National genetic registries: The Australian experience

CARE-NMD
Budapest
18-19 April 2013

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PERSPECTIVES

- Registry background [context]
- Update on NMD registries [status]
- Update on rare disease registry development [ontology]

CONTEXT

REGISTRY BACKGROUND

BACKGROUND

- Duchenne Foundation initiated a nationwide campaign for a National DMD Registry to collate clinical and genetic data
  - Received strong collaborative support from Muscular Dystrophy Associations and families living with DMD across Australia
- Clinical Technical and Ethical Principal Committee (CTEPC)
  - Principal committee of the Australian Health Ministers Advisory Council (AHMAC) requested a report with recommendations for a National Registry
- Office of Population Health Genomics (WA Health)
  - Established and led a working group
  - Engaged and collaborated with jurisdictions, stakeholders & TREAT-NMD
  - Recommendations endorsed by all stakeholders
  - Report and Recommendations submitted

STAKEHOLDER POSITION

All stakeholders supported establishing a National Registry for DMD, and endorsed the Purpose Statement and Objectives

Purpose Statement

- To provide Australian Duchenne muscular dystrophy families with an opportunity to improve the outcomes of individuals affected by DMD.

Objectives

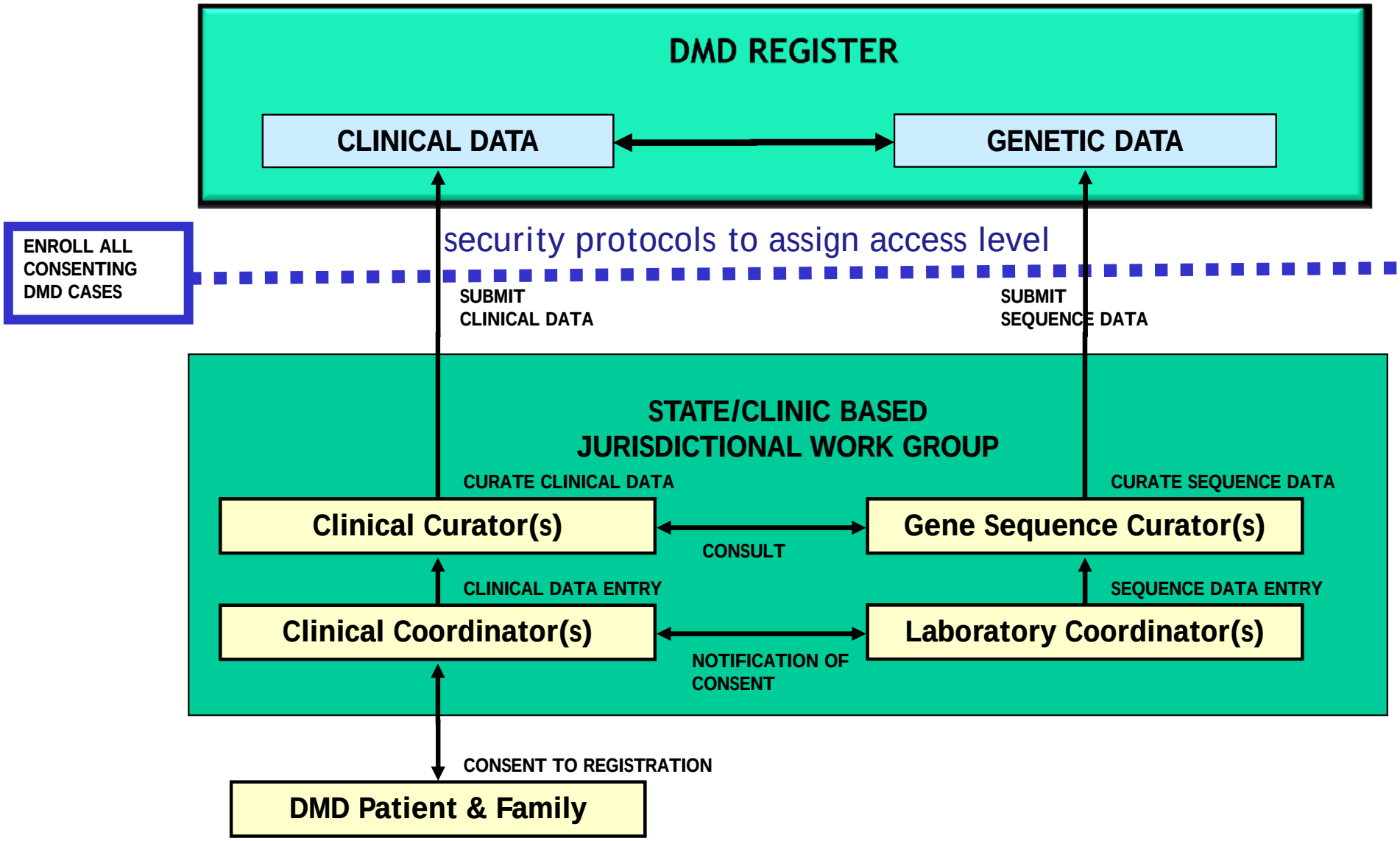
- To improve care of Duchenne muscular dystrophy patients through the coordination of diagnosis and therapy and by ensuring new intervention strategies are available on an equitable and consistent manner across Australia; and
- To improve opportunities for international collaboration by facilitating and accelerating recruitment process of Australian DMD patients into new clinical trials and for participating in studies for the benefit of the world DMD community and the advancement of medical science.

