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## Australians' Knowledge and Perceptions of Direct-to-Consumer Personal Genome Testing

Savard, J, Mooney-Somers, J, Newson, A, Kerridge, I. (2014)

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### Abstract:

**Background:** As direct-to-consumer personal genome testing (DTC-PGT) is increasingly available in Australia, knowledge of Australian's perceptions and attitudes towards his technology is needed in order to assess the (potential) impact it might have on the Australian public and health care system.

**Aims:** To explore the knowledge and perceptions of direct-to-consumer personal genome testing (DTC-PGT) in an Australian sample.

**Methods:** An online survey asking about knowledge and perceptions of DTC-PGT, undertaken between October 2011 and April 2012, of 270 Australian residents. Results were analysed using SAS.

**Results:** Our study found limited consumer knowledge of, and interest in pursuing DTC-PGT in Australia. 93% of respondents correctly identified DTC-PGT as available to consumers directly, but only 40% correctly identified its availability in Australia. When asked about the content and value of the information DTC-PGT provides, the majority of respondents identified that DTC-PGT could provide information about one's health and/or ancestry (82% and 74%). Additionally, respondents indicated they believed this information to be equally important as non-genetic information about one's ancestry and health.

**Conclusion:** While few respondents expressed an intention to pursue DTC-PGT (27%), the majority of people, irrespective of whether they wished to pursue it or not, believed that genetic information was as important as non-genetic information in regards to their health and their ancestry. The value ascribed to genetic information suggests genetics plays a role in people's lives and at this time, further qualitative research could explore the ways in which people might use and understand the genetic information provided by DTC-PGT.

**Key Words:** predictive genetic testing, consumer health information, ethical issues, survey methodology

## **Introduction**

Traditionally, people have accessed medical diagnostics and therapeutics through a health practitioner. However the emergence of the Internet and other information technologies has profoundly changed this dynamic and consumers are now able to access sophisticated biotechnologies without medical assent or advice. One of the most striking examples of this is direct to consumer personal genome testing (DTC-PGT). In this situation, consumers purchase an 'at home' DNA collection kit (usually a kit that collects a saliva sample) which they return to the company for testing. The company usually then emails a report of the test results to the consumer. This report may provide information about the consumer's carrier status for several genetic conditions and risk estimates for common multi-factorial diseases, such as diabetes, heart disease, various cancers and Alzheimer's disease. Testing may also provide information about ancestry, including an ancient ancestor migration report, an estimate of the likelihood that the consumer is from a particular ethnic group and a description of whether or not the consumer has ancestral genetic ties to certain historical figures. Since its introduction in 2007, DTC-PGT has functioned as a tool that individuals, groups (and families) can use as a means of accessing genetic information about themselves outside of the clinical setting.

The emergence and consumption of DTC-PGT can be explained, at least in part, by the increasing impact of genetic information in health care. It is also due to the increasing prominence of the notion of personalised medicine. Both of these factors emphasise the importance of genetic influences on and understandings of health and illness, and of clinicians and consumers 'knowing' and acting upon this information. DTC-PGT, it is argued, promotes health by providing individuals with information they need to make informed health care decisions (1).

There has, however, been considerable debate about the value of the information DTC-PGT provides, including how it is understood and/or used by consumers. Recent studies have explored characteristics and motivations of early DTC-PGT adopters outside Australia, including how consumers interpret and act on their risk reports (2), and whether health-related actions correspond appropriately to the genetic information received (3). Almost all of this research has been done in the United States, the most prominent market to date for DTC-PGT. While there are growing markets in China, Korea and India (4-6), cultural similarities between Australia and the United States make the former body of literature the most appropriate source of critical commentary. This research has also tended to focus on specific groups, including: 'early adopter' populations (7, 8), health and 'high-tech' company employees (9), social networkers (10), individuals who are affected or have a family member affected with a genetic condition (11-13) or individuals enrolled in particular studies to study the impact of genetic susceptibility testing (14). There is currently no data regarding the knowledge and perceptions of DTC-PGT in any Australian population.

We conducted an online survey between November 2011 and April 2012 to explore the Australian's knowledge and perceptions of DTC-PGT and their expressed interest in pursuing DTC-PGT for themselves.

