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04 June 2014  
National Health and Medical Research Council  
GPO Box 1421  
Canberra, ACT, 2601  

To the NHMRC Public Consultations Board,  

Re: Submission on NHMRC Draft Statement and Resource for Consumers: Direct-to-Consumer Genetic Testing  

Please find attached our submission in response to the NHMRC Public Consultation on the following documents:  

- Direct-to-Consumer Genetic Testing – A Statement from the NHMRC; and  
- Understanding Direct-to-Consumer Genetic DNA Testing – An information resource for consumers  

We sincerely appreciate your willingness to accept this submission after the deadline.  

This submission is from an organization: the Centre for Values, Ethics and the Law in Medicine (VELiM), School of Public Health, The University of Sydney. We are happy for this submission to be made publicly available.  

Sincerely,  

Ms. Jacqueline Savard  
Dr. Ainsley Newson  
Associate Professor Ian Kerridge
Direct-to-Consumer Genetic Testing – A Statement from the NHMRC

Question 1: Is the draft Statement presented and written in a manner that is easy to understand?

• Yes, the draft statement is written in a clear manner and in our view will be easy to understand.

• However, in our view the structure is hard to follow. For example, consumers are inherent to the commencement of the DTC testing process, so we wonder why they are discussed last in the draft statement. We feel the consumer should be discussed first, and that this content be followed by the description of the need for a robust evidence base and then the role of professional involvement and education, concluding with overarching ethical, legal and social issues (ELSI) DTC genetic testing raises. We comment further on ELSI aspects of DTC testing below.

Question 2: Is the draft Statement comprehensive and does it cover all of the key issues?

No, the draft Statement is not comprehensive. The following issues should also be included:

• It is unclear as to what the overall purpose of the statement is in its current draft iteration. It does mention “highlighting the important issues for the appropriate delivery and use of health-related genetic testing” (page 2); however, it does not explicitly say which health care professionals are expected to use or access this resource. Also, is this statement intended to guide best-practice for health professionals to follow when they encounter a consumer with DTC genetic test results who may be seeking professional advice? This has not been made clear and given that this scenario is mentioned in the draft consumer statement, it might be pertinent to include something here too; perhaps also cross-referencing to the advice for health professionals document.

• Currently, the information from DTC genetic tests, while of a questionable accuracy (analytically and clinically) are described in the statement as sources of genetic information about one’s health. Describing DTC genetic tests as a source of health information is a misnomer and this should be clearly stated under the heading: “The need for robust evidence.” In this section, the types and levels of accuracy and validity could be clearly articulated.

• The regulation and legal standing of DTC genetic tests is unclear – both overseas and here in Australia. Therefore, it would be beneficial if this point is made clear and the status of the tests and their results are made explicit. Papers by Savard and Nicol and Hagger provide an overview of the current legal status of DTC genetic tests in Australia1.

The statement “The growing uptake of DTC genetic tests is in part driven by individuals who are taking more responsibility for their own health” is a context- and situation-dependent claim. Either this statement should be appropriately referenced, or it could be revised as follows: “there are a variety of reasons why an individual might seek DTC genetic information about themselves.” Significantly, current research in Australia (underway by the authors of this submission) on the experiences of Australian consumers of DTC genetic testing indicates that consumers of DTC genetic tests in Australia pursue DTC genetic testing for a range of reasons, each imbued with personal, familial and contextual meaning to the consumer as it relates to their life and expectations.

The identification of appropriate health care professionals with the expertise and background to reasonably understand and advise on the results of DTC genetic tests needs to be identified. In the NHMRC draft Resource for Consumers, several mentions of directing consumers to their general practitioners are made throughout the document. In this Statement, however, it is not made explicit that general practitioners would be assisting consumers with this kind of interpretation nor advising them on a course of action in light of health-relevant genetic information accessed through DTC genetic testing. It would be helpful to have guidance on who in the health care system could or should be the first point of contact for consumers seeking expert advice and how they could go about accessing such assistance. This is currently unclear.

Mention is made of the complex ethical, legal and social issues that arise as a result of genetic testing, but there is neither an identification of what these issues are, nor a discussion of how they could impact the individual being tested, their family nor their larger implications in society. We suggest adding a brief a section on the ethical, legal and social aspects of DTC genetic testing. Several relevant (but not exhaustive) ethical, legal and social issues that could be included and expanded upon are:

- Privacy and Discrimination
- A right to know versus not to know
- Consent (as, for example, parents are able to submit tests on their child’s behalf)
  - This could also include the dynamic nature of genetic information and that with future research, current information has the potential to change, which could lead to different meanings and interpretations (both inside and outside of the clinic).
- Family Communication of both the intention to test and results of testing

The statement currently does not contain reference to empirical research nor reviews of supporting claims. Any empirical claims in the text of this draft statement should be appropriately referenced. Examples include:

- That genetic tests can cause unnecessary physical or psychological harm (2nd paragraph of section entitled “The Importance of Professional Involvement and Education.”; and

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Q3: Is there any further advice that should be included in the draft statement on the accuracy of DTC genetic testing?

- Yes, further advice is needed. We believe a definition of the levels of accuracy would be helpful for this statement, which indicates the level of accuracy current medical genetic tests are held to and the current level the DTC genetic tests are rated at and perhaps where they have yet to establish accuracy. We feel this would correctly inform health care professionals about the level of accuracy they should look for when interpreting DTC genetic tests and would help them explain to consumers why DTC tests are not equivalent to clinical genetic testing.

- Additionally, we'd suggest removing the statement on companies providing different interpretations of the same sample unless it can be referenced with a peer-reviewed citation. This information may not continue to hold true (for example if testing becomes more robust) and so may prematurely render this statement out of date.

Q4: Is more advice needed in the draft statement on legal risks, risk rated insurance or privacy issues?

- Yes, we recommend more advice on legal risks, risk rated insurance or privacy issues. Following up on the suggestion we made under question 2 above, in a section dedicated to the ethical, legal and social issues raised by DTC genetic testing, a clear discussion of the benefits and possible consequences in terms of legal obligations and privacy concerns could perhaps be included in this proposed new section.

- While these are mainly concerns for the consumer being tested, it would be wise for advising health professionals to be aware of the legal standing of the tests and legal consequences of testing for consumers so they could advise them correctly if the consumer is thinking of being tested or has already been tested and is seeking advice on how to proceed.

- The third paragraph of the section on the “need for consumer information…” seems to imply that insurers will definitely want the results of DTC tests; whereas they may not. This paragraph could be re-worded to add “typically” or “often” before “are required”

Q5: Is more advice needed in the draft statement on the usefulness of DTC genetic testing?

We believe the current level of content is appropriate for the aims of the statement. However, there are a few sections (highlighted throughout this submission) where we feel further justification and a degree of depth would strengthen the current statement. There are also the following organising recommendations we wish to suggest:

- Reordering the introduction. First identify the challenges presented by DTC genetic testing and then proceed to state the reason for this statement and
intended aims. This would then be followed by a brief description of DTC genetic testing, including ALL types of information that can be procured through this service. The focus on only health related genetic information (of which ancestry can play a role) is understandably the focus of this statement, but it should be acknowledged that other information is also available through these services. This would then lead into a discussion on the information-seeking behaviours of consumers, specifically reframing the current statement of:

- “Individuals may undergo DTC testing out of curiosity, to discover whether they possess particular genetic variations. Such motivation allows greater engagement and appreciation for science and what makes us unique.”

  - In response, our research indicates a wide range of reasons for testing, and most consumers are not interested in ‘genetic variations’ as it is currently stated. The motivation for engagement in science and the ‘uniqueness of an individual’ are actually contradicted by our findings, the results of which appear to confirm how common or usual the attitudes of individuals are. We would suggest the following wording:

    “Individuals may undergo DTC testing for a range of reasons, which often reflect personal, familial and societal priorities.”

- We would recommend re-ordering the contents of the statement to address main issues in the following order:
  1. Consumer information and support;
  2. A robust evidence base;
  3. Professional involvement and education; and
  4. Ethical, Social and Legal Issues arising from DTC Genetic Testing

This order places the consumer at the centre of the issue, which as the main actor who initiates testing should be the primary concern of health care professionals. This builds on what is needed (strong evidence) to guide professional involvement and then include any overarching concerns relating to the process as a whole.

- We suggest removing “out of curiosity” in the reasons for testing, as consumers will have different reasons for choosing to take a DTC genetic test

- We would suggest removing the reference to the recent FDA action regarding DTC testing as this will date the statement, particularly if the action is resolved.

- We suggest adding ‘usually’ before “without the involvement” in the first paragraph.

- Regarding the sentence on testing without consent (in the consumer information section): we are not sure what this adds to this statement? It could perhaps be expanded upon or removed? If a consumer was considering surreptitious testing they would unlikely be swayed by this statement, and a person who was testing without their knowledge would not know to read it.
• The statement seems to assume that all DTC genetic testing is web or internet based and offered overseas. However there is DTC genetic testing in Australia, both on and offline. An example of offline onshore DTC genetic testing is testing DNA for the purpose of providing nutritional advice. Many of the same issues arise, so the statement should be revised throughout to encompass DTC testing wherever it occurs, both on and offshore and within and outside Australia. Likewise, there are varieties of DTC that are purchased by consumers but involve a health professional. The statement should either address this (perhaps by way of a quick taxonomy?) or explicitly exclude them.

• We would suggest re-wording a paragraph in the final section to read: “While it is encouraging that individuals are engaging more actively with their own health and wellbeing, the provision of potentially unnecessary and/or inaccurate DTC genetic tests is concerning. It may lead to individuals spending money on tests they don’t need; or it could produce an expanding cohort of ‘worried well’ and to place unnecessary strain on Australia’s health system. Conversely [but why conversely?] risks also exist with the inappropriate self-management of susceptibility and conditions and the possibility of delayed recognition of preventable or treatable disease.”

Reference List:
Understanding Direct-to-Consumer Genetic DNA Testing – An information resource for consumers

Question 1: Is the draft resource for consumers presented and written in a manner that is easy to understand?

The current draft resource is rather negative in tone. We submit that the draft could be revised to be just as informative and meaningful but with more neutral content. Additionally, the resource alludes to concepts and concerns relevant to DTC genetic testing but provides little by way of explanation or follow-up for consumers.

Taking these points in further detail:

- The tone of the draft resource is negative, which discourages readers from seeking help for further information regarding what these tests offer, their meaning or the implications of the results from such a test. As a potential or actual consumer, the reader is left with a very negative view of these tests which could contribute to stress or anxiety for those who were considering being tested or for those who might have already been tested. The resources provided and overall idea that consumers should refer to their doctor (in this case a General Practitioner) is arguably merely referring the problem on, as recent research suggests that while GP’s are aware of these tests, they do not necessarily have the confidence to accurately interpret and explain the results.

- There are several sections missing in terms of content and context that would clarify information provided and why it was provided in the manner in which it appears (please see our response to question 2 below).

Question 2: Is the draft resource for Consumers comprehensive and covers all of the key issues?

No, the draft is not comprehensive and there are several key areas that require further content and addressing, as follows:

- The structure of the content does not address issues as they arise, and the reader has to look ahead in the resource for information that is then not provided. For example, why someone might get this type of testing and what the information they receive is a key question many consumers have regarding this technology – and this information is not included in this resource.

- A section describing motivations for testing is not present in this resource. Being able to identify with consumers and why they might seek this information creates a level of acknowledgement that they do have an interest in this information and that it is an important part of the testing process. This builds confidence on the part of the consumer that their concerns or interest are not misplaced, but are legitimate and deserve follow up and consultation by professionals who can offer guidance. Further, the section from the current (2012) consumer resource commencing with: “you might like the idea…” could be re-introduced into the

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section with the heading “Important issues to consider…” in the Draft Consumer Statement (2014).

- The limitations of the test are missing from this current draft. By mentioning and discussing the types of information a consumer might receive from such tests, consumers are better informed when they make their final decision to pursue testing or not. The broad term of ‘health information’ is not informative enough and if a consumer has taken the time to search out and consult this resource, it would be helpful to provide them with a detailed description of the types of information provided by DTC genetic tests. Such sections were present in the 2012 version of ‘Direct-to-consumer DNA genetic testing: An information resource for consumers’ which provided an adequate level of detail. If drafted in generic terms it is unlikely that this information will become outdated quickly.

- The draft is missing key evidence as to why doctors (in this regards, general practitioners) are identified at the first point of contact for the interpretation of their test results. Research conducted in Australia identifies a gap in knowledge – where current general practitioners do not have the confidence to interpret or provide meaningful guidance on how to understand these tests. As a first point of contact to access further or repeat testing, it is understandable that GP’s are identified. At present, however, there is simply not any sort of systemic architecture in place for consumers once they have visited their GP and could then be referred on to genetic counselling.