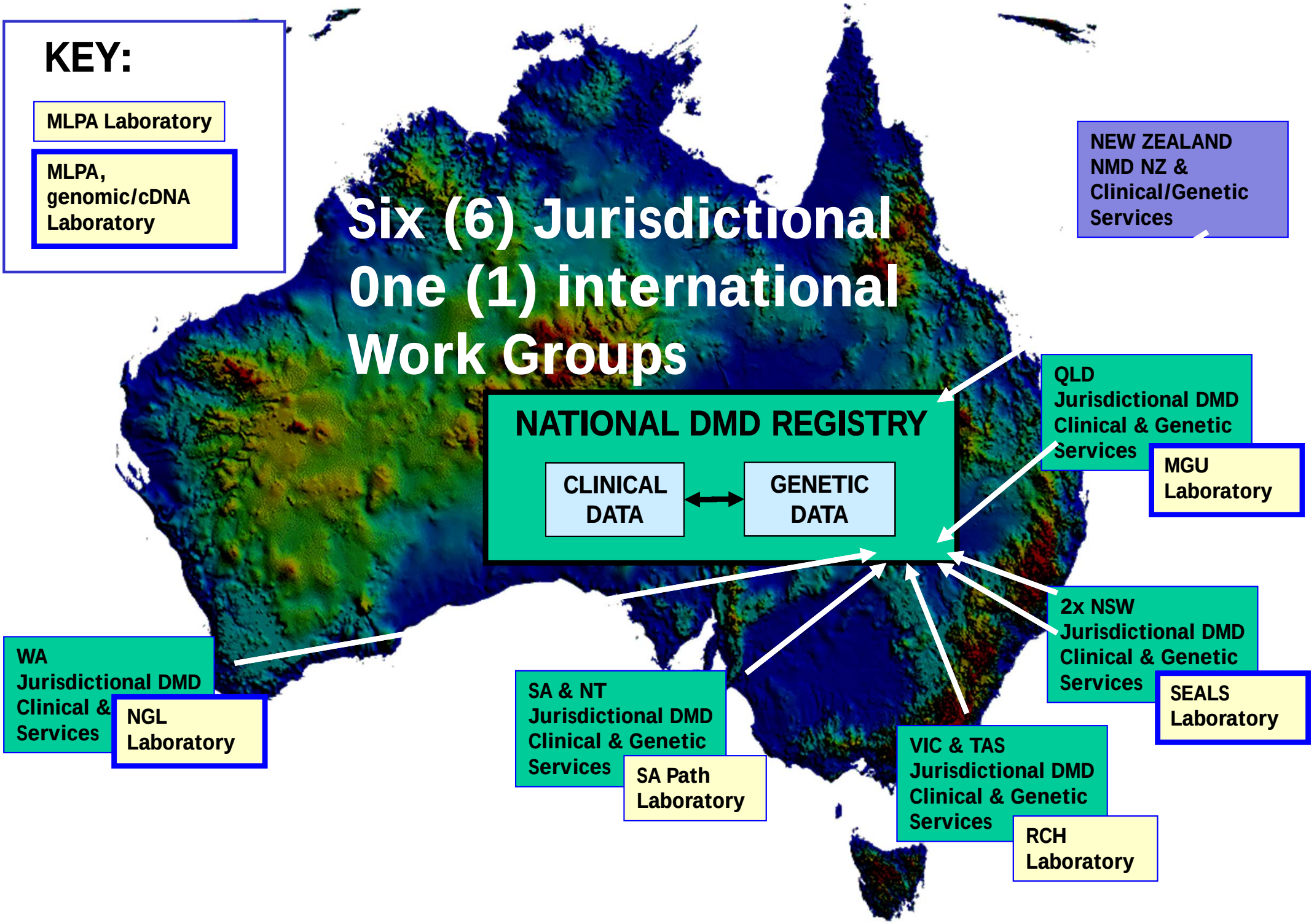
REPORT RECOMMENDATIONS

- Accuracy of the clinical and genetic data as a critical success factor of the Registry;
  - Jurisdictions provide clinical and diagnostic services;
  - Specialist testing laboratories to cross curate genetic data entry; and
  - Clinical service units provide support to register and check the accuracy of clinical data entry.

REGISTRY ARCHITECTURE & FLOW

DNA variation module, clinical data module and report modules within an Internet based application that is restricted from public viewing



