Rare Disease Day Policy Event at the United Nations
Second High-Level Event of the NGO Committee for Rare Diseases
21 February, 2018

Establishing an APEC Rare Disease Registry Network QUT and a framework for collaboration

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Asia-Pacific Economic Cooperation

21 Member Economies

2.8 billion people

Facilitates economic growth in region

59% of the worlds GDP

Responsible for 49% of world trade

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Source: https://www.apec.org/About-Us/About-APEC

APEC and Health

APEC Healthy Asia Pacific 2020 Strategy

> *“Recognizing that health is central to development; that it is a prerequisite for, an outcome of, and effective indicator of sustainable development; that health and health equity are integral to public policies of all sectors; that the value of health capital in an economy dwarfs any other form of capital…”*

APEC Roadmap to Promote Mental Welness in Healthy Asia Pacific (2014-2020)

- Recognises that health is a driver of economic growth and prosperity
- In 2014, APEC Leaders endorsed the development of the Healthy Asia Pacific 2020 Strategy

1. Continuing to advance unfinished health-related Millennium Development Goals (MDGs) in a manner that complements activities of the post-2015 development agenda, including de-stigmatization and equality.
2. Strengthening the prevention and control of non-communicable diseases, including mental illnesses, disabilities, violence and injuries. This includes adopting a holistic and multi-sectoral approach that provides continuous health management and early diagnosis/treatment.
3. Strengthening health systems to support Universal Health Coverage, providing the whole population with access to safe, effective, quality, affordable and sustainable health care.
4. Improving health emergency preparedness, surveillance, response and recovery systems for public health emergencies, including pandemic events and natural disasters.

Source: APEC Health Working Group & APEC Life Science and Innovation Forum, Health Asia Pacific 2020 Roadmap (2014).

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Rare Disease in APEC

Problem

- Approximately 400 million people affected globally, with approximately 200 million individuals impacted across the APEC Member Economies
- 50% of cases occurring in children
- Many patients lack access to appropriate diagnosis and access to therapy

Progress

- Variety of approaches to rare disease policy within member economies
- Ongoing development of healthcare systems and expansion of coverage
- Barriers to care remain significant due to variable factors including health infrastructure, cost and access to diagnostic services

Opportunity

- Engage with Governments and stakeholders across the region to focus on rare disease policy
- Partner with policy makers as health systems evolve to ensure they adequately address the needs of the rare disease community.

Source: APEC Rare Disease Network. Presentation to the APEC Life Science and Innovation Forum – SOM 3 Meeting, August 2018.

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APEC Rare Disease Expert Network

Workplan Overview

1. LSIF Endorsement of RDN Concept Note
2. APEC Rare Disease Economy Survey/Issues paper
3. Undertake Economy Consultations
4. Hold APEC Rare Disease Policy Dialogue
5. APEC Rare Disease Action Plan
6. Develop implementation plan

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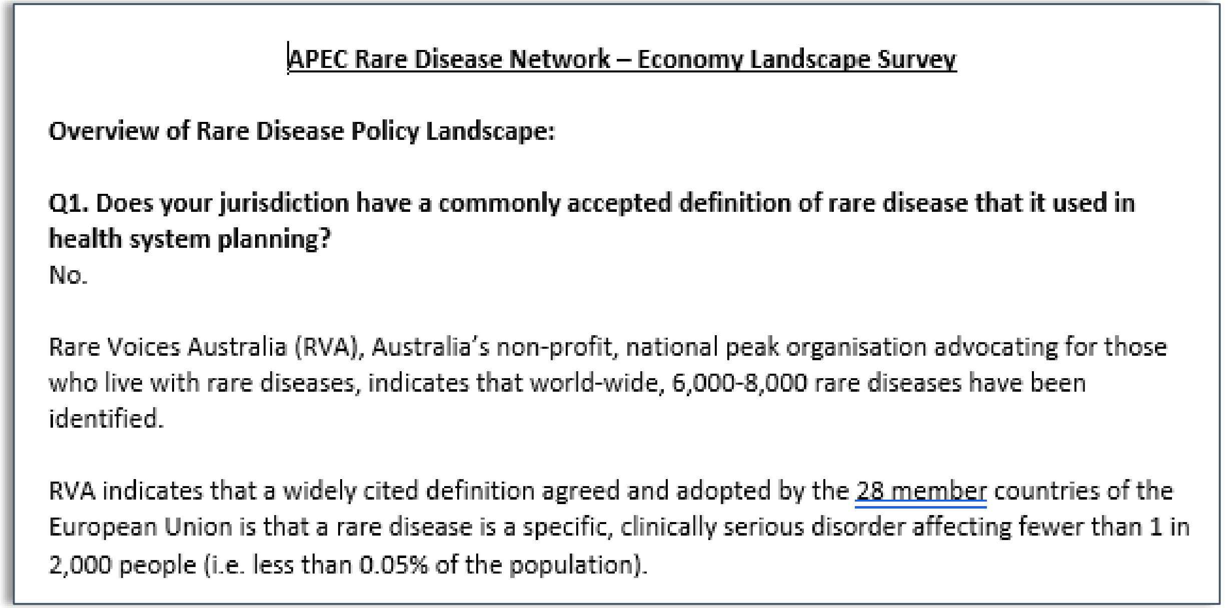
Capturing Base Line