## **Methods**

Survey Instrument and Recruitment:

An online survey consisting of 30 questions was designed to capture a sample of the Australian public's knowledge and perceptions of DTC-PGT. The survey consisted of four sections.

Section one requested demographic information relating to age, gender, country of birth, country of residence, level of general education and education around genetics. Section two asked respondents about their general knowledge of DTC-PGT, such as what does DTC-PGT provide information about, who is the test available to, how can the test be obtained, how they heard about testing and whether they believed the test was legal or available in Australia. Section three asked respondents about their perceptions of the value information from DTC-PGT could provide. The final section asked those respondents who had undergone DTC-PGT about their experience of DTC-PGT, while non-consumers of DTC-PGT were asked if they were planning on pursuing DTC-PGT.

The survey was distributed through online mailing lists to a university student and staff population, through online networks to patient support groups for people living with genetic conditions, via a consumer health information website, a science and technology website and through societies associated with genetic conditions in Australasia. The survey was launched in November 2011 and closed at the end of April 2012, and used the online survey tool [peoplepulse.com.au](http://peoplepulse.com.au). The survey took approximately 5 minutes to complete. As a link to the survey was distributed through list serves and posted on a website for individuals to access, there were no tracking statistics attached to the survey link – meaning that the reported responses are of the number of surveys commenced and completed. The University of Sydney Human Research Ethics Committee approved this study and survey instrument (Approval and Reference #2012/2264).

#### Statistical Analysis:

Data was analysed using SAS 9.3 (SAS Institute Inc, Cary, NC, USA) using the PROC SURVEYFREQ procedure to calculate frequencies based on the 270 responses included for analysis. Demographic factors (age, gender and education level) were tested in PROC SURVEYFREQ in tables using adjusted Pearson chi-square analysis at a  $P < 0.05$  level of significance. Summary statistics for relevant variables are reported using PROC SURVEYFREQ and are reported with 95% confidence intervals.

## Results

### *Demographics*

Overall, 402 surveys were commenced – of which 282 were completed. Of these – 270 were completed by individuals who resided in Australia and were included in the analysis.

Table 1 summarises the demographic data. Respondents were categorised into three age groups according to characteristics that we felt may be relevant to awareness and uptake of DTC-PGT. The first age group (18-31 years) was chosen to include a population demographic that is generally familiar with genetic concepts (for example via school science education) and with several of the media platforms used to promote DTC-PGT (for example, social media and online blogs). The second age category (32-45 years) was chosen to encompass potential consumers with permanent jobs, disposable incomes and still in their reproductive years. This group also includes those who have completed their post-secondary or postgraduate education. The final age category (46 and up) was chosen on the grounds that it was likely to include people who may be less interested in the implications of genetic information for their

own reproductive choices, but who might still have an interest in some of the information DTC-PGT provides about future health and ancestry.

**Table 1: Demographic information frequency summary**

	Percentage of Respondents N=270 (95% CI)
<b>Age</b>	
18-31	55.9% (55.6% - 56.3%)
32-45	20.7% (20.4% - 21.0%)
46 and up	23.3% (23.0% - 23.6%)
<b>Gender</b>	
Male	27.0% (26.8% - 27.3%)
Female	72.9% (72.7% - 73.2%)
<b>Education</b>	
Secondary Education or Less	21.4% (21.2% - 21.8%)
Post-Secondary Education and Higher	78.5% (78.2% - 78.8%)

*Expressed Intention to Pursue DTC-PGT*

When asked if they planned on pursuing DTC-PGT, 73.0% (95% CI, 72.7% - 73.2%) indicated that they were not, while 27.0% (95% CI, 26.8% - 27.3%) said they were intending to or were currently having DTC-PGT done. No demographic variables – including age, gender or education – were predictive of an interest in pursuing DTC-PGT.

*Knowledge about DTC-PGT and its Availability*

In section two of the survey, all questions were ‘forced’ answers – without an option of ‘I don’t know.’ When asked what type of testing could be done using DTC-PGT, most responded that DTC-PGT could be used for health testing (82%; 95% CI, 81.2% - 81.8%) and ancestry testing (74.4%; 95% CI, 74.1% - 74.8%). This is consistent with respondents’ expectations of the information provided by DTC-PGT; with most indicating that DTC-PGT would provide information about one’s health (particularly one’s future health) and ancestry (Table 2).

