Taking phenotype to the population

The ultimate challenge

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Director, Generation Victoria

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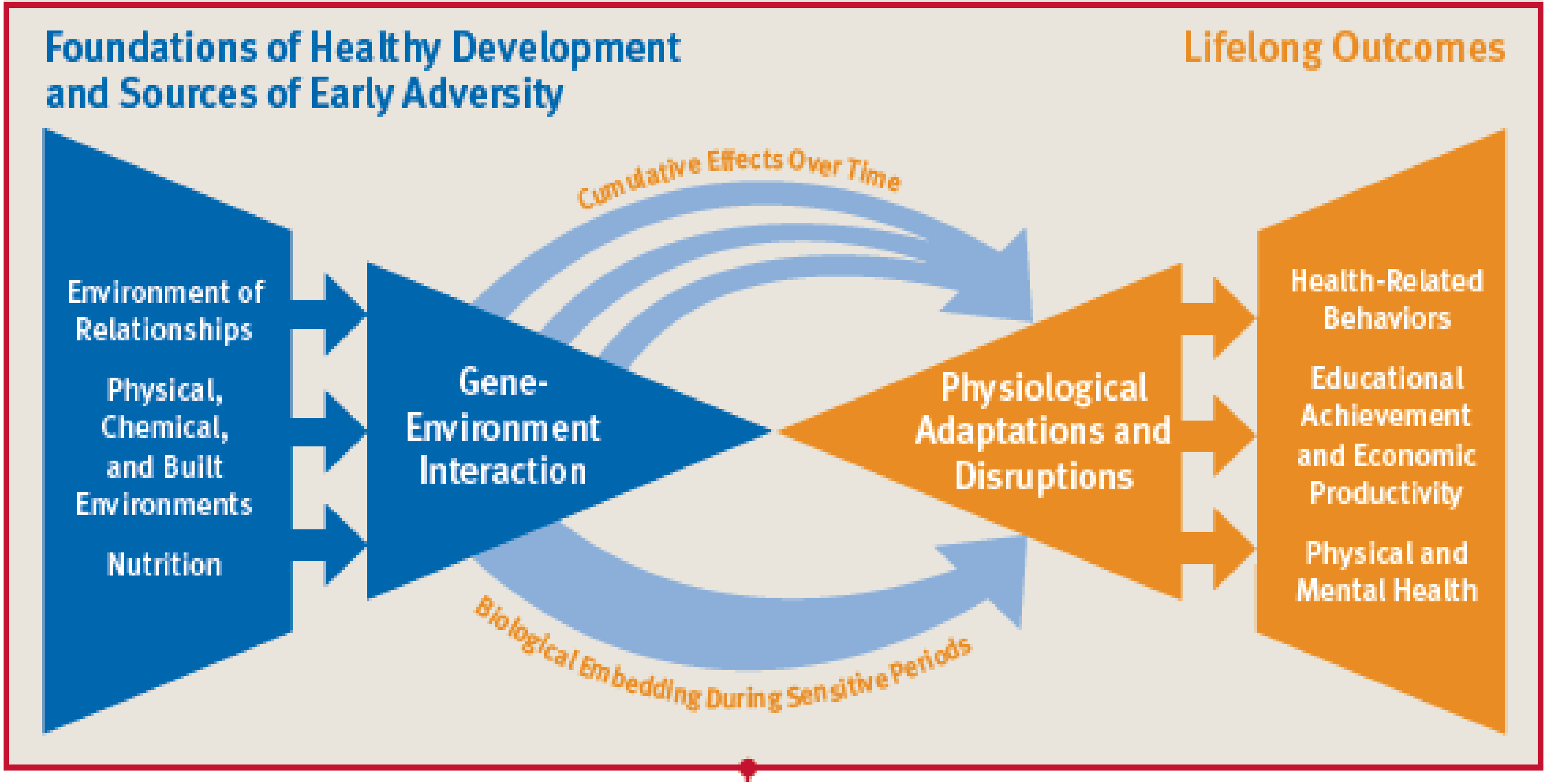
Melbourne Children's
A world leader in child and adolescent health

Longitudinal Study of Australian Children

- Growing Up in Australia
- Growing Up in Scotland
- Growing Up in New Zealand
- Growing Up in Ireland
- Millenium Cohort Study

HOW EARLY EXPERIENCES GET INTO THE BODY: A Biodevelopmental Framework



Shonkoff et al, Center on the Developing Child, 2012

The Child Health CheckPoint

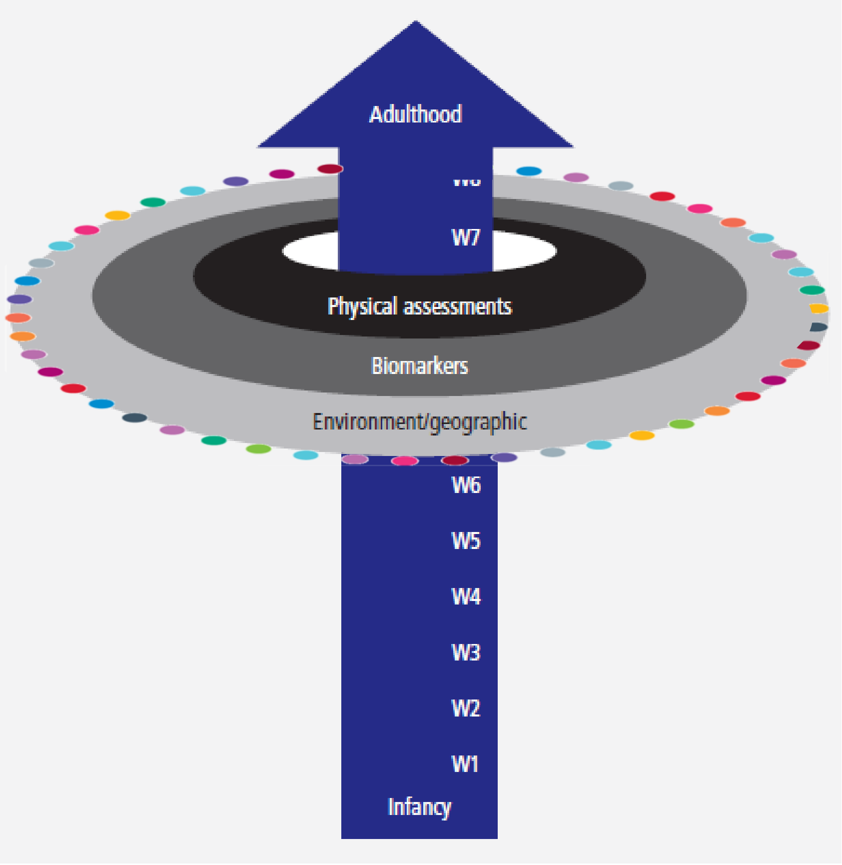


Figure 1: Conceptualisation of the integration of the Child Health CheckPoint into LSAC

The Child Health CheckPoint

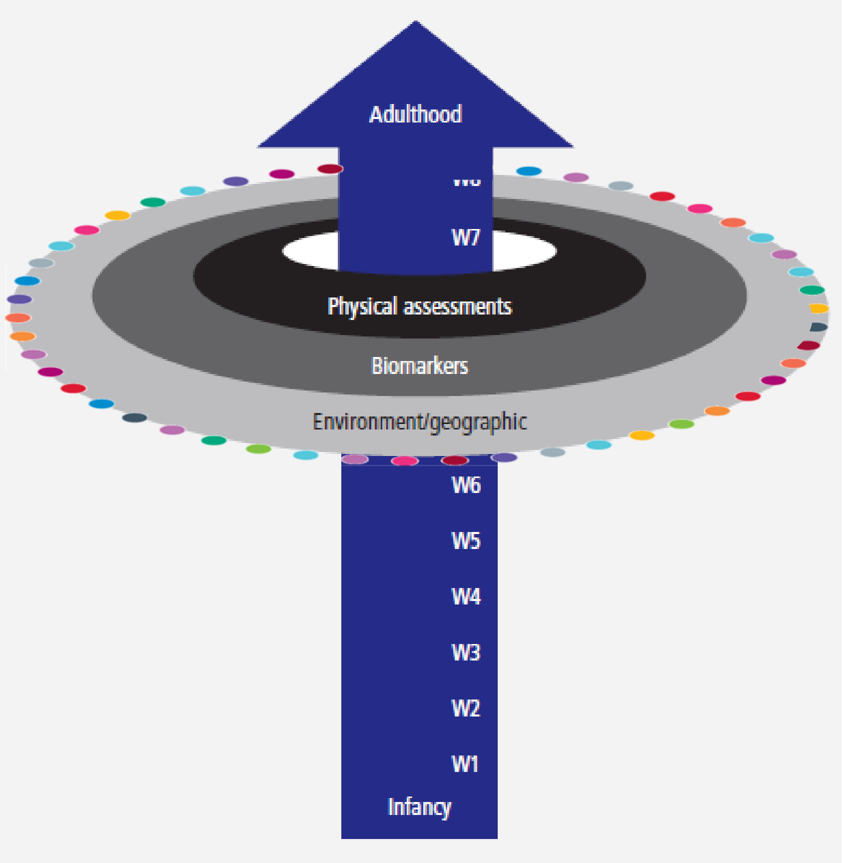


Figure 1: Conceptualisation of the integration of the Child Health CheckPoint into LSAC

The Child Health CheckPoint

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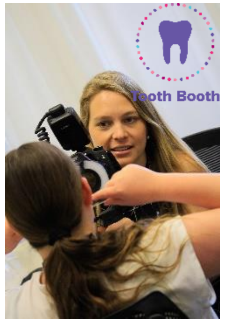

Finish | Start

GROWING UP IN AUSTRALIA'S Child Health CheckPoint

CheckPoint Check-in

Parent Trap
Consent
Parent questionnaire

Life at 25
Child written story

Measure Up
Height
Weight
% body fat
Waist girth

Heart Lab
cIMT
Pulse wave analysis
Pulse wave velocity
Blood pressure

Lung Fun
Spirometry
Biospecimens (urine, toenails, hair, saliva)

Listen Up
Hearing threshold
Auditory processing
Language processing
Vocabulary

Tooth Booth
3D dental images
3D head image

Sit and Click
HRQol, Pain, Puberty, Diet, Positive wellbeing

Bone Zone
pQCT bone scan

Young Bloods
Venous blood
Finger prick

Food Stop
Food choice and consumption

Jumping Beans
Broad jump

Bike Hike
PWC170 fitness test

See Here
Retinal photography
Visual acuity

About Time
MARCA time-use diary

Endgames
Accelerometry

KEY:
Child attends station
Parent attends station

The Child Health CheckPoint

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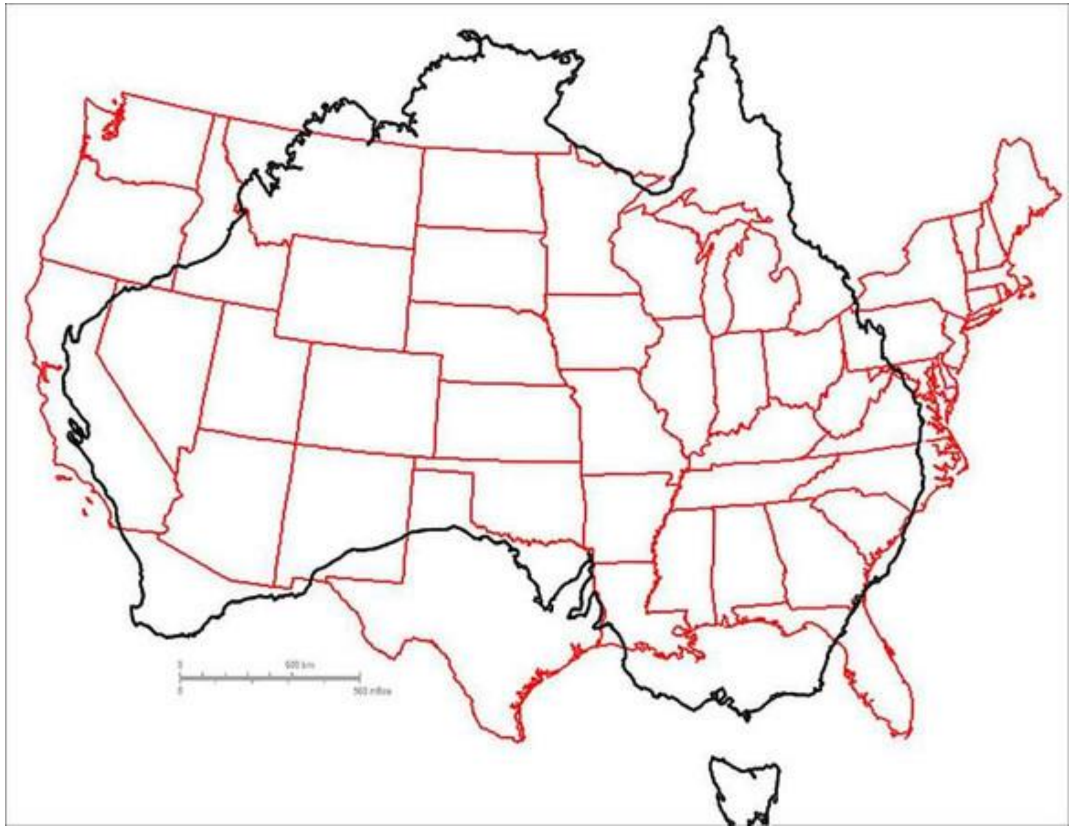

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G E N V

The Child Health CheckPoint

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Budget

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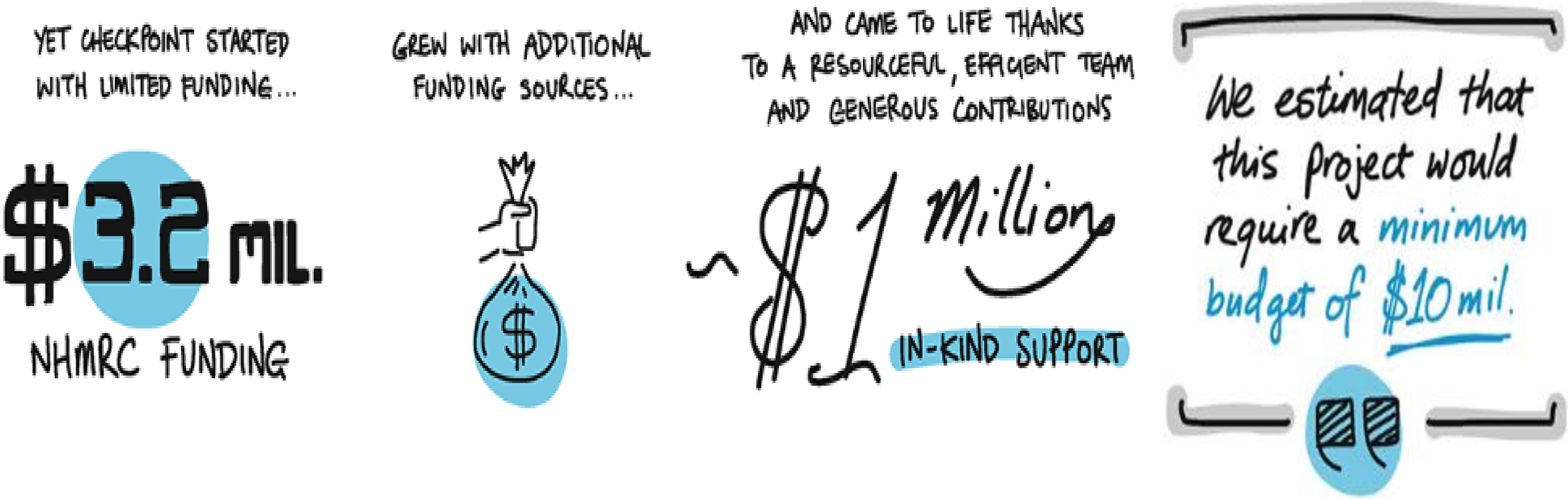


YET CHECKPOINT STARTED WITH LIMITED FUNDING...

$3.2 MIL.

NHMRC FUNDING

GREW WITH ADDITIONAL FUNDING SOURCES...

AND CAME TO LIFE THANKS TO A RESOURCEFUL, EFFICIENT TEAM AND GENEROUS CONTRIBUTIONS

~$1 Million

IN-KIND SUPPORT

We estimated that this project would require a minimum budget of $10 mil.

Child Health CheckPoint – we're celebrating!

2015-16

- 18 Australian cities
- 17,000 km in the CheckPoint trucks

Biostore

130,000 aliquots stored at the MCRI
A unique cross-generational bioresource

January 2019

Dataset released for international use
85 registered data users

GROWING UP IN AUSTRALIA'S
Child Health CheckPoint

A physical and biomarkers module for the Longitudinal Study of Australian Children

July 2019

- BMJ Open Special Issue
- 16 papers, 16 health domains

Results reached >8 million people

"…their **forward-thinking, broad and far reaching research** was why we dedicated our first Special Issue to a cohort study." BMJ Open Editor Adrian Aldcroft

2000 child-parent dyads

Data released: cardiovascular, respiratory, oral and kidney health, body composition, bone and muscle, facial morphology, physical activity, fitness, sleep, time use, vision, hearing, language, HRQL

Biosamples data

Data released: Metabolomics (Nightingale), Polygenic Risk Scores, Inflammation (GlycA), telomere length (ageing)

To be released: Metabolomics (Nightingale), CRP, micronutrients (lipid & water soluble vitamins, one-C), heavy metals + more

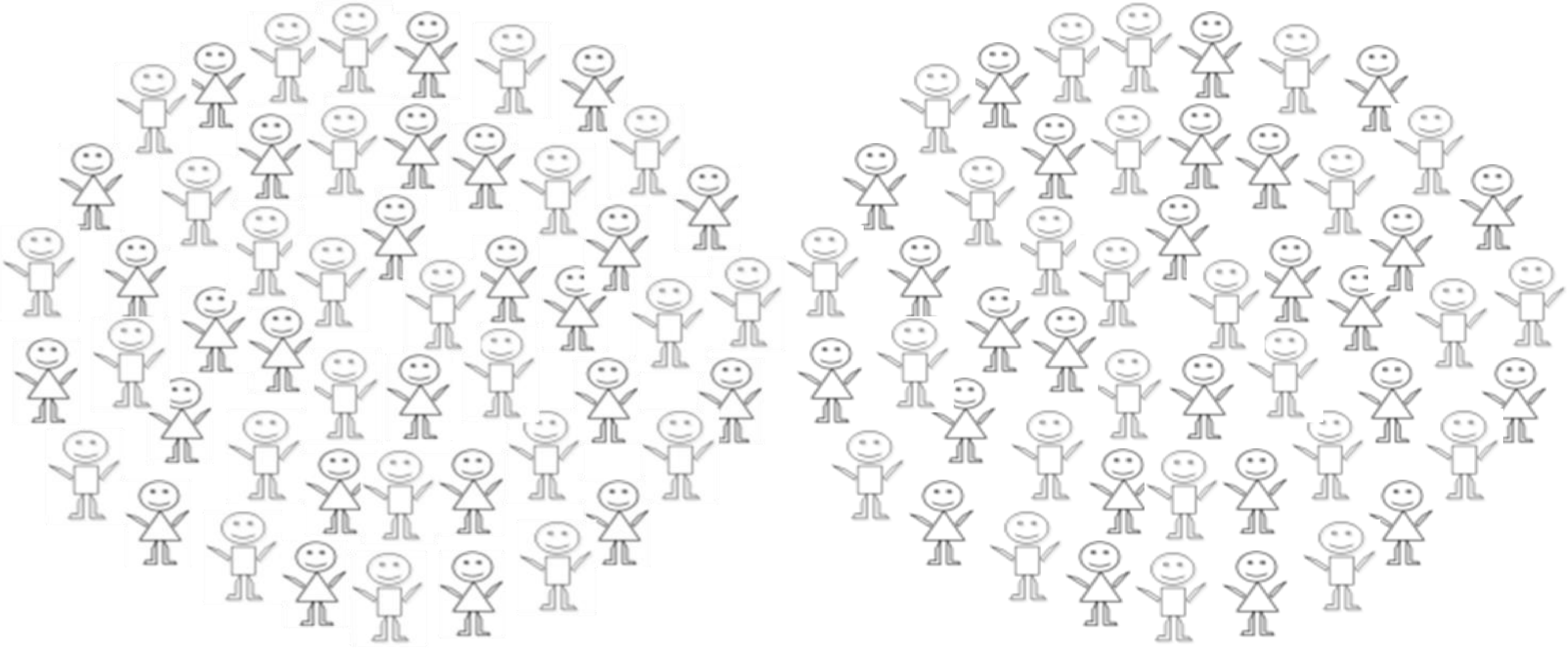
Learnings

Measuring phenotype:

- It’s possible
- It’s faster and cheaper than you think
- Children and parents enjoy it - more than questionnaires!
- They think it’s useful
- It takes lots of planning – but it doesn’t need experts to collect
- Researchers want it
- In order to have it, PIs are willing to commit to Open Science
- Unique, quality data begets funding

Generation Victoria (GenV)

How can we keep people, as individuals and as populations, healthy and developing well?



GenV

Targeting all 1700,000 Victoria babies born in 2021-22 and their parents

Consented cohort

Biosamples

Linked admin & services data

GenV collected data

GenV

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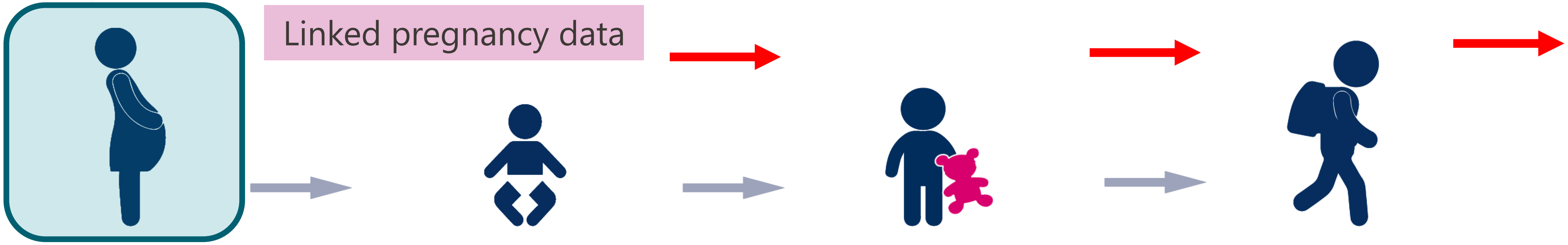


Linked pregnancy data

- Ultrasounds
- Maternal Serum Screening
- Birthing Outcomes System
- Non-invasive Prenatal Screening (NIPT)

- Newborn Screening
- Infant Hearing Screening
- Consent soon after birth
- Child and Parent DNA
- Cord blood bank

- Maternal & Child Health Visits
- Immunisation Register
- Kinder Childcare
- Brain Development

- School Entrant Health Q
- Australian Early Development Census (AEDC)
- NAPLAN
- School health checks

Health & Education service encounters: Administrative Data (e.g. VAED, VEMD, Medicare, My Health Record)

Clinical & Developmental Repository

Geographic Information System (GIS)

■ ■ ■ Existing but joined-up collections ■ GenV Specific

By 2035, GenV's vision is to have solved complex issues facing our children and the adults they become

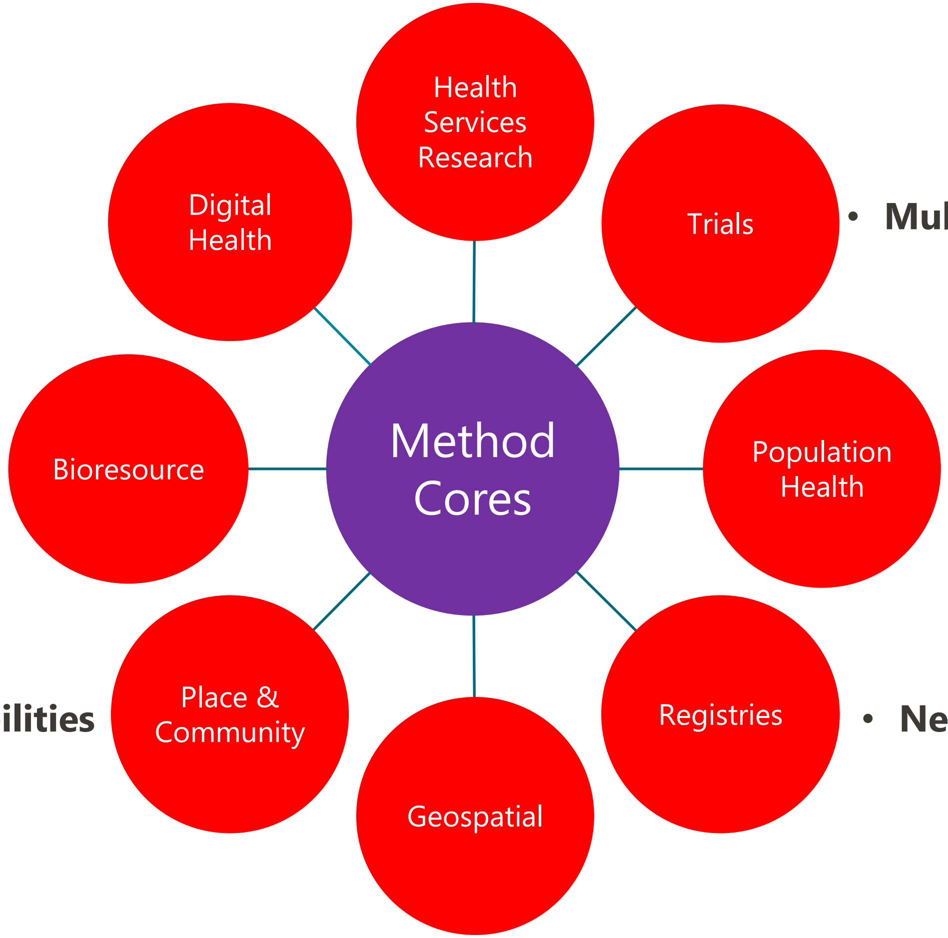
GenV will give us a complete picture of the health and wellbeing of a generation of Victoria's children and their parents.

Helping us to discover new ways to predict, prevent and treat many of today's most challenging conditions.

So that together we can make this our healthiest generation ever.

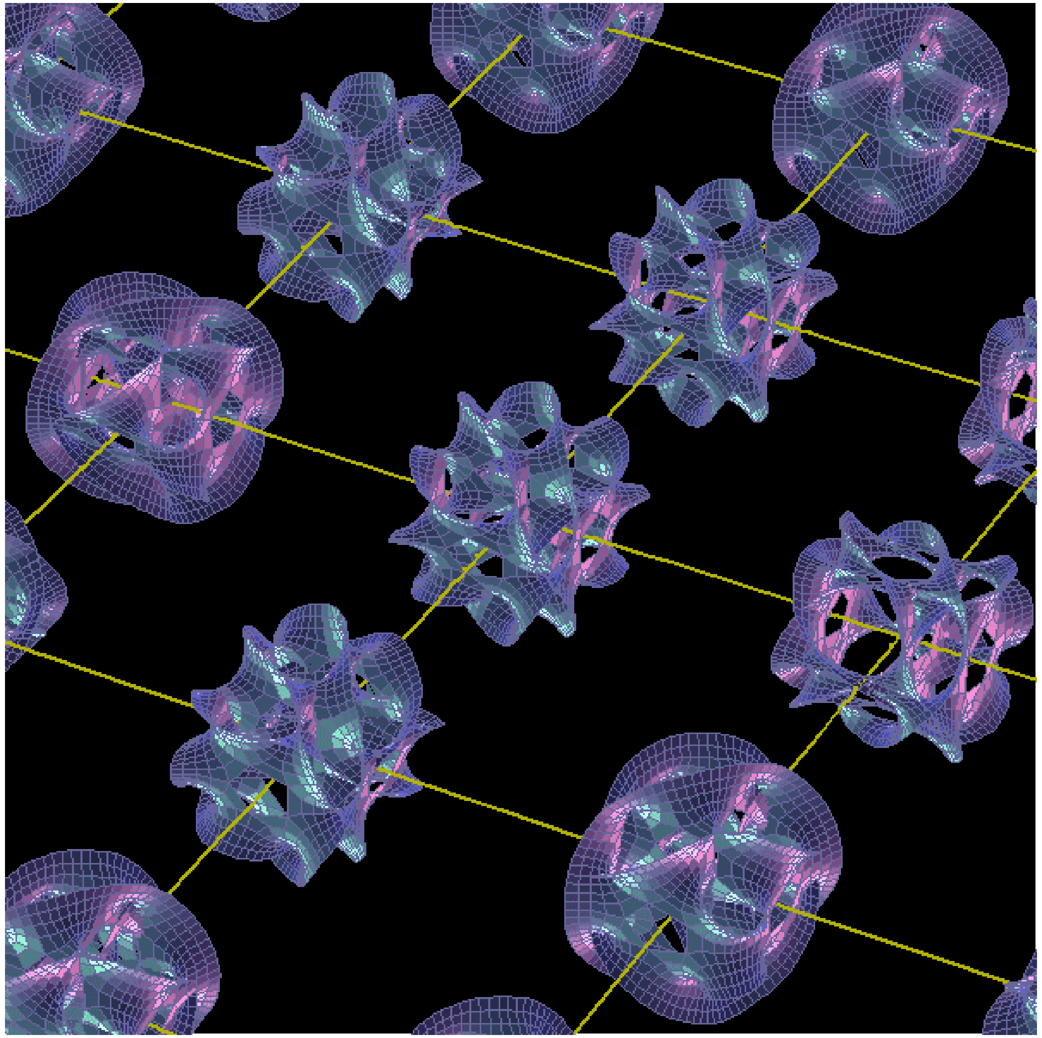
But what is ‘a complete picture’….?

…and what does it mean to be solution-focused?



- **Variations in care, costs and outcomes**
- **ePhenome**
- **Multiple RCTs overarching protocol**
- **Curate existing biosamples**
- **Simulations/causal modelling**
- **Local rich datasets & capabilities**
- **New registries – high-risk pregnancy, SCN**
- **The BELL – Built Environment LifeCourse Laboratory**

Taking precision health to the population



- Fit our research to their lives, not their lives to our research

- All layers for everyone – cut and slice creatively
- Microbiome
- Genomics
- Biology
- Phenotype
- Time
- Generations
- Lifecourse
- Services
- Population metrics
- Geospatial

- Deep phenotyping – low burden, digital reach

- The rawest data possible now (images, audio, traces) – computation later

The GenV Guiding Principles

1

Collaboration

GenV should **increase partnerships** and links between researchers, clinicians, policy makers, service providers and the community

2

Inclusivity

GenV should be **inclusive** of all partners, participants and data users

3

Sustainability

GenV should be **financially sustainable, leverage infrastructure and capabilities**, and be **scalable** to respond to emerging insights and needs

4

Enhancement

GenV should be **low burden to its participants** and **enhance services**

5

Systematised processes

GenV should operate via **standardised, high throughput processes** that can be embedded to become 'normal practice' in real time

6

Value

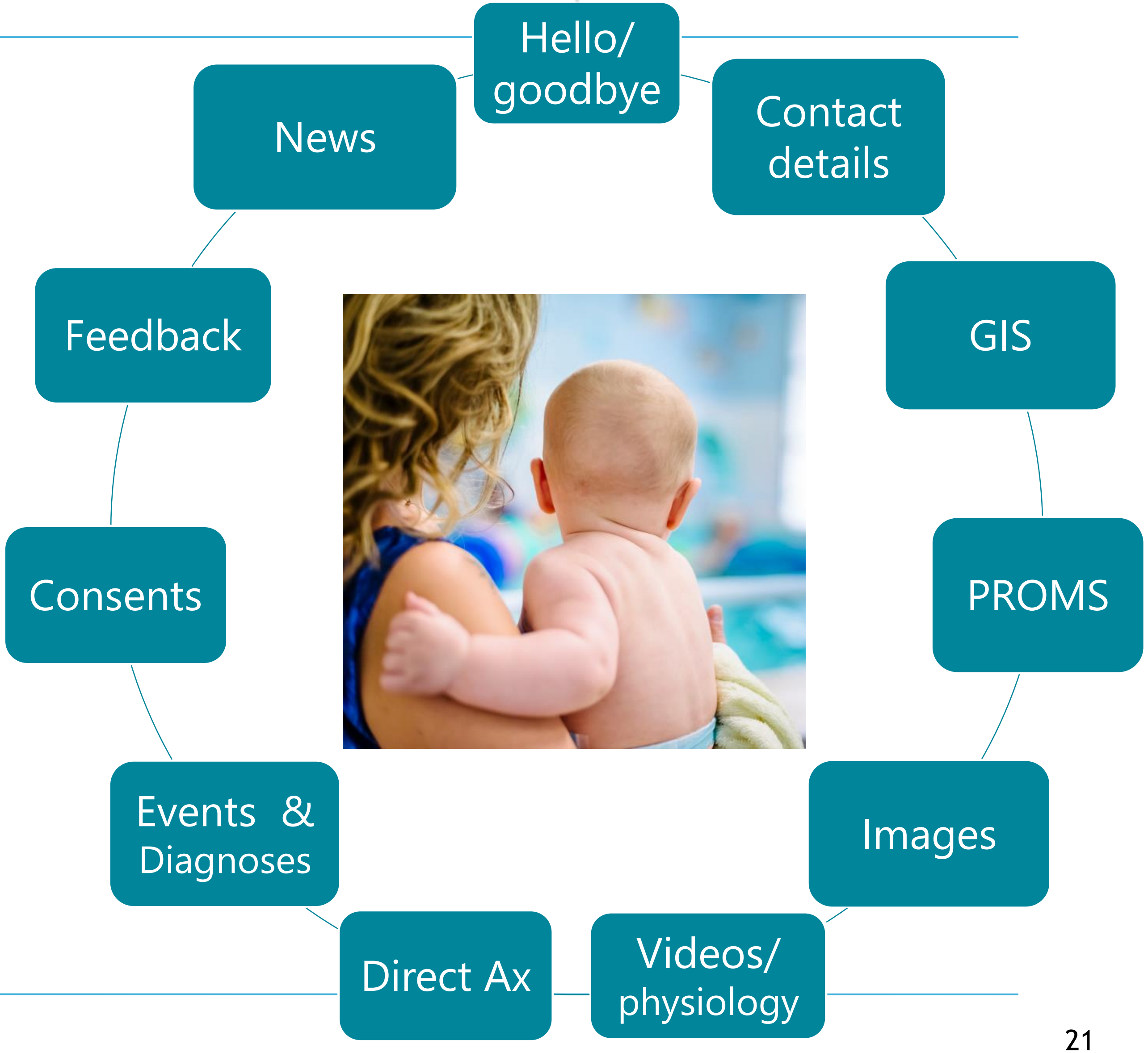
GenV should generate outstanding science and solutions that maximise children's outcomes

e-Phenome: Outcomes measurement **at scale**

From ‘black box’ to ‘open window’:

- Genomes need phenomes
- AI prediction needs outcomes
- Trials need endpoints

….digital maps of the developing child and maturing adult



Thank You

GenV Directors (Melissa Wake, Sharon Goldfeld, Richard Saffery)
GenV Program Manager (Dino Asproloupos)
GenV Stream Leads (Jessika Hu, Michael Stringer, Will Siero, Joan Leong)
GenV Investigator Committee
GenV's many collaborators
Paul Ramsay Foundation
Victorian government
NHMRC
MCRI

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Trials