APEC RDN Economy Survey and Issue Paper



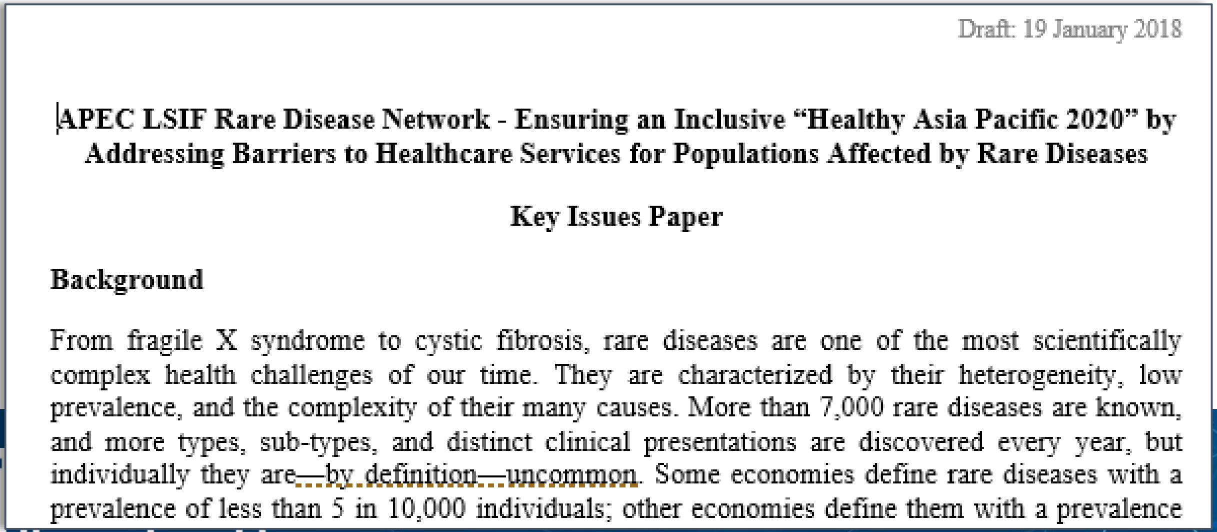
APEC Rare Disease Network – Economy Landscape Survey

Overview of Rare Disease Policy Landscape:

Q1. Does your jurisdiction have a commonly accepted definition of rare disease that it used in health system planning?
No.

Rare Voices Australia (RVA), Australia's non-profit, national peak organisation advocating for those who live with rare diseases, indicates that world-wide, 6,000-8,000 rare diseases have been identified.

RVA indicates that a widely cited definition agreed and adopted by the 28 member countries of the European Union is that a rare disease is a specific, clinically serious disorder affecting fewer than 1 in 2,000 people (i.e. less than 0.05% of the population).



