Australian Health Genetics/ Genomics Survey 2017 Communications Package: Department of Health

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RCPA
The Royal College of Pathologists of Australasia

This report has been prepared on behalf of the Royal College of Pathologists of Australasia, for the Department of Health. The views expressed in this document do not necessarily represent the views of either organisation. This report is based on data provided by participating laboratories.

HIGH LEVEL PROJECT EVALUATION

NUMBER AND TYPE OF LABORATORIES

Eighty-seven Australian laboratories delivered various categories of genetic testing, including biochemical genetics, newborn bloodspot screening, pregnancy-related or population screening tests during the 2016 to 2017 financial year (1 July 2016 to 30 June 2017). All were invited to participate in the Stocktake. Eighty-three of 87 laboratories (95.4%) submitted full or partial returns.

Eighty of these laboratories provided details about their NPAAC category, accreditation status and test numbers. Of these, 51.3%, were in the public sector and delivered 45.1% of all completed tests; 30% were in the private sector delivering 53.6% of all completed tests; 15% were research delivering 0.2% of all completed tests, and 3.8% were Catholic/ schedule 3 delivering 1.1% of all completed tests.

LABORATORY ACCREDITATION

Seventy-two of 80 laboratories (90%) were NATA accredited. Non-accredited laboratories performed 0.14% of all tests. Compared with 39 participating NATA accredited laboratories in 2011, the number of accredited laboratories has risen by 85% over 5 ½ years. Thirty-two of the 72 laboratories offering genetic/ genomic testing were also accredited for massively parallel sequencing, of which twenty-two (69%) were accredited under the 2017 NPAAC requirements.

NUMBER OF TESTS

A total of 1,181,923 tests were reported. They comprised 660,150 genetic/ genomic tests (constitutional – 545,029; cancer – 115,121); maternal serum screening (146,719); newborn bloodspot screening (307,770), and biochemical genetic diagnostic tests (67,284).

Genetic/ genomic tests in 2011 were grouped as molecular or cytogenetic. Using these categories, the volume of molecular genetic tests has increased by 73% over the past 5 ½ years. By contrast, the volume of cytogenetic tests fell by 40%.

REFERRAL SOURCES AND TEST INDICATIONS

The most common referral sources for genetic/ genomic tests in 2016/17 were General Practitioners (27.7%), Obstetricians/ Fertility/ Fetal Medicine Specialists (21.1%) and Pathologists (15.4%). Other significant clinical referral sources were Paediatricians (8.1%), Clinical Geneticists (6.6%) and Oncologists (5.4%).

The most common reasons for testing were for diagnostic purposes for constitutional genetic conditions (55% of requests) or for cancer (12%). Other clinical indications included various forms of “cascade testing” of relatives for familial variant(s); therapy selection; minimal residual disease (leukaemia) and transplant monitoring; population screening; several categories of prenatal testing, and preimplantation genetic screening.

TEST CATEGORIES

In the constitutional setting, targeted analysis to assess for the presence or absence of a predefined genomic variant(s) represented the largest test category (78.3%). This category also accounted for most cancer tests (71.1%), although FISH/ ISH analysis represented a higher proportion of cancer-related targeted tests (20.2%) than constitutional targeted tests (1.2%). Chromosomal karyotyping represented 17.5% of cancer-related and 10.2% of

constitutional tests. By contrast, chromosomal microarray analysis accounted for 7.0% of constitutional and 1.3% of cancer tests. The other listed test categories were sequencing-based: grouped as single gene; 2-49 genes; 50+ genes; exome, and genome, as well as gene expression studies.

WORKFORCE

The total number of FTEs identified by this survey was 1287.8. This represents a 27% increase in workforce compared with 2011; however, account should be taken of the wider scope and the inclusion of non-accredited laboratories in the 2016/17 Stocktake.

LABORATORY SUPERVISION

Sixty-one of 81 laboratories (75.3%) included staff with locally-recognised professional qualifications indicating scopes of practice in genetics (FRCPA Genetics, FFSc Genetics, FHGSA, recognised overseas qualifications or a combination).

