

Precision Public Health:

Translating new (genomic) knowledge into care pathways and health service planning

Rare Disorders Research Forum

Christchurch

Aotearoa New Zealand

5th and 6th August 2026

Rare Disease Policy

Policy has the power to transform the health system and patient experiences

GOOD POLICY

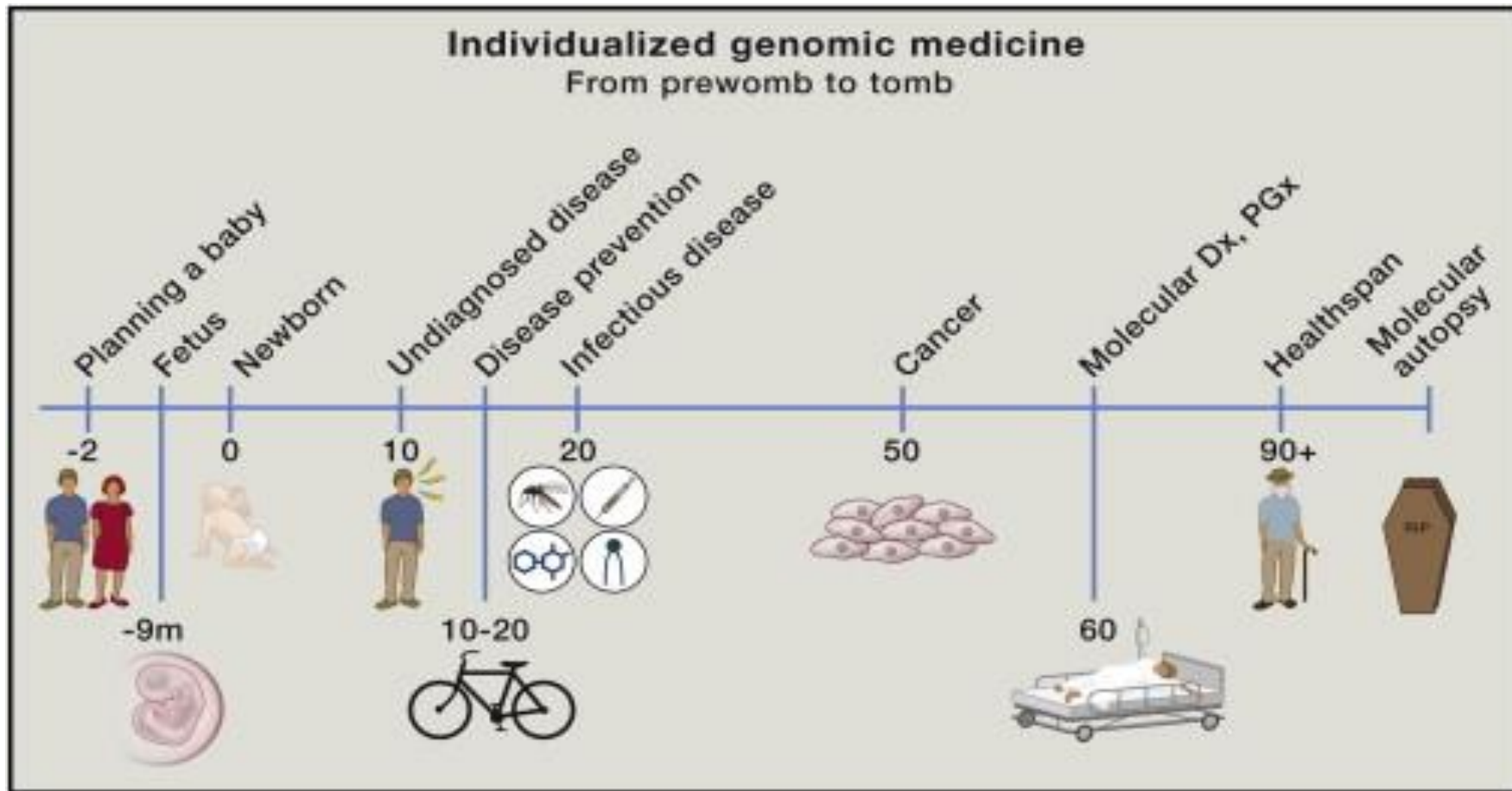
- **Supports clinical innovation**; goes beyond well-written documents
- **Begins and ends with stakeholders**; uses substantive co-development that ensures industry and public are deeply invested
- **Ensures outcomes benefits the most vulnerable**; equitable access and optimised outcomes
- **Capitalises on appreciative approaches** that capture ‘what works well’ and builds baseline trust
- **Capable of incorporating new knowledge and cohesively integrating multiple sources of truth** [e.g. medical literature and clinical knowledge; with real world evidence and data collections; with lived-experiences]
- **Uses deliberative approaches to resolve perceived points of disagreement to build genuine consensus and creates deeper trust**
- **Is able to achieve the highest attainable standard through adoption of pragmatic approaches** that can be **agile efficaciously integrated** into health system design and create holistic patient care pathways
- **Is dynamic, it embraces serendipity**, and thus capable of driving improvement and efficiencies through iterative innovation steps