**Table 2: What information DTC-PGT provides to consumers: Survey responses to the question, “DTC-PGT provides genetic information to consumers. What do you think this information could tell you about?”**

What the information from DTC-PGT could tell you about	Frequency of Responses N=1174 (95% CI)
Your future health	89.3 (89.0 - 89.5)
Your ancestry	84.4 (84.2 - 84.7)
Your current state of health	64.1 (63.7 - 64.4)
Your family	58.9 (58.5 - 59.2)
Your future children	48.5 (48.1 - 48.9)
Your physical appearance	35.6 (35.2 - 35.9)
Your children	35.2 (34.8 - 35.3)
Your personality	18.9 (18.6 - 19.2)
NOTE: Respondents could select more than one answer. The frequency count is expressed as a percentage of the total number of responses, not the total number of respondents.	

The vast majority (93.3%; 95% CI, 93.2% - 93.5%) of respondents correctly indicated that DTC-PGT was available to anyone (as it is now), with 75.2% (95% CI, 74.9% - 75.5%) (correctly) indicating that consumers themselves could purchase a DTC-PGT. It should be noted that in the introduction to the survey, respondents were provided with a definition of direct-to-consumer. Fewer respondents - 24.4% (95% CI, 24.1% - 24.8%) incorrectly believed that a doctor must request DTC-PGT. Of those surveyed, 40.4% (95% CI, 40.0% - 40.7%) correctly thought it was available in Australia (as it is now), while 53.3% (95% CI, 53.0% - 53.7%) were unsure. In addition, 54.8% (95% CI, 54.5% - 55.2%) of respondents did not know if DTC-PGT was 'legal' in Australia, while 43% (95% CI, 42.6% - 43.3%) correctly believed that it was (and remains so). At the time of the survey, Australians were able to access DTC-PGT online through providers located overseas; there was also one Australian company offering these tests (15).

#### *Perceived Value of the Information Provided by DTC-PGT*

Respondents were asked how they rated the value of the information that DTC-PGT could provide in terms of its importance to the individual. Respondents were asked to indicate whether this data had medical, personal or familial importance (or any combination of these) or whether the information was simply 'interesting.' Respondents could choose multiple answers. The frequency count is expressed as a percentage of the total number of responses, not the total number of respondents. The results are summarised in Table 3.

**Table 3: Perceived value of information provided by DTC-PGT: Respondents' answer to the question: "Do you think the information DTC-PGT provides is (select all that apply)"**

How do you view the importance of the information gained through DTC-PGT	Frequency of Responses N=492 (95% CI)
Medically Important	37.0 (36.7 – 37.4)
Personally Important	28.9 (28.6 – 29.2)
Important to your family	22.6 (22.3 – 22.9)
All the above	43.3 (43.0 – 43.7)
A combination of the above	19.6 (19.3 – 19.9)
None of the above	0.37 (0.36 – 0.41)
It is just interesting information	30.4 (30.0 – 30.7)
NOTE: Results are presented in the same manner as the options appeared to the respondents in the survey and respondents could select more than one answer. The frequency count is expressed as a percentage of the total number of responses, not the total number of respondents.	

Respondents were also asked how they rated the value of information provided by DTC-PGT, including the value of genetic health information and genetic ancestry information for themselves and for their family. On a Likert scale, for *genetic health information*, over half of respondents indicated genetic health information was important for themselves (64.8%; 95% CI, 64.5% - 65.2%) and their family (58.1%; 95% CI, 57.8% - 58.5%). When asked about *genetic ancestry information*, less than half of respondents found genetic ancestry information important for themselves (44.8%; 95% CI, 44.4% - 45.2%) and their family (38.9%; 95% CI, 38.5% - 39.2%). Respondents were also asked which type of information they believed was more important in determining their health: (i) their genetically predicted health status; or (ii) their health history and current health status. The majority of respondents selected they were equally important (62.2%; 95% CI, 61.9% to 62.6%). Likewise, respondents were asked which type of information they believed was more important in determining their ancestry – their

known family ancestry and stories or their genetically determined ancestry. Again, the majority of respondents also selected they were equally important (53.7%; 95% CI, 53.3% to 54.1%).

