3 March, 2019

Australian Health Genetics/Genomics Survey 2017

Project Team

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AHMAC directed the Commonwealth to lead, in partnership with states and territories, the development of a **National Health Genomics Policy Framework** to improve coordination and consistency in approach to **integrating genomics in health care**

AHMAC commissioned a **national stocktake** of genetic/genomic testing and activity

The Framework was endorsed by the COAG Health Council
The Framework identifies five Strategic Priorities:

1. Patient-centred approach
2. Workforce
3. Sustainable financing
4. Safety, quality and clinical utility of services
5. Responsible collection, storage and use of data
The Stocktake

Previous surveys of genetic testing in Australia were performed by the RCPA in 2006 and 2011.

Aims of the 2017 Stocktake:

• To provide updated information on the nature, availability & volume of genetic/genomic tests arranged for Australian patients
• To make comparisons with historical data
• To provide information about workforce change requirements
• To facilitate modelling for future service provision
AHMAC directed the Commonwealth to lead, in partnership with states and territories, the development of a **National Health Genomics Policy Framework** to improve coordination and consistency in approach to **integrating genomics in health care**

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The Framework was endorsed by the COAG Health Council
Background

Stage 1:
The Commonwealth engaged the RCPA to undertake preliminary scoping to support the stocktake.

Stage 2: Implementation
- Survey open (8 weeks) Feb – Apr
- Data cleansing, aggregation & analysis May – Sep
- Report submitted

Pilot survey Jan/Feb

2016 2017 2018
Survey Instrument

1 July 2016 to 30 June 2017

Covering letter
Guide for participants
Confidentiality agreement

Laboratory questions

- Type of laboratory
- NATA accreditation
- Staffing
- Referral pathways
- Laboratory Information Management System (LIMS)
- Data storage and sharing
- Tests referred offshore

Test questions

- Nature and volume of tests performed
- Test targets and methodology
- Clinical indications
- Turnaround times
- Funding
- Geographical origin of samples

3 March, 2019

The Royal College of Pathologists of Australasia
Participation

Eligibility - Laboratories known to have offered human genetic and genomic tests that yielded results with medical utility during the 2016/17 financial year

87 Laboratories identified & invited to participate

83 Submitted data (95% participation)

• 6 did not provide test information
• 8 did not provide details about accreditation status, clinical referrers, staffing and supporting infrastructure

4 No response
### Industry Sector: Biochemical Genetic Tests

Laboratories offering biochemical genetic tests (analytes/enzymology/maternal serum screening/newborn screening):

<table>
<thead>
<tr>
<th>Industry sector</th>
<th>No. of labs.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Public</td>
<td>5</td>
</tr>
<tr>
<td>Private</td>
<td>5</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>10</strong></td>
</tr>
</tbody>
</table>
Industry Sector: Constitutional/ Somatic Tests

Laboratories offering genetic/ genomic tests (constitutional/ cancer):

<table>
<thead>
<tr>
<th>Industry sector</th>
<th>No. of labs.</th>
<th>Percentage of constitutional tests</th>
<th>% of laboratories delivering % of tests</th>
</tr>
</thead>
<tbody>
<tr>
<td>Public</td>
<td>38</td>
<td>52%</td>
<td>34%</td>
</tr>
<tr>
<td>Private</td>
<td>20</td>
<td>27%</td>
<td>63%</td>
</tr>
<tr>
<td>Catholic/ Schedule 3</td>
<td>3</td>
<td>4%</td>
<td>2%</td>
</tr>
<tr>
<td>Research</td>
<td>12</td>
<td>16%</td>
<td>0.4%</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>73</strong></td>
<td><strong>52%</strong></td>
<td><strong>34%</strong></td>
</tr>
</tbody>
</table>

Percentage of constitutional and cancer tests across sectors:

Constitutional tests:
- 71% Public
- 29% Private
- <1% Catholic/Schedule 3
- <1% Research/Academic

Cancer tests:
- 60% Public
- 28% Private
- 10% Catholic/Schedule 3
- 2% Research/Academic
NATA Accreditation

<table>
<thead>
<tr>
<th>NATA Status</th>
<th>Number of laboratories</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Service</td>
<td>Research</td>
</tr>
<tr>
<td>Already NATA accredited</td>
<td>67</td>
<td>5</td>
</tr>
<tr>
<td>Not NATA accredited</td>
<td>1</td>
<td>7</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>68</strong></td>
<td><strong>12</strong></td>
</tr>
</tbody>
</table>