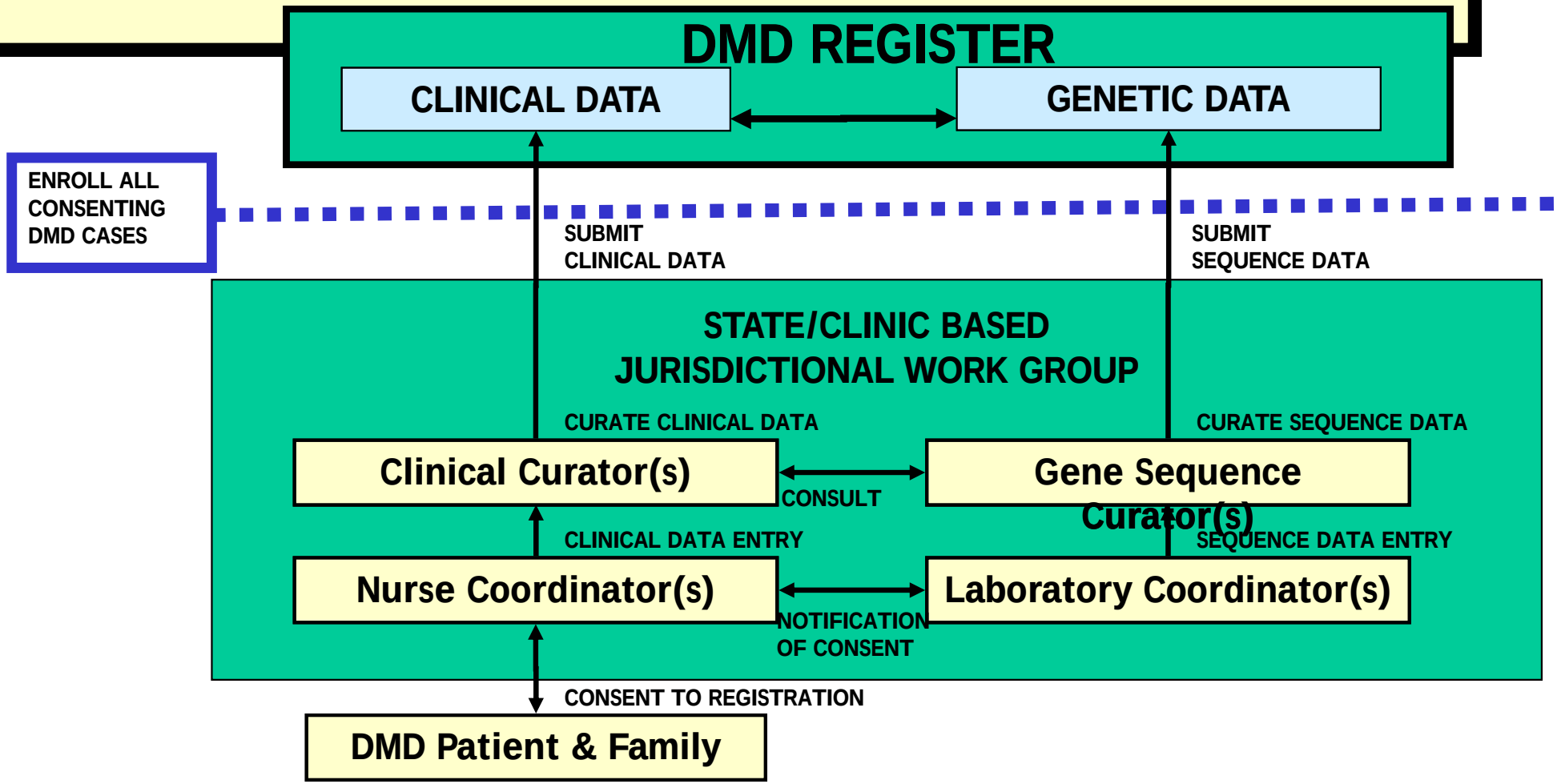

KEY:
MLPA Laboratory
MLPA, genomic/cDNA Laboratory
Six (6) Jurisdictional
One (1) international
Work Groups
NEW ZEALAND
NMD NZ &
Clinical/Genetic
Services
NATIONAL DMD REGISTRY
CLINICAL DATA
GENETIC DATA
QLD
Jurisdictional DMD
Clinical & Genetic
Services
MGU
Laboratory
2x NSW
Jurisdictional DMD
Clinical & Genetic
Services
SEALS
Laboratory
WA
Jurisdictional DMD
Clinical &
Services
NGL
Laboratory
SA & NT
Jurisdictional DMD
Clinical & Genetic
Services
SA Path
Laboratory
VIC & TAS
Jurisdictional DMD
Clinical & Genetic
Services
RCH
Laboratory