Precision Medicine Initiatives: atomising disease into genome-phenome **building knowledge on the n of 1**

Precision Public Health Initiatives: policies that regroup the people in the **Precision Medicine** landscape to ensure equitable access and equitable (optimised) outcomes for all citizens. **Transformative translation** applying the n of 1 to “**1 to the many**”

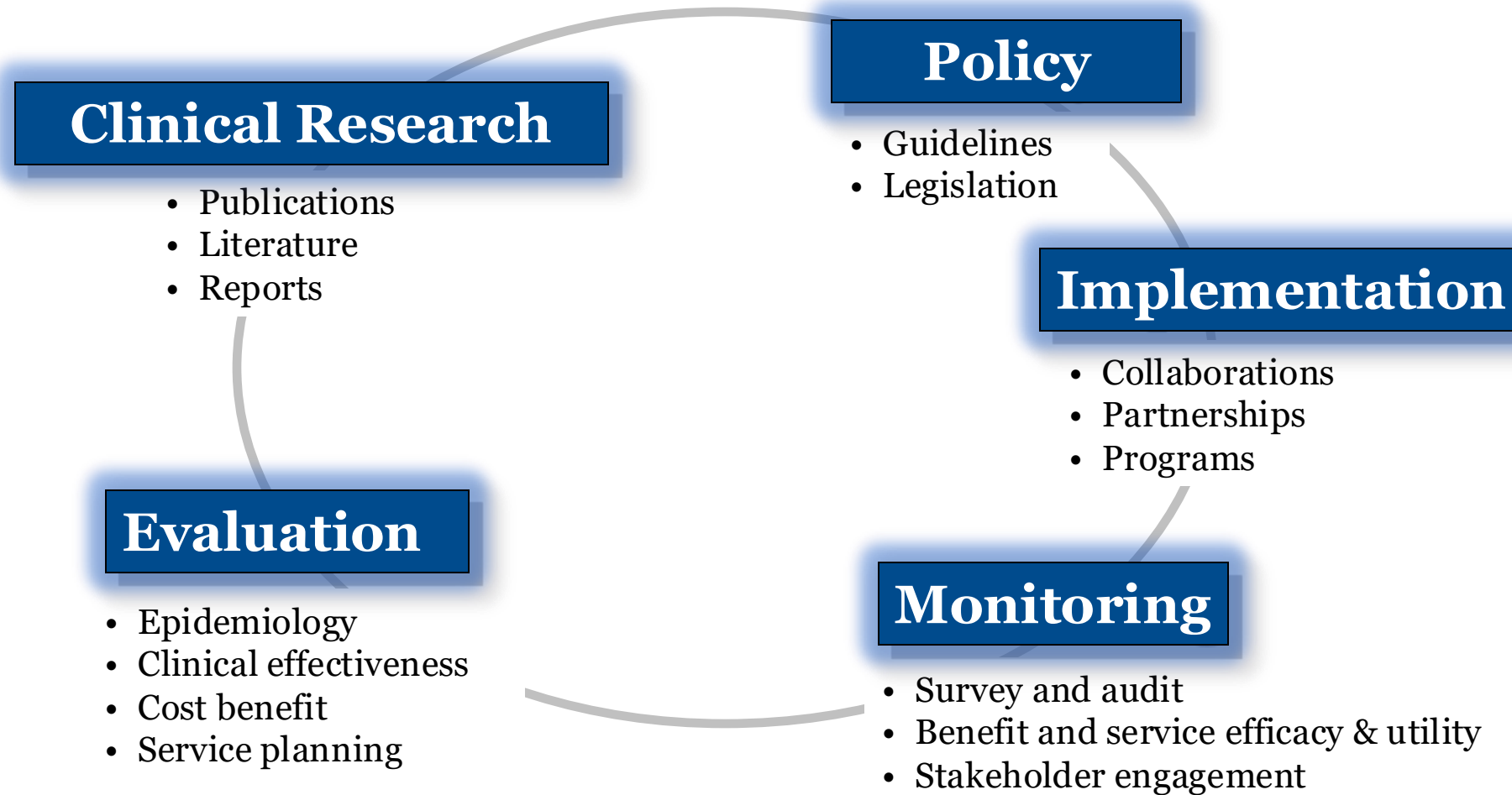
Precision Public Health