- Non-accredited laboratories performed 0.14% of all tests.
- The number of accredited laboratories has risen by 85% over 5 ½ years.
- 32/72 laboratories were also accredited for MPS, of which 22 (69%) were accredited under the 2017 NPAAC requirements.
Number of tests performed

Total tests performed: 1,181,923

- 545,029 constitutional tests
- 115,121 cancer (somatic) tests
- 146,719 maternal serum screening tests
- 307,770 newborn screening tests
- 67,284 diagnostic biochemical tests
Comparison with 2006 and 2011

Cytogenetic and molecular test volumes (2006, 2011 and 2016/17)

10% increase in tests per capita since 2011*

*Likely an underestimation due to differences in the definition of a single test between surveys

Cytogenetic data were not collected in 2006.
Referral sources

- General Practitioners: 28%
- Obstetricians/Fetal Medicine: 21%
- Pathologists: 15%
- Oncologists: 5%
- Clinical Geneticists: 7%
- Paediatricians: 8%
- Other Medical Practitioners: 16%
Test indications

- Diagnosis (constitutional) 55%
- Diagnosis (somatic) 12%
- NIPS 9%
- Therapy selection/monitoring 9%
- Family cascade testing 5%
- Pre-implantation genetic testing 2%
- Population screening 6%
- Testing of fetal tissue 2%
## Test categories

<table>
<thead>
<tr>
<th>Test Category</th>
<th>Constitutional (% of tests)</th>
<th>Cancer (% of tests)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Targeted testing for predefined variants:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- By a molecular method</td>
<td>77</td>
<td>57</td>
</tr>
<tr>
<td>- By FISH/ISH</td>
<td>&lt;1</td>
<td>14</td>
</tr>
<tr>
<td>Testing for undefined variants in 1 gene</td>
<td>1</td>
<td>&lt;1</td>
</tr>
<tr>
<td>Testing for undefined variants in 2 - 49 genes</td>
<td>3</td>
<td>8</td>
</tr>
<tr>
<td>Testing for undefined variants in ≥ 50 genes</td>
<td>&lt;1</td>
<td>2</td>
</tr>
<tr>
<td>Karyotyping</td>
<td>10</td>
<td>17</td>
</tr>
<tr>
<td>Microarray</td>
<td>7</td>
<td>1</td>
</tr>
<tr>
<td>WES</td>
<td>&lt;1</td>
<td>0</td>
</tr>
<tr>
<td>WGS</td>
<td>&lt;1</td>
<td>0</td>
</tr>
<tr>
<td>Gene expression studies</td>
<td>0</td>
<td>&lt;1</td>
</tr>
</tbody>
</table>
Distribution of tests across laboratory disciplines

Constitutional Tests
- Genetic Pathology: 73%
- Haematopathology: 2%
- Anatomical Pathology: 7%
- Chemical Pathology: 2%
- Immunopathology: 1%
- Reproductive Genetics: 8%
- Research: 1%

Cancer Tests
- Genetic Pathology: 56%
- Haematopathology: 18%
- Anatomical Pathology: 18%
- Chemical Pathology: 4%
- Immunopathology: 2%
- Reproductive Genetics: 2%

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Workforce

Senior medical/scientific staff

- 27% increase in FTE since 2011 (1011 to 1288)

Emerging groups within the laboratory workforce:
- Clinical bioinformaticians (23.2)
- Computer scientists (18.2)
- Genetic counsellors (14.3)
- Other medical staff (18.3)

Scientific/technical staff

- Technician
- Other Medical Scientist (without PhD)
- Other Medical Scientist (PhD)
- Other Medical Scientist
- Other Fellow/Postgrad. qualification
- MHGSA

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Laboratory supervision

<table>
<thead>
<tr>
<th>Supervising staff</th>
<th>No. (%) of laboratories</th>
</tr>
</thead>
<tbody>
<tr>
<td>FRCPA Genetics, FFSc Genetics, FHGSA, or a combination</td>
<td>60 (75%)</td>
</tr>
<tr>
<td>FRCPA Genetics</td>
<td>34 (43%)</td>
</tr>
</tbody>
</table>

No. of laboratories without access to a supervising Pathologist (FRCPA, any discipline): 17 (21%)

No. of laboratories without access to a supervising Pathologist (FRCPA, any discipline) or scientist with FFSc Genetics or FHGSA: 9 (11%)

Complex tests performed in the absence of staff with scopes of practice in genetics (FRCPA Genetics, FFSc Genetics or FHGSA): 12% (no. ~4,600) microarrays
45% (no. ~300) WES/ WGS
Interstate transfer of samples for testing

