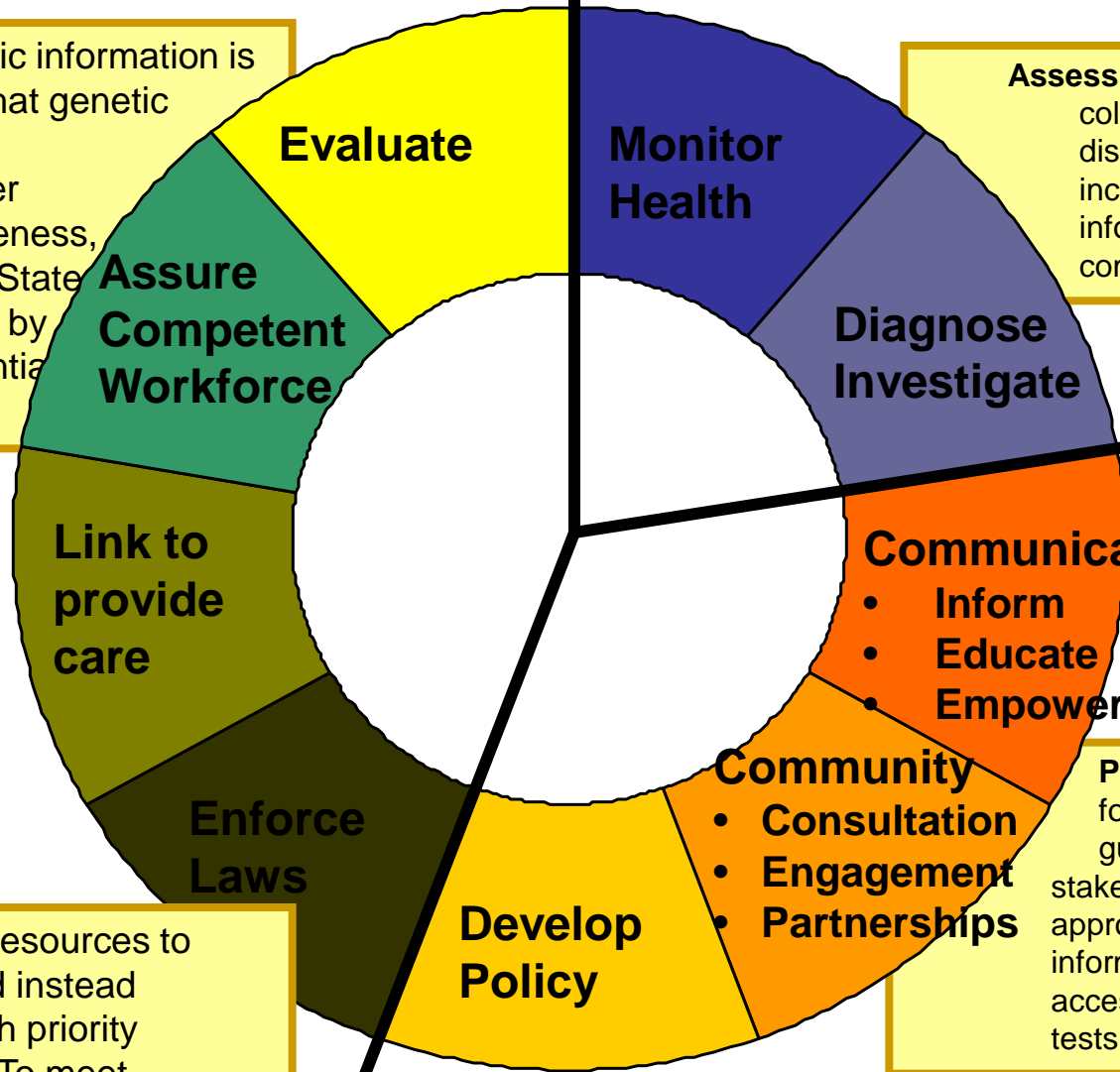
Strategic Intent 2010-2015:

- Caring for individuals and the community;
- Caring for those who need it most;
- Making best use of funds and resources; and
- Supporting our team.



Core Public Health Genomics Functions

Assurance: That genomic information is used appropriately and that genetic tests and services meet agreed goals (stakeholder engagement) for effectiveness, accessibility and quality. State fulfil these core functions by conducting various essential public health services



Assessment: The regular systematic collection, assembly, analysis and dissemination of information, including genetic epidemiologic information on the health of the community

- Communication**
- Inform
 - Educate
 - Empower

Policy development: The formulation of standards and guidelines in collaboration with stakeholders that promote the appropriate use of genomic information and the effectiveness, accessibility and quality of genetic tests and services

OPHG: do not have the resources to perform all 9 aspects and instead strategically focus on high priority functions for WA Health. To meet objectives OPHG rely heavily on networks and partnerships with other jurisdictions, academic cs, helath care

OPHG strategy & policy development

RARE DISEASES



Rare Disease

- frequently life-threatening or chronic debilitating diseases
- a low prevalence in the community.
- internationally accepted classification five or less per 10,000 head of population.
- Currently over 7,000 rare diseases have been identified, about 80% of which have a genetic origin.
- Taken cumulatively, the number of people affected by rare diseases has been estimated at 1-in-12 people, or about 8% of the population.
- Australian data, and interpolation from international prevalence figures, suggest that approximately 1.8 million Australian people are affected by rare diseases; including more than 400,000 children.
- The burden of disease is not confined to those affected; it also impacts the emotional and financial well-being of the family and the wider community as a result of loss of productivity and through the provision of disability services and health services.
- In Australia, and internationally, public advocacy and engagement are helping to form strong community endorsed policy and a mandate for change in health services.
- Rare disease advocacy is also driving change in industry and technology development agendas for treatment and management of rare diseases.
- Technology *per se* is insufficient to bring about the change and policy development required by the rare diseases community.
- Genomic, biotechnology and pharmaceutical developments represents only a component of the expressed and unmet needs of the rare diseases community.




Social & Community Research

Caron Molster, Gaenor Kyne, Fiona Hope

“(WA Health) commit to fostering and supporting community & consumer engagement in health system performance”

INCREASING LEVEL OF PUBLIC EMPOWERMENT

CONSULT	INVOLVE	COLLABORATE	EMPOWER
<p>Community Surveys (1 way) Identifying data Mandatory fortification of folate Genetics knowledge Information seeking behaviours Genetic technologies (e.g. tests) Family health history Cascade screening for FH Evaluation of resources</p>	<p>Deliberative Forums (2 way)</p> <p style="text-align: center;">Biobanking in WA</p> 		



Government of **Western Australia**

Department of **Health**

Public Health Office of Population Health Genomics