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First Ever Australian Genetic Testing Survey report released

The Royal College of Pathologists of Australasia (RCPA) has released the first ever national Australian survey reporting on the extent of genetic testing performed in the country.

The Report, entitled "*Report of the Australian Genetic Testing Survey 2006*" will be launched at the RCPA's annual Pathology Update / 25th WASPaLM Conference on Friday, March 13th 2009.

Funded by the Australian Department of Health and Ageing, through the Quality Use of Pathology Program, and involving close collaboration with the Human Genetics Society of Australasia, the report provides an overview of the availability and prevalence of more than 400 genetic tests which were offered in 2006.

Chairman of the RCPA Genetics Advisory Committee, Dr Graeme Suthers, said advances in genetic knowledge have led to the introduction of genetic tests for clinical purposes. The survey was designed to provide valuable information regarding the current level of testing in Australia to assist in the development of policy, planning and resource allocation for such tests.

"Importantly, the Report identified some key baseline levels from which to benchmark testing availability and changes in access for coming years," said Dr Suthers.

The Report data exposed an extraordinary variation between testing (assay) frequency across different genetic tests.

"At the time of the surveyed period, in 2006, five genetic tests were funded under the Medicare Benefits Scheme (MBS), and a further 437 non-MBS rebated tests were offered and available to greater or lesser degrees across the country.

"While we expected some level of divergence between the frequency of tests performed across the range available, the survey demonstrated a gap to the magnitude of thousands to tens of thousands-fold between the numbers of certain genetic tests being performed, with some being performed more than 10,000 times, while others were performed once or twice, if at all, in 2006.

"Overall, the total number of genetic tests performed in 2006 were low, with only 41,497 assays for MBS molecular genetic tests and 119,354 assays for non-MBS tests.

“To put this into perspective relative to other forms of pathology testing, there were more than a million clinical chemistry tests performed per million population in 2006 in Australia, while genetic testing was much less common, with roughly a thousand tests per million population.”

The survey provides unprecedented access to statistical data and information regarding non-MBS tests, which previously had never been compiled, and found the following key results:

- 56 laboratories in Australia were identified as providing molecular genetic testing for clinical purposes in 2006 that was not Medicare rebated. 93% provided data for this survey.
- Of the 437 types of non Medicare-rebated molecular genetic tests offered by Australian laboratories in 2006, 55% were offered by only one lab, and a further 21% were offered by only two laboratories.
- 40% of the assays were for medical screening purposes; e.g. pre-transfusion testing or neonatal screening; 28% of the assays were for diagnostic purposes; 8% were for non-heritable variants in cancer; 5% were to define the genetic status of unaffected relatives in families with a documented mutation; 1% low-volumes tests of different types; and in 18% of the assays, the reason was unidentified.
- Half of the types of test were provided by laboratories offering less than 10 types of test; 10% of the laboratories offered 40 or more types of test.
- 17% of the laboratories reported doing less than 100 assays during 2006; 27% reported doing more than 1,000 assays during this period.
- The majority of types of test were provided by laboratories in only one State or Territory, while only 56 types of test (13%) were provided by laboratories in four or more regions.

President of the RCPA, Dr Beverly Rowbotham says the survey illustrates the complexity surrounding genetic testing access in Australia.

“There are tremendous variations between access and availability across the country,” says Dr Rowbotham.

“Importantly, the RCPA recommends that the survey be repeated on a regular basis due to the rapid advances and changes in availability of genetic tests. It is essential that we keep a handle on this.

“In 2007, the diversity of types of test offered increased by approximately 8%, while the number of assays rose by 67%, reflecting an increased volume of MBS testing rather than an increase in non-MBS testing.

“Many challenges identified in the process of conducting the Report are being addressed.

“For example, the lack of clarity about which laboratories are conducting which tests is being rectified, with the launch of an online catalogue providing an overview of which gene tests are performed by which laboratories.

“This resource tool, which is particularly important for laboratories, referring doctors and specialists, will be hosted on the RCPA Manual website (<http://rcpamanual.edu.au/>).

“The strength of data within this Report underpins the RCPA’s calls for the need for a ‘National Genetics Framework’ to ensure Australia is not left behind in the genetics revolution that is dramatically altering medicine.”

The survey Report is available for download from the RCPA website:
<http://www.rcpa.edu.au//static/File/Asset%20library/public%20documents/Media%20Releases/AustralianGeneSurvey2006.pdf>

The RCPA is the leading organisation representing pathologists in Australasia. Its mission is to train and support pathologists and to improve the use of pathology testing to achieve better healthcare.

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