Newborn Screening in Victoria

Prof Agnes Bankier
In the beginning
Translating new knowledge to clinical practice
David Pitt and David Danks

- 1933 Dr Folling discovered PKU
- 1959 Dr David Pitt diagnosed a 14 yr old girl
- 1961 Wet-nappy test Infant Welfare Centres
- 1964 Guthrie test developed as a give-away kit
- 1966 Screening at the Children Kew Cottages → Mont Park, Ivan Frances “public laboratory”
- 1968 - 70% of Victorian babies tested
- 1970 – Vic NBS David Danks Lab moved to BDRI (MCRI) “private organisation”
Newborn Screening in Victoria

- Phenylketonuria (PKU) since 1966
  - 1:12000 ie 7 babies/year

- Congenital hypothyroidism since 1977
  - 1:3,500 ie 35 babies per year

- Cystic fibrosis since 1989
  - 1:2,500 ie 24 babies per year

- TMS in Feb 2002: >many metabolic disorders.
  - 11 babies per year
The Expanded Newborn Screening Program 2002

- TMS detects **biochemical markers** in the blood spot
- This expanded program will detect about 9 babies per year in Victoria with conditions such as:
  - MCAD deficiency
  - homocystinuria
  - maple syrup urine disease
- **Early treatment can prevent death or disability**
  - “treating babies before they get sick”
- Genetics was not mentioned
Population Screening
World Health Organisation guidelines

• Condition of known cause
• Frequent condition and significant problem
• Safe reliable test available (+/-)
• Known treatment intervention

• Cost benefit established
• Counseling services available
Variance in practice between the States in 2002

- **Vic** – cards kept indefinitely since late 1960s
- **NSW**- cards kept for 18 years
- **SA**- cards kept indefinitely
- **QLD** – cards kept for 25 years
- **WA**- cards kept for 2 years only
  - 5 year collection destroyed
- **NZ** - cards kept indefinitely
Concerns: Potential Misuse

• Cards seized by the Police and used for law enforcement purposes – WA 1997 - cards destroyed

• Inadvertent DNA bank - potential database
• Cards potential sale for commercial gain to eg a drug company
• Unlawful research
• Cards potential use for secondary purposes eg paternity testing, insurance assessments
• Vic Program in a “private” setting
How your DNA is falling into private hands

If you are under 34 and were born in Victoria, a private company has your DNA on file — and people are worried. Tom Noble reports.

A child’s birth is a special moment — the labour, the joy, the chaos. And about 48 hours into a new life, each baby has a medical test that has saved countless children’s lives.

Blood from the baby’s heel is soaked into an absorbent card and then tested for a range of rare, usually treatable conditions. Most parents hear nothing more and life goes on.

But what becomes of the card? In Victoria, they are tested and stored by Genetic Health Services Victoria, a company set up 10 years ago. It is paid by the State Government to do the tests, yet believes it owns the cards, which date from 1965. All Victorian babies were tested from about 1970. The Parkville company ultimately controls who can get access to the blood and DNA, of more than 2 million people born in Victoria. Its collection is the largest in Australia and the only one not in government hands.

“I don’t believe it is appropriate for the state to grant ownership of its citizens’ DNA to a private company,” Health Services Commissioner Beth Wilson said.

“I think custodianship of samples such as the newborn screening cards should be a state responsibility and I believe most members of the public would agree with this.”

Liberty Victoria vice-president Brian Walters, SC, said he was deeply troubled that a private company was being given DNA from a publicly-funded screening program.

“Where it’s a state activity there’s better recourse to protection of the information,” he said. “Assurances of good faith are all very well — and they may be very nice people — but companies change hands; people see opportunities and things change.”

Opposition health spokesman David Davis said a joint parliamentary committee should investigate the ownership of the cards and the ethical-legal aspects of who uses the cards and when.

“This is a bipartisan issue. The only protection for Victorians is transparency and light. There’s no other way,” Mr Davis said.

Mr Walters said concerns needed to be heeded to stop otherwise parents could decline a universally supported test.

“Who has control over the information and what purposes it can be used for will determine whether people give consent and have trust in the system,” he said.

In recent years, the cards have been used for medical research approved by an ethics committee at the Royal Children’s Hospital.

People may be concerned about Guthrie cards being released for use in “research”, especially by researchers in private agencies such as pharmaceutical companies. But if blood spots are released for research, they are almost always “de-identified” — the scientists who receive them do not know whose samples they are and the “donors” cannot be traced.