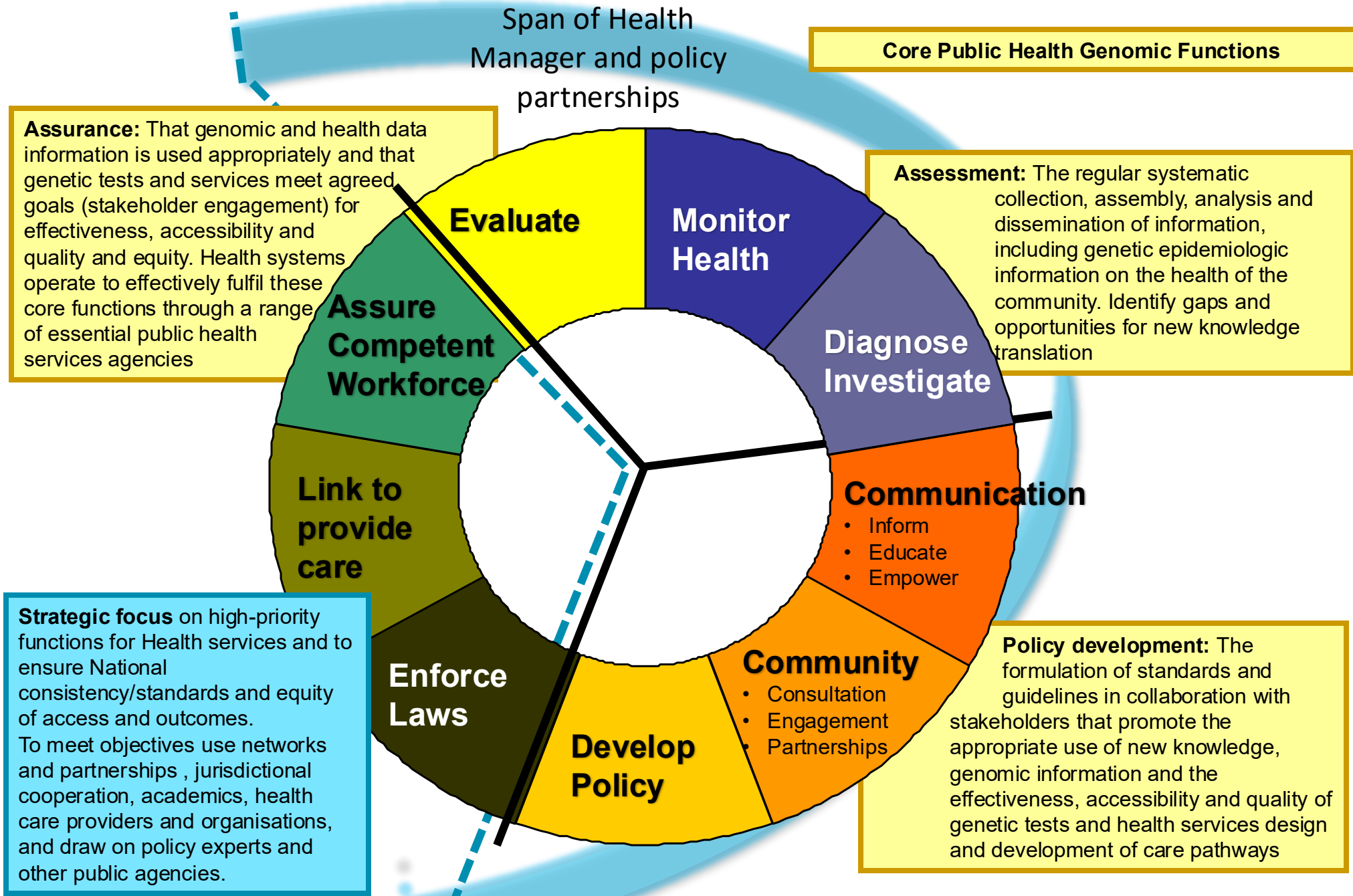
“The application and combination of new and existing technologies which precisely describe individuals and their environment, in order to tailor preventive interventions and improve overall population health”



Timeline of Sequencing Applications in Medicine from Prewomb to Tomb: The medical application of genomics is relevant to many points during an individual's lifespan.