NATIONAL REGISTRY CURATOR: H. Dawkins
NATIONAL REGISTRY COORDINATOR: L. Youngs

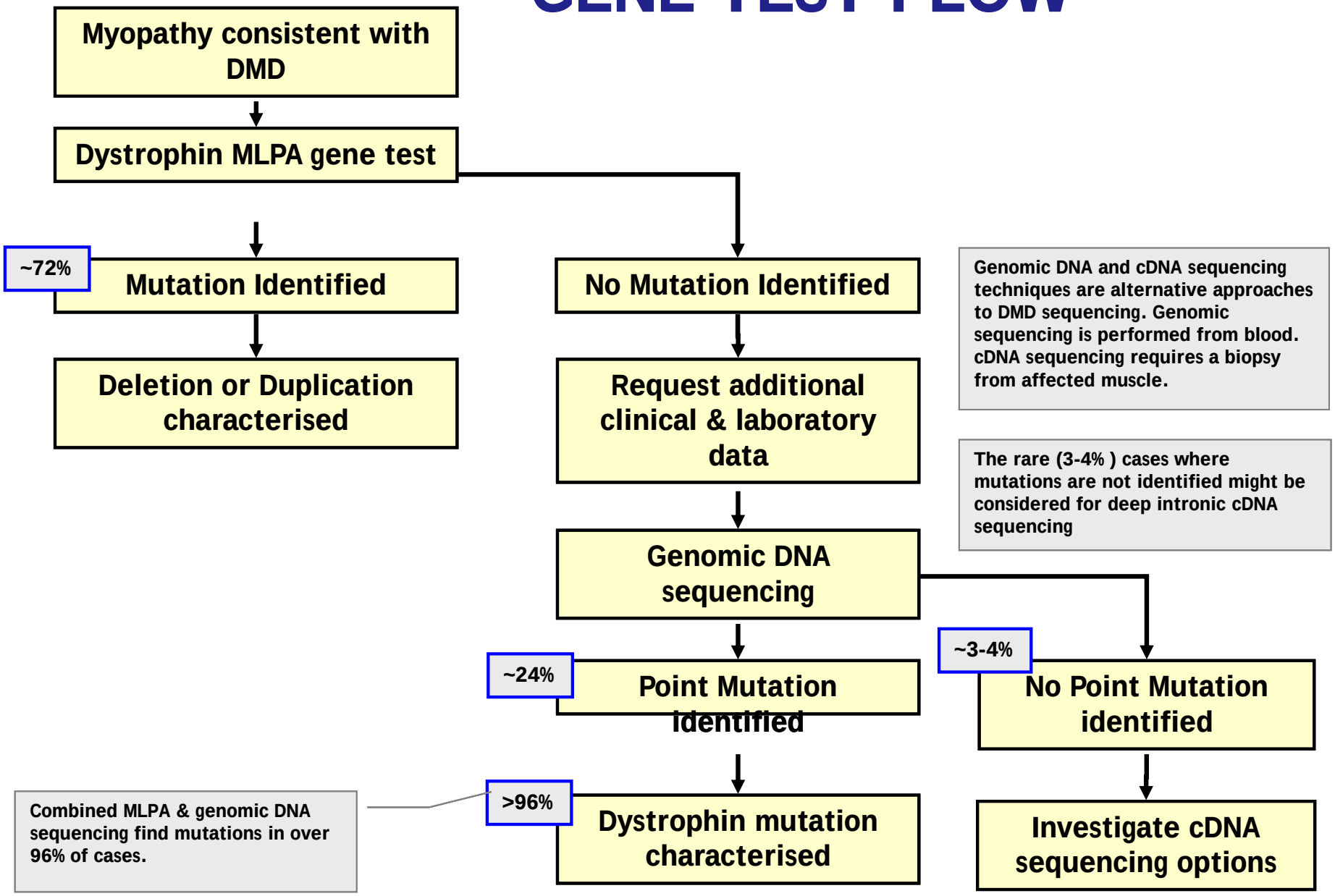
CLINICAL SERVICES
- M. Farrar (NSW)
- K. North (NSW)
- M. Ryan (VIC)
- P. Lamont (WA)
- K. Sinclair (QLD)

GENETIC SEVICES
- P. Taylor (NSW)
- V. Hyland (QLD)
- M. Davis (WA)
- S. Yu (SA)

STAKEHOLDERS
- J. Gummer (MDA Australia)
- D. Robins (Duchenne Foundation)
- B. Struck (MDA Victoria)
- Ethics representative



GENE TEST FLOW



STATUS

UPDATE ON NMD REGISTRIES

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

AUSTRALIAN NMD REGISTRY

- Overarching NMD Registry structure
  - Linear modular architecture
  - Open source
  - Secure
  - Interoperable [TREAT-NMD; Rochester; other registries]
- Ethics Variation
  - NMD registries using same modular format
  - Inform Human Research Ethics Committee
  - Develop NEAF for all jurisdictions
- Provision for New Zealand
- Ontology layers

REGISTRATIONS
February 2013

NMD Registry Registrations (Feb 2013)		
	DMD	SMA
New South Wales	64	
Queensland	42	
South Australia & Northern Territory	10	
Victoria & Tasmania	49	
Western Australia	29	
New Zealand	54	21
Total	248	21