Draft: 19 January 2018

APEC LSIF Rare Disease Network - Ensuring an Inclusive "Healthy Asia Pacific 2020" by Addressing Barriers to Healthcare Services for Populations Affected by Rare Diseases

Key Issues Paper

Background

From fragile X syndrome to cystic fibrosis, rare diseases are one of the most scientifically complex health challenges of our time. They are characterized by their heterogeneity, low prevalence, and the complexity of their many causes. More than 7,000 rare diseases are known, and more types, sub-types, and distinct clinical presentations are discovered every year, but individually they are—by definition—uncommon. Some economies define rare diseases with a prevalence of less than 5 in 10,000 individuals; other economies define them with a prevalence

- Economy Landscape Survey sent to all 21 APEC Economies

- Utilised survey responses to supplement desktop research

- Drafted an APEC Rare Disease Key Issues Paper
  - *Circulated to Member Economies for review/feedback*
  - *Provided to stakeholders during consultations*

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Developing real-world insights

APEC RDN Country Consultations

Peoples Republic of China

Thailand

Viet Nam

Republic of Korea

Chinese Taipei

Australia

6

Government

Patient Orgs

Clinicians

Academia

55

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Articulating problems, developing solutions

APEC Rare Disease Policy Dialogue – 6-7June, Beijing CHINA

Overview

- 2 days
- 75 attendees
- 14 Economies
- 30 presentations across 9 critical policy areas
- Views represented:
    - Government
    - Academia
    - Clinicians
    - Patients
    - Industry

*"Talk is good, action is better"**

* Summit participants call to action

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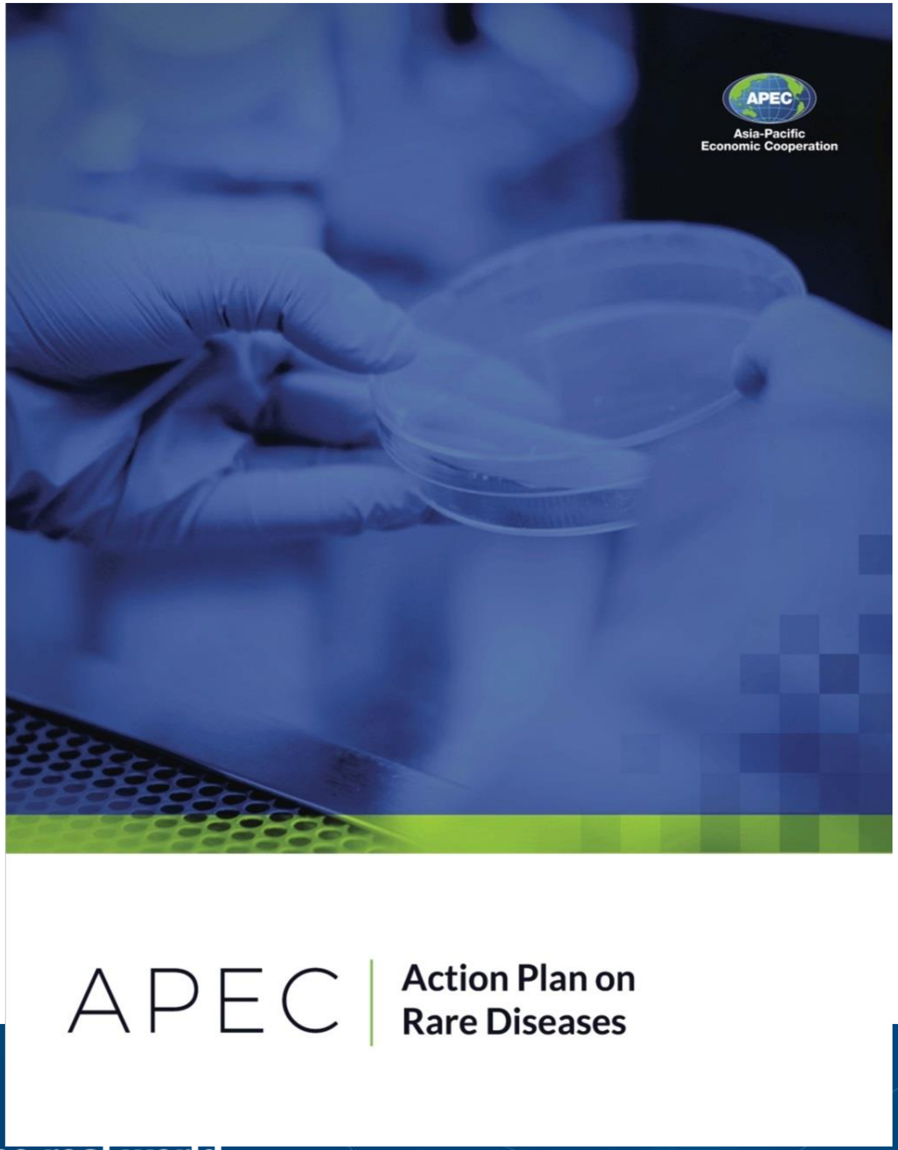
A Road Map for Rare Disease

APEC Rare Disease Action Plan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APEC | Action Plan on Rare Diseases

- Sets out a clear vision for APEC Member Economies to improve the economic and social inclusion of people living with rare diseases

- Identifies 10 policy pillars that require action:
  - Summarises issues
  - Identifies targets for policy action
  - Outlines indicators to measure progress
  - Develops a clear action plan for economies to follow

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APEC Action Plan on Rare Diseases Overview

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Action Plan Pillars

1. Define rare disease and orphan products with policies and processes
2. Raise public and political awareness of rare disease issues
3. Promote innovative research and development
4. Build human resource capacity in medical and non-medical sectors
5. Facilitate early, accurate, and systematic diagnosis
6. Coordinate patient-centred care across medical speciality, life course and location
7. Deliver new and accessible treatments to patients
8. Support financial and social needs of patients and their families
9. Manage pooling and usage of patient data securely and effectively
10. Prioritize comprehensive domestic rare disease policy

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10 Prioritize comprehensive domestic rare disease policy

Recommendation 10.1

2025 Target: APEC member economies each have developed and published non-binding but comprehensive, whole-of-government, and medium- to long-term plans for addressing rare diseases in each of their domestic contexts.
Indicator: Percent of economies with a non-binding, comprehensive domestic rare disease plan.

Recommendation 10.2

2025 Target: APEC member economies each have integrated legislative provisions for rare diseases into other areas of legislation outside healthcare such as social security, disability, employment, and housing.
Indicator: Percent of economies with binding, legislative rare disease provisions in other policy areas.

Recommendation 10.3

2025 Target: APEC member economies each have enacted enforceable, comprehensive legislation, policy, or mechanism at least covering provisions on the research, diagnosis, and treatment of rare diseases.
Indicator: Percent of economies with comprehensive domestic legislation, policy, or mechanism.

RECOMMENDATION 10.1

2025 Target

- 2025 Target: APEC member economies each have **developed and published non-binding but comprehensive, whole-of-government**, and medium- to long-term **plans for addressing rare diseases in each of their domestic contexts**.

Indicator

- Percent of economies with a non-binding, **comprehensive domestic rare disease plan**.

Actions

- Generate political will in partnership with industry, academia, clinicians, and patient groups to **develop and publish an economy-wide plan on rare diseases and orphan products** that
    1. **incorporates actionable strategies** on key policy areas that require development;
    2. **targets and prioritizes research and development areas** depending on economy strengths and needs;
    3. **evolves over time** to match the domestic rare disease context and community; and
    4. **integrates monitoring and financing components** to accelerate action and maintain momentum.
- Identify a **government focal point** to convene various government entities relevant to addressi**ng rare diseases.**

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Rare Disease base-line data is essential