The virtuous cycle of public health





INCREASING LEVEL OF PUBLIC EMPOWERMENT (IMPACT)

	Inform	Consult	Involve	Collaborate	Empower
Participation Goal	To Provide balanced and objective information to assist them in understanding the problem, alternatives, opportunities and/or solutions	To obtain public feedback on analysis, alternatives and/or decisions	Work directly with the public(s) throughout the process to ensure that concerns, and aspirations are consistently understood and considered	Partner with the public(s) in each aspect of the decision including development of alternatives and identification of the preferred solutions. Manage all parties	To place final decision-making in the hands of the public
Promise to the Public	We will keep you informed	We will keep you informed, listen to and acknowledge concerns and aspirations, and provide feedback on how public opinion influenced the decision	We will work directly with you and the wider public(s) to ensure concerns expressed are directly reflected in the alternatives developed and the how public input influenced the decisions	We will partner with you and the wider public(s) in each aspect of the decision and innovation processes. Your advice, recommendations will be incorporated to the maximum extent possible.	We will implement your decisions

THE RARE DISEASE ECO SYSTEM **is an exemplar of** **PRECISION PUBLIC HEALTH**

Policy has the power to transform the health system and patient experiences

BIOINFORMATICS ECO SYSTEM AND KNOWLEDGE MANAGEMENT

EXPERT HUMAN KNOWLEDGE

- Implicit and explicit knowledge

MACHINE SYSTEMS

- Information capture
- Systems Integration
- Analysis
- Interpretation

VOICE OF THOSE AFFECTED

- RD patients, families, carers
- Clinical & Academic
- Industry & Manufactures
- Government
- Hospital (private & public)

PEOPLE LIVING WITH RARE DISEASES

- Equity of Access
- Lead the best life possible
- Quality of life choices and measures
- Improve health system experience
- Empowered with knowledge
- Influence health care
- Opportunity to influence policy

PHENOTYPE
ENABLED
DIAGNOSTICS

SYSTEM PLANNING

Evidence, Behaviour, Demographic,
Socio-Economic, Environment

DATA
MATCHMAKING

VOICE OF EXPERTS

- Technology
 - devices, instruments, bioinformatics, systems
- Clinical expertise/experts
- Natural History
- Integrated Omics
- Interpretation & application

VOICE OF DATA (EVIDENCE)

CONTINUOUS KNOWLEDGE
MANAGEMENT CYCLE

NEW
KNOWLEDGE

WHAT TO USE (NOT TO USE)

- Genotype:Phenotype
- Treatment monitoring
- Therapeutic targets
- Biomarkers
- Trial Optimisation

PRECISION
MEDICINE

WHERE & WHEN TO INTERVENE

- Disease classification & codification
- Incidence, prevalence, Burden of Disease
- Best practice
- Knowledge transfer
- Capacity building

PRECISION
HEALTH

POLICY
IMPLEMENTATION

THE SOLUTION

Policy Hooks

Align objectives to fit Government/Health system imperatives **to improve health outcomes** for their respective population; **by reducing the burden of chronic-multisystem diseases** on health system budgets through early diagnosis and disease classification (Snowmed and ICD-11); **enabling hospital metrics and economic data supporting care** and helping inform health system design changes and ensure sustainability of quality health systems; **using RD data to help drive innovation and discovery for new treatments and therapies**; and to reduce economic productivity losses due to multi-system chronic conditions .

ENABLERS: Build cogent compelling arguments for government and health system policy makers using currently available sociological-psychological-productivity data/evidence into to high level briefs and support impacts in relation to sustainability and reducing burden of RD and multi-system disorders.

R&D Hooks

Initiate new study design by first scoping out regulatory requirements that will (i) guide and inform the implementation of new knowledge, and thus translation of outcomes into direct health benefits for their target population; (ii) will be supported by their power calculation requirements for epidemiological purposes; (iii) ensure FAIR and CARE principles are adhered to for equity of accesses and equitable outcomes for the beneficiary population(s).

ENABLERS: FAIR and CARE principles to improve equity for Indigenous populations , minority ethnic groups , and LMIC and resource poor and impoverished populations.