Data Completion
Feb 2013

DMD Registry proportion of clinical and genetic data entered	
Clinical data	
Complete	60%
Partially complete	9%
Incomplete	31%
Genetic data	67%

Registry Issues

- Clinician curator time to enter data
  - a self reported patient questionnaire has been developed for the Myotonic dystrophy registry that requires curator acceptance
  - Adapt for patient registration and compiling registration demographic
  - Adapt to inform clinical data and demographic information updates
  - Deploy across all platforms to alleviate this bottleneck to data entry
- Jurisdictions ethics approvals for NMD registry
  - DMD Registry data represents possibly only 30% of forecast registrations
  - Rate limiting spinal muscular atrophy and Myotonic dystrophy [NZ and WA only jurisdictions with ethics approval]
- Clinical Data Currency Certificate
  - Clinical data which has not been updated for more than 12 months loses currency certification
- Semantic reporting and exchange of information
  - Expansion of registries highlights the need for semantic (ontological)standardisation of integrated genetic reporting ,disease codes and clinical phenotype and common data elements

NEW REGISTRY DEVELOPMENT

Facioscapular humoral dystrophy [FSHD]

- FSHD core dataset endorsed by working group
- Registry development in progress

Congenital Muscular Dystrophy [CMD]

- CMD core dataset endorsed by working group
- Collaborating with CMDIR to maximise interoperability and data exchange
- CMD Spalsh page [external link to CMDIR] for those on the CMDIR and those wishing to be on CMDIR
- Still being discussed feedback from CMDIR to show Australian and New Zealand registrations to reconcile self reported registration information
- Interoperability – aggregated de-identified data; data sharing

FURTHER ITERATIONS

- Ethics variation to store longitudinal data [proxy for natural history]
- Upload ethics PDF – AA+ formats
- Reporting tools and open access data visualisation
- Family history tools
- Semantic standardisation and integration of ontology elements [phenotype; disease classification code; common data elements]
- Enhanced interoperable communication layer
- Upload data to TREAT-NMD

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.