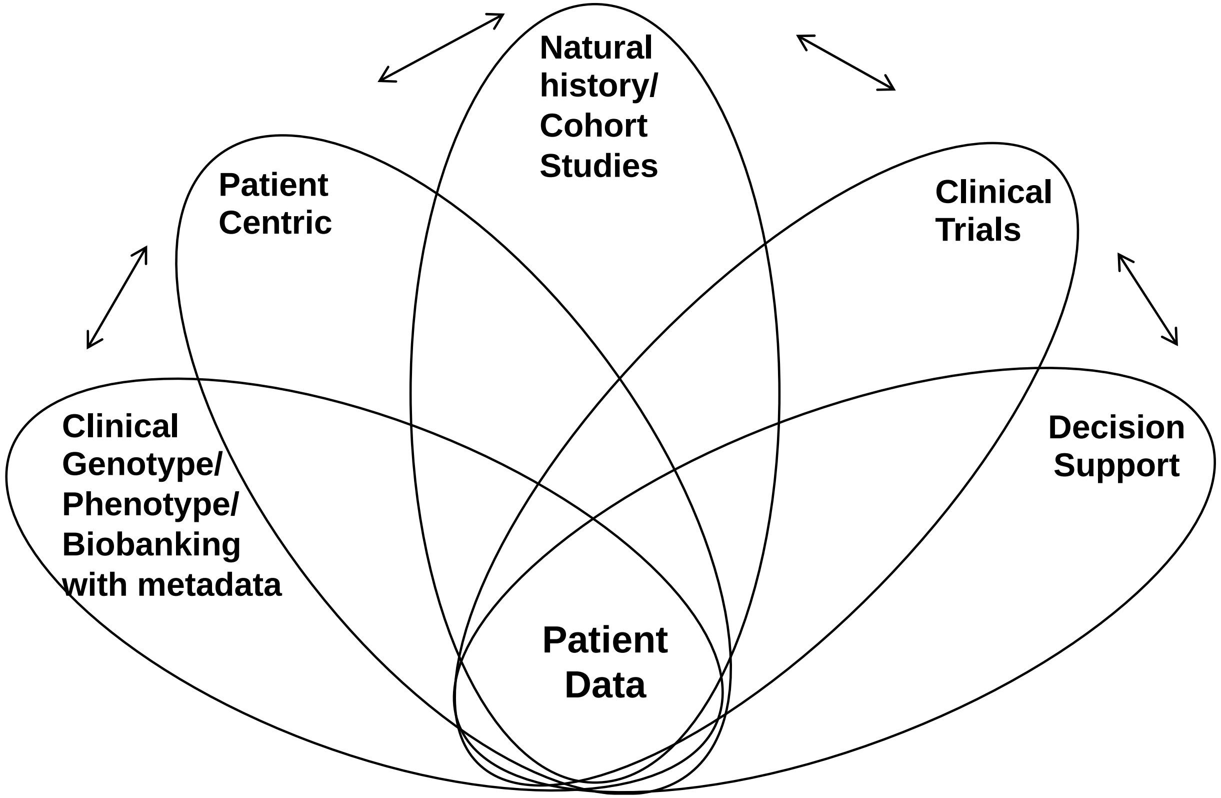
- **Better utilization of patient data** provides a significant opportunity to better support those living with a rare disease – enabling **adaptive/optimal/informed decision making**
  - For example, the right treatment, for the right patient and leveraging what has been learnt from past decisions to optimise future decisions
- In rare diseases there is **inherent heterogeneity** in the population such that individualised treatment/care is needed
  - However, of equal importance is **building the evidence base for the rare disease population**
- We need to **learn from individual experiences** and **aggregate** across the **rare disease population** to find **generality in disease progression, management and treatment**
- We can then catere to individual needs and also learn from collective experiences
  - better **disease diagnosis and management**, **personalized therapeutic interventions**, and as a catalyst for **new and innovative research and development**
  - **underpin public health and clinical research** and to **inform health service design and delivery**
  - **promote and disseminate** new knowledge to inform best **clinical practice and care**, **identify and recruit volunteers for clinical trials, and to enable seamless integration with clinical trial**

Registry should be a dynamic platform that captures and interrogates data

- Registry **time-scales**
    - Short-term, medium-term, Long-term time-frames

- Registries system **should** capture data required by all stakeholders and make outcomes accessible and useful for decision making

- It should have the **capacity to add** additional conditions/data collection centres as required