RARE DISEASES ARE MEDICAL CONDITIONS FOR WHICH:

- There exists limited understanding of the currently characterised 6,172 RD and for which the current upper estimate of RD is 10,876 conditions
- **Globally**, indirect RD point prevalence is estimated between 1.2 to 1.6 persons/100,000 population
- **Collectively**, RD affect between 3.5% and 5.9 % of the of the global population affecting between 262,900,000 – 446,200,000 PLWRD at any time
- **RD without known monogenic causes include rare cancers (11% - comprising about 24% of adult and 100% of all childhood cancers), infectious diseases (2.6%), autoimmune diseases and rare poisonings (0.4%).**
- **WHO** International Classification of Diseases (ICD)10 revision had Unique RD Identifier (URI) codes for about 450 RD, less than 4% of characterised RD.
- Insufficient ICD10 URI codes means most PLWRD do not have their RD diagnosis entered into hospital records; and are invisible in the respective health system records.
- RD data when present in hospital records are sparse, scattered, or incomplete and are often not easily identified in health information systems and health data collections.
- **WHO ICD11 has more than 5,500 RD** classifications and URI codes; including URI for *Undetermined Rare Diseases*; undiagnosed RD; a 12-fold increase from ICD10.

Haendel et al. (2020) How many rare diseases are there? *Nat Rev Drug Discovery*. (2020) 19(2):77–8. doi: 10.1038/d41573-019-00180-y. Accessed 17March 2024.

Lamoreaux et al (2022) RARE-X The Power of Being Counted: a more accurate count of rare diseases and steps to getting counted <https://rare-x.org/wp-content/uploads/2022/05/be-counted-052722-WEB.pdf> Accessed 15 April 2024

Nguengang Wakap et al. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* **28**, 165–173 (2020). <https://doi.org/10.1038/s41431-019-0508-0> Accessed 29 February 2024.

Vicente et al (2021) Regarding the estimations of people affected by rare diseases. *Eur J Hum Genet*. 2021 Jun;29(6):1032-1033. doi: 10.1038/s41431-020-00763-z. Accessed 29 February 2024

World Health Assembly Resolution on Rare Diseases

On 24 May 2025, the Seventy-eighth World Health Assembly (WHA78) voted unanimously for the resolution
“Rare diseases: a global health priority for equity and inclusion,”

- **WHO has formally elevated rare diseases to the top tier of its agenda.**
- The text **urges governments to integrate rare-disease policies in national health plans, expand newborn screening and timely diagnosis**, and guarantee affordable access to medicines and assistive technologies under universal health coverage. It also **encourages investment in digital tools, patient registries and centres of excellence, while promoting the active participation of patient organisations in policy design.**
- **Member States acknowledged that more than 300 million people live with one of over 7 000 distinct rare conditions**—70 % of which begin in childhood—facing delayed diagnosis, fragmented care and significant social and financial burdens.
- **A central provision in the resolution instructs WHO to draft a comprehensive 10-year global action plan by 2028**, complete with measurable targets and an accountability framework. The plan will map existing standards, identify technological innovations (including AI-enabled diagnostics) and outline mechanisms to share data across borders
- **Policy analysts agree the resolution creates fresh momentum for cross-disciplinary research consortia and public-private partnerships to work with governments** is an unprecedented mandate that could reshape research pipelines and regulatory pathways worldwide.

DRAFT GPW14 THEORY OF CHANGE

OVERALL PRINCIPLES & APPROACHES

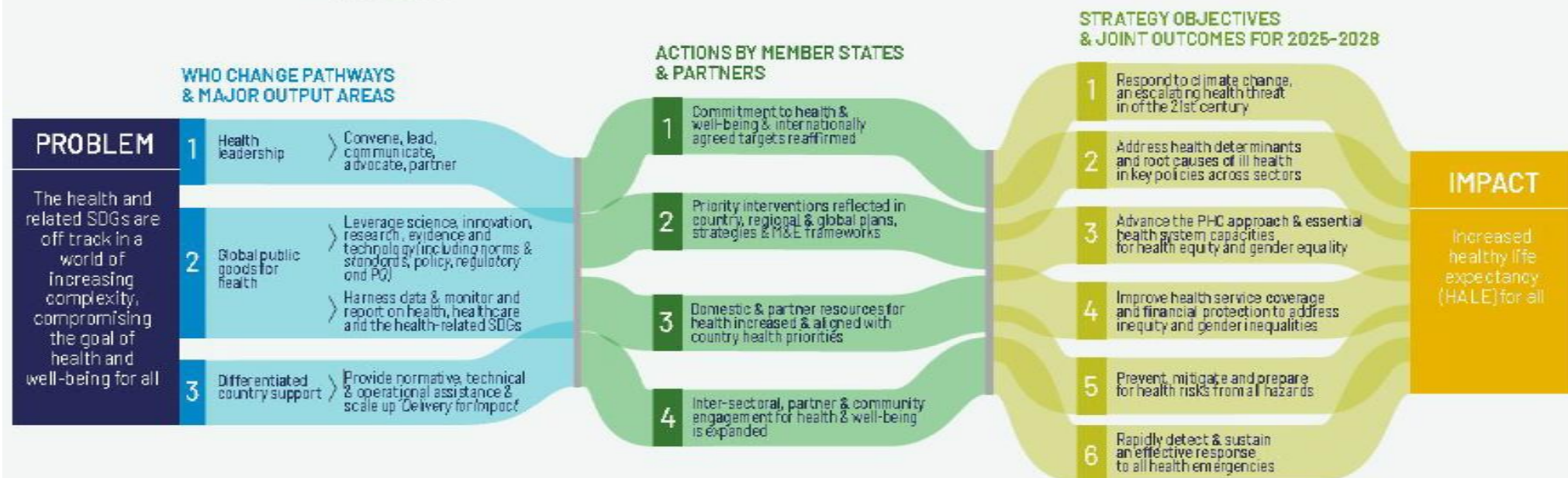
Scale up the Primary Health Care approach for UHC & health security, to promote equitable, cost-effective, integrated, people-centred care, especially for the underserved

Respect and empower national leadership, structures & processes for the governance of health to ensure the alignment of health players

Maintain a relentless focus on delivering measurable impact at the country level using approaches that enhance programmatic accountability

Advance gender equality, health equity and human rights to overcome barriers to health and well-being for all

Enhance and expand partnerships, community engagement and intersectoral collaboration



ENABLERS (WHO CORPORATE OUTCOME #4)



Strengthened WHO Country and Regional Office capacities



A sustainably, flexibly financed WHO



A diverse, motivated, fit-for-purpose and empowered WHO workforce



Enhanced vertical & horizontal integration and ways of working



A more effective, efficient and accountable WHO

ASSUMPTIONS & STRATEGIC RISKS (CONDITIONS FOR THE TOC TO BE VALID)



The expanding use of new and emerging technologies, such as artificial intelligence, in a rapidly evolving communications landscape, does not exacerbate and accelerate the spread of mis- and dis-information



Member States & health partners remain committed to the SDGs and there is sustained political will to keep health high on the agenda



Sufficient financing is available for critical health priorities (i.e. domestic income & fiscal policy remain conducive to health priorities)



Focus can be maintained over time (e.g. no major health emergency requiring a significant repurposing of the global health architecture)

WHO theory of change for the draft GPW 14. Reproduced

PROBLEM

The health and SDGs are off track in a world of increasing crises and complexity, compromising the goal of health and well-being for all.

OVERALL STRATEGIC APPROACH FOR 2025-2028

1. Scale up the **Primary Health Care** approach for UHC and health security, to promote equitable, cost-effective, integrated quality care, especially for the vulnerable
2. Fully empower **national leadership, structures & processes** for governance of health to ensure alignment of health players
3. Maintain a relentless focus on delivering **measurable impact at country level** (incl. *Delivery for Impact*, SDG stocktaking)
4. Enhance action of **gender equality, health equity and human rights** as preconditions for health & well-being for all
5. **Expand partnerships, community engagement, and intersectoral collaboration**

IMPACT

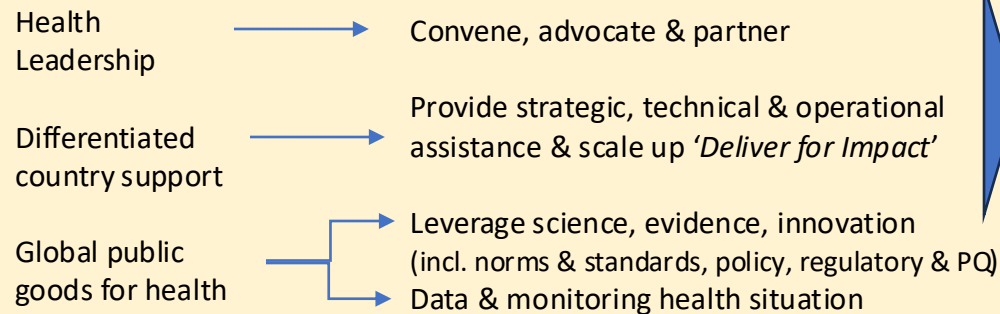
Increased healthy life expectancy (HALE) for all

GPW14 STRATEGIC OBJECTIVES

- Respond to **climate change**, the greatest health threat of the 21st century
- Address **root causes of ill health** by embedding health in key policies across sectors
- Advance the **Primary Health Care approach & essential health system capacities** for equity & gender equality
- Improve equity in **service coverage & financial protection**
- **Prevent, mitigate & prepare** for health risks from all hazards
- Rapidly **detect & sustain an effective response** to all health emergencies