OFFICE OF POPULATION HEALTH GENOMICS

Established 2001

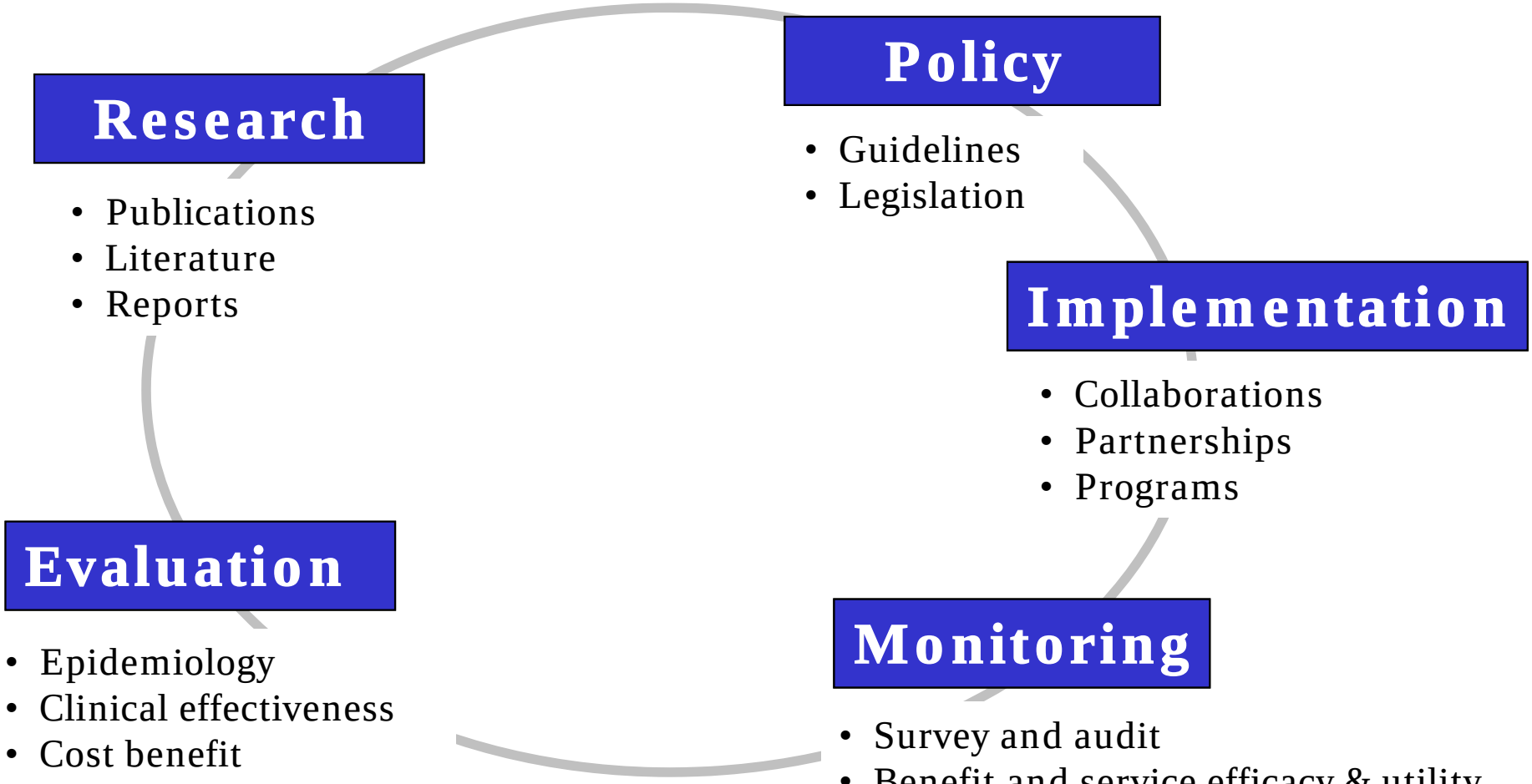
PURPOSE

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

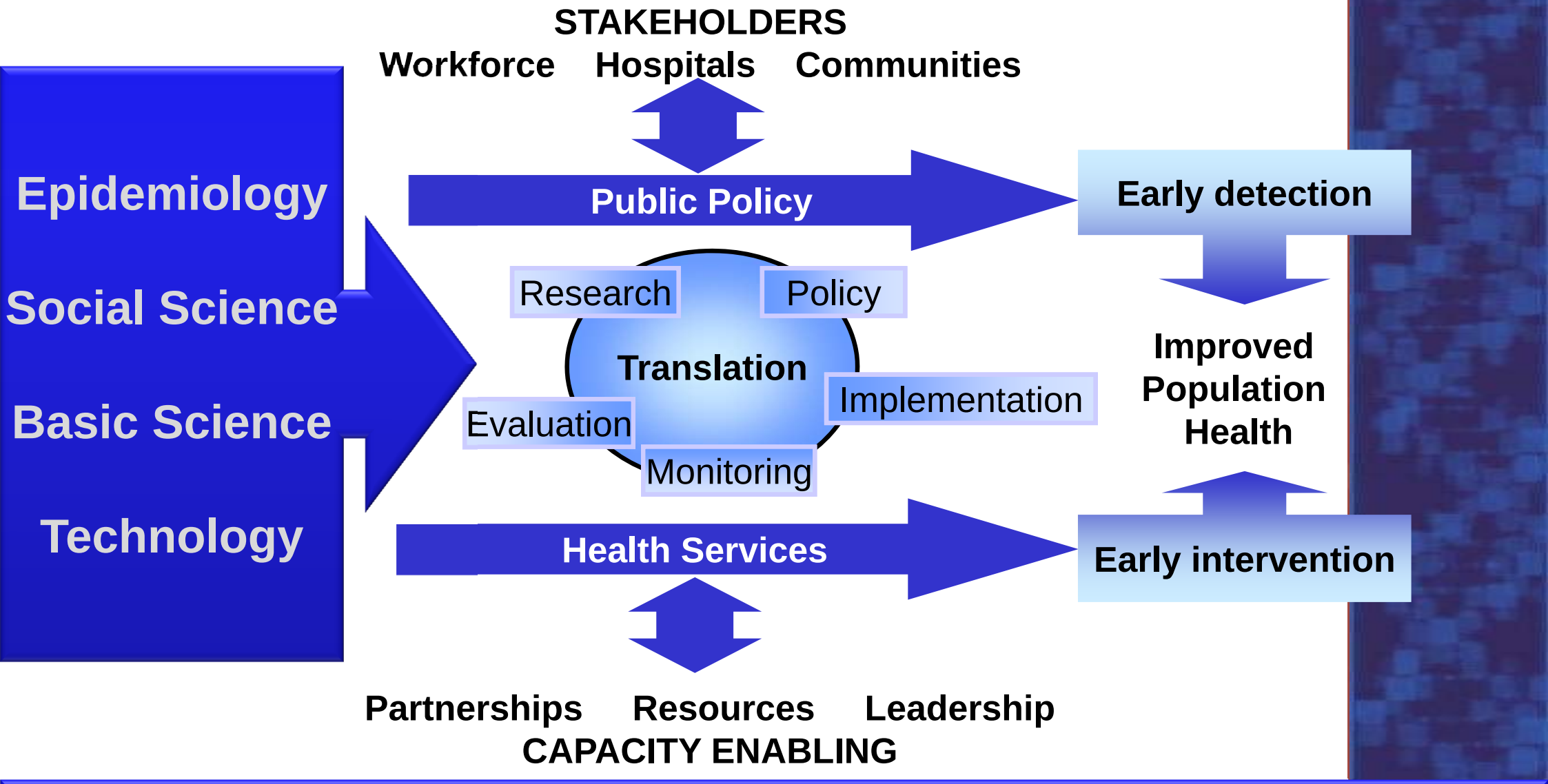
AIMS

- 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.

The virtuous cycle of public health



TRANSLATION OF KNOWLEDGE INTO HEALTH BENEFIT



RARE DISEASE ALLIANCE ORGANISATION [UNMET NEED, AUTHORATIVE VOICE, PARTNERS]

International Rare Disease Research Consortium (IRDiRC)

Co-operation at international level to stimulate, better co-ordinate & maximise output of rare disease research efforts around the world

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC Full Funding Members (25)

North America (12)