Natural history/ Cohort Studies

Patient Centric

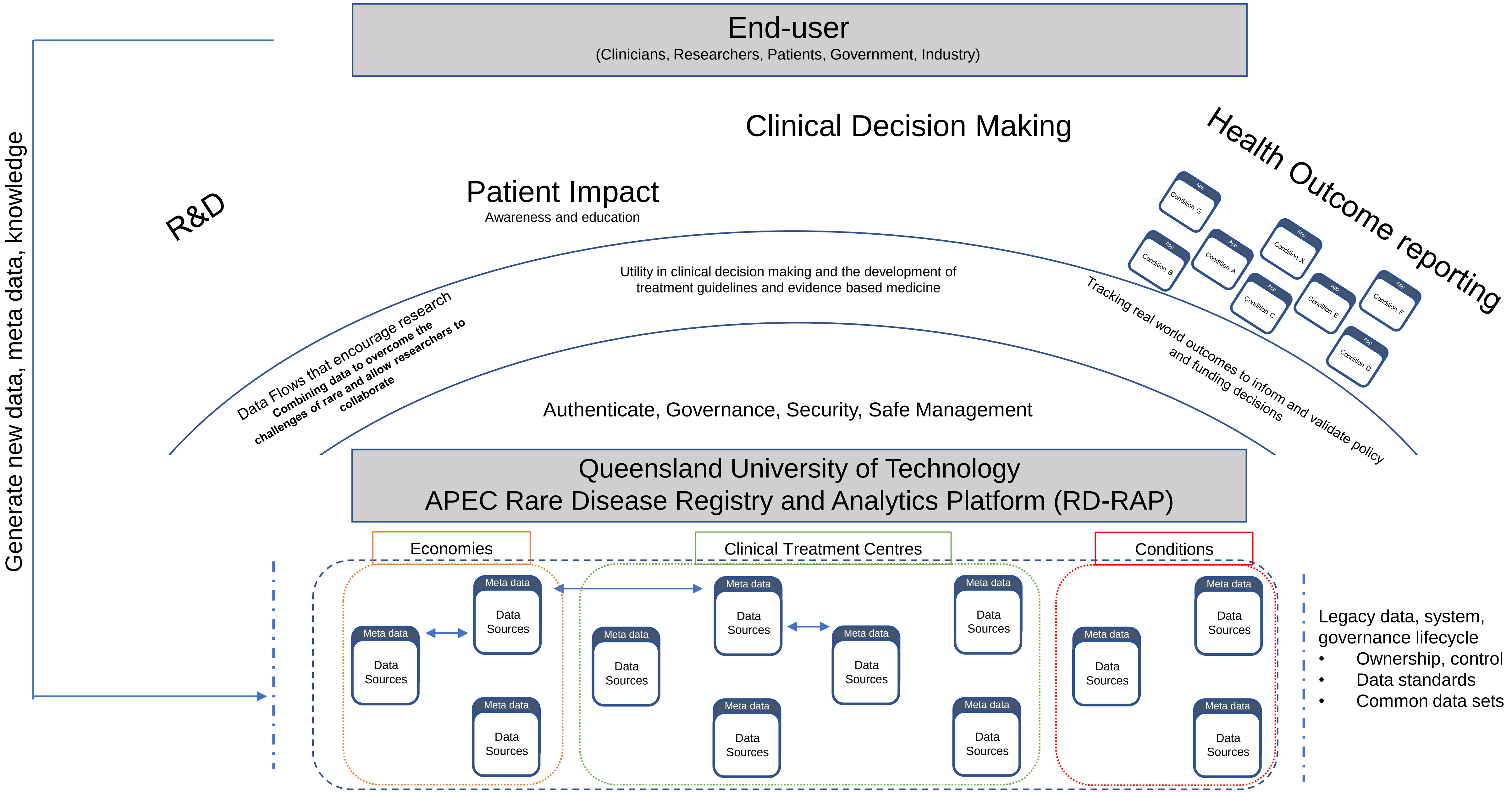
Clinical Trials

Clinical Genotype/ Phenotype/ Biobanking with metadata

Decision Support

Patient Data

Bellgard M, Embracing digital disruption to advance clinical research, 2017 ACS Distinguished Oration.



End-user

(Clinicians, Researchers, Patients, Government, Industry)

Clinical Decision Making

Patient Impact

Awareness and education

R&D

Health Outcome reporting

Utility in clinical decision making and the development of treatment guidelines and evidence based medicine

Data Flows that encourage research

Combining data to overcome the challenges of rare and allow researchers to collaborate

Tracking real world outcomes to inform and validate policy and funding decisions

Authenticate, Governance, Security, Safe Management

Queensland University of Technology
APEC Rare Disease Registry and Analytics Platform (RD-RAP)

Economies

Clinical Treatment Centres

Conditions

Legacy data, system, governance lifecycle

- Ownership, control
- Data standards
- Common data sets

Generate new data, meta data, knowledge

Issues to be addressed to make an APEC RD-RAP a reality

- Governance
  - Security and operations - Data sharing access
  - What data should be collected
  - Who should get access to data
- Regulation
  - Economies have different regulatory frameworks covering collection of data
  - Sharing of data across national borders
  - Privacy
    - different economies regulate privacy differently
- Sustainability
  - How is the registry paid for?
  - How does it grow and change to meet the evolving needs of stakeholders?

QUT
BRISBANE
AUSTRALIA

- QUT to lead development of an APEC Rare Disease Registry and Analytics Platform
  - Partner with stakeholders through APEC to address these issues
  - Host the registry
  - Lead research efforts to address the needs of the rare disease community