PATHWAYS OF CHANGE & KEY CONTRIBUTIONS TO ACHIEVE THE STRATEGIC OBJECTIVES

WHO CHANGE PATHWAYS



CRITICAL ACTIONS BY MEMBER STATES, PARTNERS & KEY CONSTITUENCIES

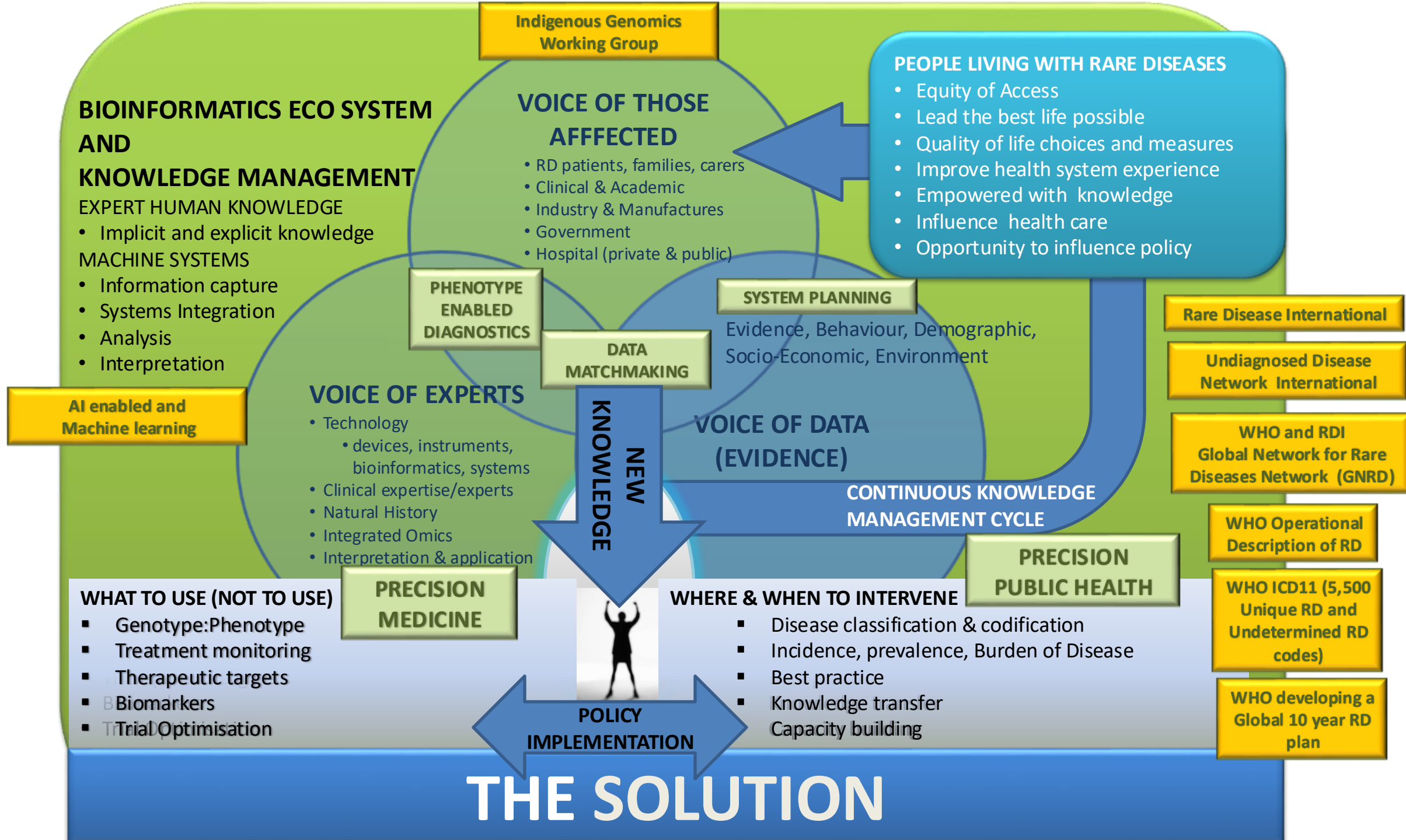
Commitment to health & well-being & internationally agreed targets reaffirmed

Priority health interventions reflected in **country, regional & global plans, strategies M&E frameworks**

Domestic & partner resources increased & aligned domestic with country health priorities