Canadian Institutes for Health Research (CA)
Genome Canada (CA)
Sanford Research (US)
Mendelian Disorders Genome Centres(US)
National Centre for Translational Therapeutics (US)
National Cancer Institute (US)
National Institute of Neurological Disorders and Stroke (US)
National Institute of Arthritis and Musculoskeletal and Skin Diseases (US)
National Institute of Child Health and Human Development (US)
National Eye Institute (US)
Office of Rare Diseases (US)
Food and Drug Administration (US)

Europe (12)

European Commission
German Federal Ministry of Education and research
Italian Higher Institute of Health Research
Italian Telethon Foundation
French Association against Myopathies
French National Research Agency
Netherlands Organisation for Health Research and Development
Lysogene (FR)
Prosensa (NL)
Spanish Carlos III Health Institute
UK National Institute for Health Research
Shire (IE)

Western Australian Department of Health

IRDiRC Australia
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

2020 Vision & Milestones

YEAR	DIAGNOSTICS	THERAPIES
2012	• Mapping of sequencing & characterisation efforts • Identified & prioritised gaps in sequencing & diagnostics	• Prioritisation of 100 new or repurposed therapies
2015	• 5000 sequenced/characterised • 3000 diagnostics	• 50 new applications for market authorizations for new or repurposed therapies
2020	• 6000 diagnostics	• 200 new market authorizations given for new or repurposed therapies

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM