To infinity and beyond: Partnerships utilising hearing screening program infrastructure

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Directors, VIHSP
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Context

ANHS 2017 - Partnerships with clinical and research initiatives related to hearing loss:

• Mild-moderate congenital hearing loss: secular trends in outcomes across four systems of detection (Peter Carew, Concurrent Session 1: Audiology)

• Paediatric hearing services and research in Victoria: working towards predicting prognosis in the era of genomic medicine (Valerie Sung, Concurrent Session 5: Aetiology/Medical)

• Whole Exome Sequencing in infants with congenital hearing loss (Lilian Downie, Concurrent Session 5: Aetiology/Medical)
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Partnerships with research that goes beyond hearing loss
VIHSP screening protocol

VIHSP screen #1
- Pass result in both ears
  - NFA
- Refer result in 1 or 2 ears
  - VIHSP screen #2
    - Pass result in both ears
      - NFA
    - Refer result in 1 or 2 ears
      - Referral to audiology by VIHSP Area Manager
VIHSP: Key performance indicators

**Screen:** Percentage of newborns screened by 1 month of age; **target >97%**

**Support:** Percentage of newborns referred for early support who are processed within three business days; **target >90%**

**Diagnosis:** Percentage of newborns with a positive screen who commence diagnosis and assessment at an audiology centre by 3 months corrected age; **target >90%**

Reported monthly
VIHSP staffing structure
VIHSP screening process

Face-to-face interaction
almost all parents of newborns
staff from the one organisation

No other service does this
(including other universal services like metabolic screening and MCH)
VIHSP databases

VIHSP operates one of few **statewide databases** that

- contains almost all birth notifications, as close as possible to real-time
- includes newborn data with parent data; and
- records family demographic information and birth location information.

Regulatory:

- Policies and procedures of the Royal Children’s Hospital
- Legislation: Freedom of Information Act 1982 (Vic); Freedom of Information Regulations 1998 (Vic); Health Records Act 2001 (Vic); Health Services Act 1988 (Vic); Privacy and Data Protection Act 2014 (Vic); Public Records Act 1973; Mental Health Act 2014; Child Wellbeing and Safety Act 2005; Children and Young Person’s Act 1989; Privacy and Data Protection Act 2014
VIHSP databases

<table>
<thead>
<tr>
<th>Attribute</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Relational</td>
<td>linkage of data across modules</td>
</tr>
<tr>
<td>Completeness</td>
<td>processes for reconciliation</td>
</tr>
<tr>
<td>Timeliness</td>
<td>data uploaded and updated daily</td>
</tr>
<tr>
<td>Customisable</td>
<td>ongoing “tweaking” for improved functionality relating to service delivery and reporting</td>
</tr>
</tbody>
</table>
VIHSP – the one stop shop

“One stop shop” for FTF access with almost all families with newborns in Victoria

VIHSP Process

Research that would have been logistically difficult or impossible is now possible.
VIHSP – the one stop shop

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VIHSP Process

VIHSP databases

“One stop shop” for rich, accurate and real-time access to data relating almost all Victorian newborns
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Research that would have been logistically difficult or impossible is now possible
Melbourne Children’s Campus Partners
Ethical considerations

• NHMRC statement 2007 (May 2015 update)
  – All research projects comply
  – Record linkage and data sharing explicitly addressed

• All have Human Research Ethics and Governance approval
Gen V ("The Big Idea")
Murdoch Childrens Research Institute

The current situation
Gen V: Addresses four key issues

• Unprecedented rates of adult diseases
• Reduce the burden of modern epidemics for children
• Change how large scale research happens (too cumbersome, short-term and costly)
• Use under-utilised data infrastructure in an innovative way

Reap the full benefit of Victoria’s investment into its outstanding health and educational services
Gen V: Vision

To create the world’s most exciting children’s health, development and wellbeing project to answer today’s pressing policy and practice questions

- Embed research into health and education services
- Cradle-to-grave approach: the first 1000 days
- Partnerships between researchers, practitioners and policy-makers
- Innovation in research and data

www.mcri.edu.au
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International research hub

Benefit both children now and the adults they become

www.mcri.edu.au
Gen V: vision

• Cradle-to-grave approach: the first 1000 days
Gen V: Methodology

- Ultrasounds
- Maternal Serum Screening
- Birthing Outcomes System

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

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

- School Entrant Health Q
  - Australian Early Development Census (AEDC)
  - NAPLAN
  - Victorian Student H&WB Survey
  - School health checks

Health & Education service encounters:
- Administrative Data (e.g. VAED, VEMD, Medicare)
- Clinical & Developmental Repository

Geographic Information System (GIS)

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Geographic Information System (GIS)

Existing but joined-up collections
Gen V Specific

www.mcri.edu.au
Gen V: When the wheel has already been created

• Goal: 100,000 babies recruited in 2 year period
• Recruitment at birth
• Without VIHSP
  – Birth notifications 70+ hospitals
  – Staff at 70+ locations
  – Data systems for birth details
  – Expensive and time consuming
• With VIHSP
  – “Piggy-back” onto hearing screen for approach to families
  – Sharing of birth details, parent demographic data
  – Time and cost savings++
VIHSP’s contribution to Gen V

• Piloting of methodology for recruitment
• Sharing of birth details information
• Sharing of parent demographic information
• Sharing of newborn hearing screen and diagnostic audiology results

• Face to face service delivery
• Databases
• In-kind support (recruitment, data sharing)
• One-stop shop
Perinatal Record Linkage (PeRL)
Briohny Hutchinson, Lisa Hui, Jane Halliday - Murdoch Childrens Research Institute

Noninvasive Prenatal Testing (NIPT)
• a screening test for Down syndrome, trisomy 18, trisomy 13
• utilizes maternal plasma
• can be performed at any stage of pregnancy from 10 weeks
• fewer risks than CVS and amniocentesis
• more accurate for Trisomy 21 than first trimester screening with nuchal translucency and maternal serum biochemical markers
PeRL: Study design

• Record linkage study
• Datasets of
  – maternal serum screening and prenatal diagnostic tests for chromosome abnormalities
  – chromosome results related to live births and pregnancy losses.
• The challenge: NIPT: pre-natal, mother’s details
  Chromosomal abnormalities: after birth, infant’s details

Pairing of NIPT results (mother) with chromosomal results (infant) is integral to this study, but is difficult to do in many cases.
VIHSP’s contribution to PeRL

- Record matching: linkage of parent details with baby’s details
- With VIHSP: more records matched, improve data completeness and study power
- Without VIHSP: incomplete data, unlinked records, less study power

- Databases
- In-kind support (data extraction)
- One-stop shop
Melbourne Genomics Health Alliance: Congenital Hearing Loss (CHL)
Clinical Lead: David Amor - University of Melbourne

Genetic diagnosis for hearing loss
• No statewide approach
• Ad hoc referrals
• Incomplete coverage
• Involved any or all of:
  – Clinical assessment
  – Imaging (CT or MRI)
  – Genetic testing (Connexin 26 only)
Melbourne Genomics: CHL flagship aims

AIMS
- Define the genetic aetiology of sensorineural congenital hearing loss
- Streamline the care of children with hearing loss
- Explore parents’ interest in genomic testing of their children in relation to
  - hearing loss
  - childhood onset medically actionable conditions
  - childhood onset conditions with some uncertainty (treatment, penetrance)
- Explore the use of genomic testing as a population tool
- Gain information on whole exome sequencing as a clinical test
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Timely access to infants with congenital sensorineural hearing loss
Melbourne Genomics: CHL – recruitment & data collection

Recruitment & data collection process

1. VIHSP
   - Identifies babies with moderate or greater severity hearing loss and sends opt out and information letter

2. RCH Genetic Counsellor (GC)
   - Contacts patient using script to explain Melbourne Genomics, VicCHILD and CHIC clinic and other services; organises appointment

3. Medical assessment
   - CHIC clinic + Geneticist/Fellow/GC

4. VicCHILD
   - Data collection
Melbourne Genomics: CHL – recruitment & data collection

Without VIHSP
• Liaison with multiple diagnostic centres, AH centres and EI providers to identify potential candidates
• Difficult to get population coverage
• Difficult to influence timing and processes of inviting into study

With VIHSP
• One organisation
• Population coverage – no selection bias
• Structured recruitment processes and timing
VIHSP’s contribution to Melbourne Genomics: CHL

- Databases
- In-kind support (opt-out letters, demographic and diagnostic data)
- One-stop shop

- Hearing loss (1:1000)
  - Genetic (50%)
  - Non-Genetic (25%)
  - Idiopathic (25%)

  - Syndromic (30%)
  - Non-Syndromic (70%)

  - Autosomal Recessive (75-85%)
  - Autosomal Dominant (15-24%)
  - X-Linked (1-2%)
  - Mitochondrial (<1%)

  - DFNB1 (50%)
  - Other (50%)
Future projects

**cCMV** (Valerie Sung)
- VIHSP contribution: timely notification of infants who return a positive (refer) result on NHS

**Family violence** (Department of Human Services, Victorian Government)
- VIHSP contribution: modelling of service delivery for delivery of a post-natal DV screen

**Victorian Clinical Genetics Services**
- VIHSP contribution: exploring sharing of data to minimise LTFU
Future projects

Digitising Health

Digitising Health, Victorian Department of Health and Human Services (2016)

dhhs.vic.gov.au/publications
Future projects

Digitising Health

Digitising Health, Victorian Department of Health and Human Services (2016)
dhhs.vic.gov.au/publications
Conclusions

• NHS programs - systems, procedures and data sets - invaluable to enabling and progressing research
• In Victoria, several research projects are underway that would otherwise be non-existant, more limited in scope, or would take longer if it were not for the contributions of VIHSP
• Reciprocal benefits – VIHSP contributes more broadly to well-being, research progresses
• Look for partnerships with researcher counterparts: the sum of the parts is greater than the whole
• Commitment to improving child health: sharing & partnerships is part of this
Thank you to the teams at

- Gen V
- PeRL
- Melbourne Genomics Health Alliance: Congenital Hearing Loss Flagship
- VicCHILD
- VIHSP
- DHHS

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The Centre for Community Child Health is a department of The Royal Children’s Hospital and a research group of Murdoch Childrens Research Institute.