Loane Skene, professor of law, University of Melbourne

Your DNA is in safe hands

Re your report “How your DNA is falling into private hands” (The Age, 5/7): “Private” health care agencies are subject to the same federal and state privacy legislation as public sector agencies. They are required not to collect or hold “health information” without the consent of the people concerned (such as parents of a newborn child); and they must take steps to hold the information securely and protect its confidentiality.

A private sector agency could sell its business, including its “assets”, to another agency; but so could a public sector agency and, in the current climate of privatisation, that is not infrequent. But purchasers of a business that holds confidential information are bound by an equitable duty of confidence in relation to that information and must keep it confidential.

Melbourne Children’s Excellence in clinical care, research and education
Retention of Newborn Screening Cards in Victoria - 2002

- Cards are kept indefinitely: NPAAC 50->25 years
- Over 2 Million cards stored since late 1960s
- Sealed boxes filed by sequential lab number by year of birth. Name of baby not recorded on the card
- Cards were on open shelves at the end of the VCGS diagnostic laboratory
- There was an information brochure for parents but no formal consent process
NBS Issues

2003- 2011

• **Consent** for collection, testing, retention and use
  - Brochure and no formal consent
  - Opt-out consent → pilot written consent 2010 → written consent 2011
    (No screening – 0.16%, No to research – 6.47%)
  - NBS Advisory Committee – policies, access, consent procedures

• **Storage**
  - NBS Cards moved from VCGS to secure off-site facility, Data base in the laboratory

• **Ownership**
  - Cards deemed public record, “owned” by Victorian Department of Human Services, under custodianship of VCGS
  - Transfer of the card to parents on request - “Disposal”, “Return” - in writing, disclaimer signed by both biological parents

• **Access**
  - Strict access protocols for **clinical use & research**
## Guthrie Card – Newborn Screening Card

### VICeTORIAN NEWBORN SCREENING LABORATORY

**Hospital Name**
and ward

COMPLETE ALL DETAILS OR USE HOSPITAL LABEL BELOW

**Baby's FULL NAME**

**Mother's FULL NAME**

**UR**

**Doctor's Name**

**Date of birth** / / time 24:00hr

**Date of sample** / / time 24:00hr

**Gestation:** weeks  Current weight:  g Twin 1/2

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<thead>
<tr>
<th>Breast Feed</th>
<th>Formula Type</th>
<th>TPN Male Female</th>
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**Relevant Clinical / Family History**

**Collector's Name**

**Newborn Screening Consent**
I have received and understood the information in the newborn screening brochure. I consent to my baby having blood collected for the newborn screening test.

Yes  No

**Secondary Research Use**
I understand that blood from stored screening cards can be used occasionally for de-identified health research. I choose to make my baby's blood sample available for this purpose.

Yes  No

**Parent Signature:**

### Hospital Name and ward

**USE BLOCK LETTERS OR HOSPITAL ID LABEL**

**UR/Comments**

**Doctor's name and initials**

**Infant's full name**

Twin 1/2

**Date of birth** / / time 24:00hr

**Date of sample** / / time 24:00hr

**Gestation:** weeks  **Current weight:**  g

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**Relevant Family History**

**Collectors Name**
National Framework for Newborn Screening for Australia

- National approach to Genetic Services, HGSA 1995
  → Public Health Genetics
- ALRC Proposal “AHMAC (Australian Health Ministers Advisory Council) …need to develop nationally consistent policies and practices in relation to collection, storage, use and access….. Guthrie cards…collected. 2003 -→ Human Genetic Commission of Australia
- Public Health Partnership -→ AHMAC -→ Population based screening framework 2008
- 2016 National Framework for Newborn Screening
# The Newborn screening team over the years

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<th>Clinical</th>
<th>Laboratory</th>
<th>Dietetic</th>
<th>Counselling/nursing/admin</th>
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<tbody>
<tr>
<td>David Pitt</td>
<td>Jan Brasch</td>
<td>Betty Lynch</td>
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<td>David Danks</td>
<td>Ivan Francis</td>
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<td>John Rogers</td>
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**Notes:**
- The Newborn screening team has been active over the years with contributions from various individuals. The table lists the team members associated with clinical, laboratory, dietetic, and counselling/nursing/administrative roles. The team has been supported by numerous professionals dedicated to newborn screening initiatives.