20% of tests were performed on interstate samples*

- 123,328 (23%) constitutional tests
- 13,974 (12%) somatic tests
- 6,796 (10%) biochemical diagnostic tests

*Likely an underestimation as details of regional origin was not provided for 16% of samples

Data include cytogenetic, molecular and biochemical diagnostic tests (MSS and NBS are excluded).
International transfer of samples

Outgoing tests (3,625*)
- 31% increase since 2011
- 1,537 NIPS
- 302 WES
- Remainder mostly MPS panels

*Substantial additional direct flow of samples to overseas laboratories

Incoming tests (8,386)
- 5,000 comprehensive HLA genotyping
- 354 pre-conception carrier screening
- 119 WES
- Remainder mostly MPS panels
Genomic data storage and sharing

Genomic data storage:
A wide range of platforms were used:

- Hospital servers (29%)
- Local laboratory servers (21%)
- “Multiple storage systems” (22%)

Satisfaction with data storage facilities:

Contribution to international databases:

Yes

No
Funding

State samples

Interstate samples

* Data on Medicare funding for interstate samples not provided in 2011.

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Reporting times

- Median and 90th centile reporting times were provided for 74% of tests (94% in 2011)

<table>
<thead>
<tr>
<th>Test Type</th>
<th>Constitution</th>
<th>Predefined variants (molecular)</th>
<th>Predefined variants (FISH)</th>
<th>MPS panel &gt;50 genes</th>
<th>Karyotype</th>
<th>Microarray</th>
<th>WES</th>
<th>WGS</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Constitutional</td>
<td>Constitutional</td>
<td>Constitutional</td>
<td>Constitutional</td>
<td>Constitutional</td>
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<tr>
<td></td>
<td></td>
<td>2016/17 median (range)</td>
<td>2016/17 90th centile (range)</td>
<td>2011 median</td>
<td></td>
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</tr>
<tr>
<td>Predefined variants (molecular)</td>
<td>Constitutional</td>
<td>11 (1-106)</td>
<td>21 (3-190)</td>
<td>28</td>
<td></td>
<td></td>
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<tr>
<td></td>
<td>Somatic</td>
<td>9 (1-54)</td>
<td>14 (3-100)</td>
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<td></td>
<td></td>
<td></td>
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<tr>
<td>Predefined variants (FISH)</td>
<td>Constitutional</td>
<td>5 (1-9)</td>
<td>8 (1-45)</td>
<td>10</td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td></td>
<td>Somatic</td>
<td>9 (1-56)</td>
<td>13 (3-70)</td>
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<td></td>
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<tr>
<td>MPS panel &gt;50 genes</td>
<td>Constitutional</td>
<td>60 (14-119)</td>
<td>96 (21-204)</td>
<td></td>
<td>17</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td></td>
<td>Somatic</td>
<td>42 (12-96)</td>
<td>100 (14-100)</td>
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<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Karyotype</td>
<td>Constitutional</td>
<td>17 (7-49)</td>
<td>22 (8-63)</td>
<td></td>
<td>17</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Somatic</td>
<td>14 (6-35)</td>
<td>21 (8-62)</td>
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<td></td>
</tr>
<tr>
<td>Microarray</td>
<td>Constitutional</td>
<td>19 (8-63)</td>
<td>25 (12-107)</td>
<td></td>
<td>42</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Somatic</td>
<td>18 (7-33)</td>
<td>32 (10-55)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>WES</td>
<td>Constitutional</td>
<td>72 (50-119)</td>
<td>120 (72-164)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>WGS</td>
<td>Constitutional</td>
<td>102</td>
<td>139</td>
<td></td>
<td></td>
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</tr>
</tbody>
</table>
Summary and limitations

- High participation rate, but many laboratories were unable to provide all details sought by the survey

- Increase in total test volumes, which was limited to the molecular sector

- Most tests were for diagnostic purposes, and half of all requests were made by General Practitioners or Obstetricians/Fertility/Fetal Medicine Specialists

- Growing rate of interstate sample transfers

- Limited insights into offshore testing

- Concerns regarding genomic data storage infrastructure

- Contribution of genomic data to international variant databases is currently limited

- Some laboratories performing complex tests without supervision from pathologists or scientists with qualifications indicating proficiency in genetic/genomic testing
Acknowledgements

• Participating laboratories
  – Especially those who contributed to the pilot studies

• Steering committees
  – Technical Working Group convened by Department of Health
  – RCPA

• A/Prof Brett Lidbury

• Dr Debra Graves

• Dr Kym Mina & Prof Graeme Suthers

• PathWest colleagues