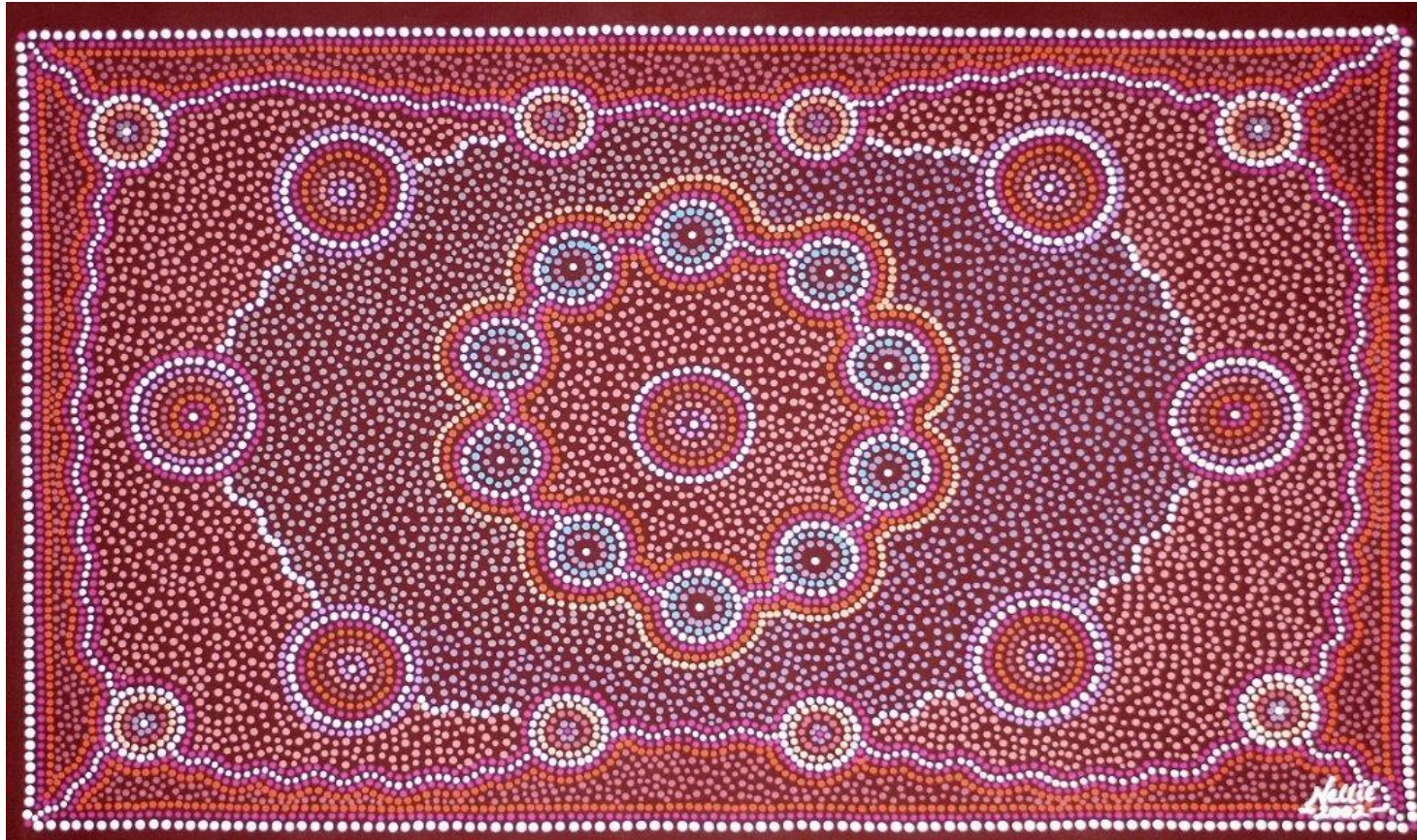
Inter-sectorial partner & community engagement for health & well-being is expanded

* Primary Health Care (PHC) approach



Transcendence

Jonelle (Nellie) Green [2002] a Badimaya woman from the Yamatji people of the Central Wheatbelt (Morawa) area in WA.



“**Transcendence** captures all the ways we transcend those things that can drag us down. Instead we link up and stay connected to those important things that are all interconnected – like a blanket of spirit from our country and ancestors that wraps us up and keeps us safe. The image represents the strong circle of connectedness that exists in Aboriginal communities. It represents the collaboration that is necessary between our communities and other stakeholders if there is to be sustained change in the health (indicators) of Aboriginal people in Western Australia”. **From WA Aboriginal Health and Wellbeing Framework 2015- 2030**