Thirty-three laboratories (40.7%) included a supervising Genetic Pathologist. Seventeen (21%) did not have access to any supervising Pathologists (FRCPA, any discipline). Forty-nine (60.5%) had a supervising scientist with FFSc (genetics) or FHGSA. Nine laboratories (11.1%) did not have access to either a Pathologist (FRCPA, any discipline) or scientific staff with any locally-recognised professional qualification indicating proficiency in genetic laboratory practice.

An assessment was also made of the numbers, and associated percentages, of tests performed in the absence of medical or scientific staff with professional qualifications relevant to genetic laboratory practice (FRCPA Genetics, FFSc Genetics or FHGSA). These included targeted testing directed towards predefined variants – approximately 52,200 tests (10.5%); targeted testing for undefined variants (1 to 49 genes) – 10,600 tests (36.4%); targeted testing for undefined variants (≥ 50 genes) – nil (0%); untargeted testing by karyotype – 3,200 (4.2%); untargeted higher-resolution testing by microarray – 4,600 (11.6%), and whole exome or genome sequencing – 300 (44.7%).

INTERSTATE AND INTERNATIONAL TRANSFER OF SAMPLES FOR TESTING

The volume of samples transferred interstate or overseas for genetic/ genomic testing continues to rise. The percentage of interstate transfers has more than doubled over the past 5 ½ years, from 9.6% in 2011 to at least 19.8% in 2016/17. For constitutional, cancer and biochemical genetic tests, the percentages of interstate transfers were 23%, 12% and 10%, respectively.

As many laboratories did not provide information on the state/ territory-of-origin of tested samples, the overall rate of increase is likely even greater.

The reported number of samples transferred to international laboratories has also risen – from 2,766 in 2011 to 3,625 in 2016/17 (a 31% increase). As described in the report, numerous additional samples are known to have bypassed the laboratories contributing to the Stocktake, which means that this number is an underestimate.

LABORATORY INFORMATIC INFRASTRUCTURE

Laboratories used a variety of systems for sample registration, tracking and report storage. Most used Laboratory Information Management Systems (LIMS), local electronic records/

databases located on either laboratory/ hospital servers, or local hard drives. Fewer than 10% of laboratories, both service and research, used laboratory workbooks or stored reports as hardcopies.

GENOMIC INFORMATIC INFRASTRUCTURE

Genomic data generated from patient samples were stored on a variety of platforms, of which hospital servers (29%), local laboratory servers (21%), and “multiple storage systems” (22%) were the commonest options. Approximately a third of service laboratories considered their data storage facilities to be suboptimal; in particular, laboratories indicated that details of locally-identified variants were not retained in a searchable local database for future reference.

INTERNATIONAL GENETIC/ GENOMIC DATABASES

Approximately three-quarters of laboratories did not contribute details of locally identified genomic variants to international variant databases.

REPORTING (TURNAROUND) TIMES

Substantial improvements were observed in the median reporting times for targeted molecular assays since 2011, however the range of reporting times was broad and a substantial proportion of results were delivered beyond published recommended turnaround times. Median reporting times for molecular tests involving screening genes for unknown disease-causing mutations have lengthened since 2011. The 2016/17 median cytogenetic reporting times were unchanged from 2011; however, the 90th centile reporting times for constitutional- and cancer-related chromosomal karyotyping both exceeded the current recommended standard for Australian laboratories.

FUNDING

The 2016/17 survey data revealed that funding arrangements for genetic tests have changed substantially. For within-state tests, federal funding (Medicare) covered almost half (49%) of within-state tests in 2016/17, compared with 35% in 2011. There have been corresponding falls in the proportion of tests funded by most other sources. Federal funding covered approximately two thirds of tests performed on interstate samples in 2016/17. Most of the remaining interstate tests were funded directly by patients. On the background of an increasing number of tests being transferred across state borders, the overall proportion of interstate tests paid for by patients has doubled to approximately one quarter of tests.