## Discussion

This study provides the first empirical account of knowledge and perceptions of DTC-PGT in an Australian context. Several findings from this survey are particularly interesting. First, the results of this study suggest limited knowledge of, and interest in, pursuing DTC-PGT. Second, when asked about the content and value of the information DTC-PGT provides, the majority of respondents identified that DTC-PGT could provide information about one's health and/or ancestry and appeared to value this information as much as non-genetic information about one's ancestry or their current/future health.

Only 40% of respondents correctly identified that DTC-PGT is available in Australia (as it is now), with the majority of respondents (54.8%) being unaware whether DTC-PGT was 'legal.' DTC-PGT was, and remains legal. But while respondents appeared uncertain about the availability or legality of DTC-PGT, respondents knew they were (as they currently are) able to access this technology independently from health care providers. The fact that respondents appeared to have a limited awareness of DTC-PGT is unsurprising; as at the time this survey was distributed (November 2011 to April 2012) there was no direct-to-consumer advertising in the Australian market. However, Australian's accessing the Internet could be exposed to online advertisements for these services. Prior to and during the time in which the survey was conducted, there was also discussion of other types of direct-to-consumer personal genome tests in popular culture, such as the television show, *'Who do you think you are?'* (16), newspaper articles about these tests and what they could offer to consumers (17-20) and radio reports (21). While only 27% of the survey sample expressed an interest in pursuing DTC-PGT, the finding that more than half of respondents indicated they believed the tests to be informative, relevant and valuable suggests the potential for greater uptake. The relational aspect of genetic information between family members who are genetically related raises the potential for DTC-PGT to inform and have an impact on an individual and perhaps their family. This impact can be in regards to their future health and/or their accepted narratives regarding their ancestry. As genetic information may be understood and acted upon by individuals, families and groups in different ways, it is imperative that the benefits and limits of this information are understood, and appropriate options are available, so people are able to act upon this information in a meaningful way.

There are two limitations to this study that caution against over-interpretation of this data. First, the data is from an online survey sent out via email lists associated with a university student and staff network, via genetic disease/disorder awareness societies around Australia, a science and technology website and through a health consumer website. As a result, there is potential for selection bias to younger, educated individuals and towards individuals knowledgeable about genetics, genetic disorders/diseases and genetic screening/testing programs. Indeed, our data tend to support this observation – with the largest proportion of respondents being younger (55.9% were aged 18-32) and respondents with a post-secondary or higher education (78.5%). This is an over-representation, as the 2011 Australian census data found only 24% of the population to be individuals between 18-34 years of age, with 26% of those within this group attending a post-secondary institution (22). Second, the overall sample size is small. To be representative of the Australian population, a larger cross-section

of the public would need to be sampled. Despite the limitations to this study, our data provides a preliminary view of knowledge of and perceptions of the Australian public towards DTC-PGT.

As the cost of sequencing continues to fall and the understanding of the information this technology provides advances, the next step in genetic technologies – whole genome sequencing – is likely to become a feature of both clinical care and the health marketplace. Until such time that whole genome sequencing is a routine part of clinical care, it is likely that Australians who wish to know more about their genetics (but who do not have a clinical ‘indication’) will seek this information through DTC-PGT services. At this stage, however, it is unclear what the clinical, public health and social impact of DTC-PGT will be. Although (only) 27% of the respondents to our study expressed an intention to pursue testing (a very large number of people if cautiously generalised to the Australian community) it is particularly noteworthy that the majority of respondents, irrespective of whether or not they wished to pursue DTC-PGT, indicated that they believed the information provided by DTC-PGT was as important as one’s ancestral stories, medical history and current health status. This apparent privileging of genetic information in understanding one’s self and one’s future health suggests that genetics has already deeply infiltrated our culture. Further quantitative research with a nationally representative sample is needed to explore who might be interested in pursuing these tests, while qualitative research could explore the ways in which people might use and understand the genetic information provided by DTC-PGT.

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