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New nationwide research on genetic testing in Australia

The Royal College of Pathologists of Australasia (RCPA) has unveiled new, nationwide research on Australia's medical genetic testing, which was conducted for the Department of Health. The findings are being reported at the RCPA's conference, Pathology Update, at the Melbourne Convention and Exhibition Centre this weekend by Project Leads, Dr Anja Ravine (Genetic Pathologist) and Dr Sarah Nickerson (Genetic Pathology registrar).

"The new findings reveal that genetic and genomic testing is becoming increasingly integrated into healthcare, reflected by a 73% increase in molecular test requests over the past 5 ½ years. The findings also show that test requests are made by a broad range of medical practitioners for a wide variety of clinical reasons. However, rapid advances in technology and genomic knowledge bring challenges, which need to be addressed if genomic technology is to become further embedded within the healthcare setting," said Dr Ravine.

The RCPA undertook prior surveys of genetic testing in 2006 and 2011. "The Federal Government agreed that repeating the survey would provide a vital update on the nature and availability of genetic tests for Australian patients to make comparisons with historical data, provide information about workforce change requirements, and to facilitate modelling for future service provision," explained Dr Ravine.

All Australian laboratories known to have offered genetic or genomic tests for medical purposes during the 2016/17 financial year were invited to participate. "Although participation was voluntary, engagement was excellent with over 95% of laboratories providing data," said Dr Nickerson.

More than 660,000 genetic/genomic tests were reported over the one-year survey period. The most common reasons for testing were for diagnostic purposes for constitutional (heritable) genetic conditions (55% of requests) or for cancer (12%). Other clinical indications included various forms of "cascade testing" of relatives for familial gene variants; therapy selection; minimal residual disease (leukaemia) and transplant monitoring; population screening; several categories of prenatal testing, and preimplantation genetic screening. The stocktake also revealed that a further 308,000 newborn bloodspot screening tests, 147,000 maternal serum screening tests, and 67,000 biochemical genetic diagnostic tests were performed over the 2016/17 financial year.

The medical practitioners most frequently requesting genetic/genomic tests were General Practitioners and Obstetricians/Fertility/Fetal Medicine Specialists – together these medical practitioner groups were responsible for half of all test requests. The remaining 50% of tests were requested by a wide variety of medical practitioners.

The survey also revealed that a substantial proportion of genetic/genomic tests are now performed by the private sector. Thirty percent of participating laboratories were in the private sector and, together, they delivered almost two thirds of the total number of requested constitutional (heritable) and cancer (somatic) genetic tests. Just over half of the 83 responding laboratories were from the public sector; these laboratories performed approximately a third of constitutional and cancer genetic tests, but were responsible for a higher proportion of biochemical genetic tests (~60%). Of the other laboratories, 15% were research laboratories, delivering 0.2% of all tests, and 3.8% were Catholic/schedule 3 delivering 1.1% of tests.

Another important observation from the most recent survey is the growth in patient samples being transferred interstate or overseas for genetic/genomic testing. "The percentage of interstate transfers has more than doubled over the past 5½ years, to at least 20% of all genetic/genomic test requests", explained Dr Ravine. "A considerable proportion of laboratories did not provide details about the state/territory-of-origin of tested samples, which means that the overall rate of increase is likely to be even greater. The reported number of samples transferred to international laboratories has also risen by 31% and, during the survey, it became apparent that large numbers of international test requests had bypassed laboratories contributing to the stocktake, which means that this value is actually a considerable underestimate," said Dr Ravine.

These latest findings from the survey data revealed substantial changes to funding arrangements. For within-state tests, Federal funding (Medicare) covered almost half (49%) of the tests completed in 2016/17, compared with 35% in 2011. "The change in the proportion of tests covered by Federal funding was largely reflective of an increase in requests for tests with longstanding Medicare Benefit Schedule (MBS) item numbers, rather than resulting from the addition of new items to the MBS since the previous survey," explained Dr Nickerson.

For test requests referred interstate, Federal funding covered a higher proportion (approximately two thirds), with the vast majority of these being performed in the private sector. Most of the remaining tests transferred interstate in 2016/17 were paid for directly by patients. "Direct patient payments for genetic tests have doubled since the 2011 survey, and this is largely reflective of the growing uptake of non-invasive prenatal screening of maternal blood for common fetal chromosomal trisomies," said Dr Nickerson.

At the same time, findings from the survey reveal that the ongoing rapid advances in genomic technologies, particularly the advent of "massively parallel" testing platforms and associated computing advances, are challenging many Australian laboratories. These testing platforms generate huge volumes of patient genetic data, necessitating secure and robust data storage solutions. A third of service laboratories considered their data storage facilities to be suboptimal for future demands.

"The big shifts going on within the industry also have major implications for laboratory staffing," said Dr Ravine. "An example is the progressive upgrading of conventional cytogenetic testing into more sensitive or efficient DNA-based methods. The net result has been a 40% fall in the volume of cytogenetic tests over the past 5 ½ years and a corresponding increase in newer molecular-based DNA tests.

"This means that there is a requirement for the workforce to upskill in order to effectively fully integrate these technologies into mainstream healthcare. A key matter requiring attention is ensuring future provision of both pathologists and medical scientists trained to the level required for safe provision of medical genomic tests to Australian patients. It is hoped that the findings from this research will further support the development of a National Genomics Policy Framework to improve coordination and consistency in approach to integrating genomics in healthcare," explained Dr Ravine.

The survey also yielded an insight into the contribution of Australian laboratories to global efforts to advance genetic knowledge and its clinical application. It is standard practice to compare patient genomic variations identified during clinical testing with variants recorded in a range of databases and within scientific literature. "Comparison with both local and

international databases is an essential step in assessing the clinical significance of test findings for individual patients," explained Dr Nickerson. "Despite this heavy reliance on international variant databases, the findings reveal that most Australian genetic testing laboratories are yet to establish systems that enable them also to contribute to this important international resource. Through sharing details of curated DNA variants, a broader repository of data is generated, which facilitates the accurate interpretation of genomic results and ultimately improves patient care."

All of the participating medical laboratories partook in this research on a voluntary basis. "We would like to acknowledge the superb contributions of each of the laboratories that participated in what was time-consuming data gathering. The survey provides representative data that can be used to describe current practices and trends in medical genetic testing in Australia," said Dr Ravine.

For further information on the RCPA, visit www.rcpa.edu.au or see our updates on Facebook or Twitter - @PathologyRCPA or Instagram - @the_rcpa.

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About the Royal College of Pathologists of Australasia:

The RCPA is the leading professional organisation representing pathologists, medical specialists and scientists who provide pathology testing in Australasia. Its mission is to train and support pathologists and to improve the use of pathology testing to achieve better healthcare.

Media enquiries:

Dr Debra Graves
Chief Executive Officer
The Royal College of Pathologists of Australasia
+61 2 8356 5858
Debrag@rcpa.edu.au

Linsey Brown
Managing Director
The Public Relations Agency
+61 425 514 005
Linsey@thepublicrelationsagency.com.au