- The content regarding the familial nature of genetic information is not well presented in the current draft form. The consequences of finding out genetic information about the self are not well articulated (for example, only one sentence is dedicated to this). The cultural and social aspects of genetic testing statement in this section seems out of place and understated, as genetic information about the self has the potential to cause harm to the self, family and others. In the 2012 ‘Direct-to-consumer DNA genetic testing: An information resource for consumers’ resource, there was an entire section dedicated to discussing the potential emotional impact of these tests, which is followed up by a section on the impact of results for family members – this is something that is entirely missing from this current draft.

- It seems a shame that the section on effects on the individual has been excluded from the updated version of the resource

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3 Ibid.
Question 3: Is there any further advice that should be included in the draft Resource for Consumers on the accuracy of DTC genetic testing?

Yes, the accuracy section requires more information regarding the levels of accuracy and how that could impact upon an individual's results.

There are two levels of accuracy that could and should be described to consumers: the analytic and clinical accuracy⁴:

- Analytic accuracy can be defined as the ability to accurately and reliably measure the genotype of interest (specifically the nucleotide sequences) and
- Clinical accuracy is the ability of the test to detect or predict the associated disorder.

Each of these types of accuracy, we acknowledge, are contentious – but identifying this to consumers is paramount, to ensure they are provided with the most accurate information upon which they might base their decision to undergo testing or their decision to pursue clarification regarding their test results.

Additionally, we would suggest re-wording the accuracy paragraph to read: “You can’t always be sure that a DTC test has been done properly or that the results are accurate. Companies offering DTC tests are mostly located overseas, even if the initial mailing address is within Australia. Overseas laboratories are not required to follow strict Australian laboratory regulations for genetic tests, though there may be regulations in the country in which the laboratory is located. DTC genetic tests often come with disclaimers that release the company from responsibility for inaccurate test results.”

Question 4: Is more advice needed in the draft Resource for Consumers on legal risks, risk rated insurance or privacy issues?

Yes, we believe there should be a better contextualisation of the information provided as well as further detail on where these guiding principles come from (as in regulation or Acts) relevant to the issues discussed. There are also two misleading statements within this section.

- What relevant Australian legal contexts refer to privacy and how do they apply (or as the case may be, not apply) to the use, storage and distribution of genetic information? There is no mention about the procedures many consumers go through when registering their purchase kits and the legally binding ‘contract’ they often enter into with the company. There was mention of this in the 2012 ‘Direct-to-consumer DNA genetic testing: An information resource for consumers’

which would require updating, as changes to the Privacy Act 1988 occurred as recently as March 2014.

The first misleading statement appears on page 3, where it states: “In general, once you agree to these further uses of your DNA, it is usually impossible to change your mind or withdraw your consent.”

- This is an inaccurate statement – consumers are able to write to the company from which they purchased their testing kit, and if they originally consented to have their genetic information included in the research bank of the company, they can request that their information be withdrawn. This frames the consent procedure as biased towards the company’s interests, which is a negative framing of the issue of consent for these tests.

The second misleading statement, on page 3 also, states: “Some DTC companies also sell information about you and your genetic results to pharmaceutical and other companies.”

- There is no empirical evidence to suggest this occurs. There is ongoing internal research by DTCPGT companies on their consumers’ data, and they conduct research in partnership with companies, but do not on-sell their data. Statements that companies on-sell their consumers’ data are, arguably, speculative.

Further, the section on consumer protection should state that it is the “Australian legal consumer protections” that are not enforceable rather than “The protections.” The dot point regarding research could have the following added: “or acceptance of research on your sample may be a condition of testing.”

**Question 5: Is more advice needed in the draft Resource for Consumers on the usefulness of DTC genetic testing?**

Yes, we believe more advice needs to be included in the Resource for Consumers along the following issues:

- A more detailed definition of ‘health information’ is needed, as it is currently too broad.

- There needs to be a section detailing what the tests provide, which could then segue to a subsequent section that would provide information about the impact and limitations of such tests.

- There needs to be an adjustment to language in the document from largely negative to a more neutral tone. This would encourage consumers to thoughtfully consider why they might be seeking this information and if this is the best course of action to answer their questions.

- A section discussing the personal, familial and societal impacts of DTC genetic testing is needed (an excellent section of this nature was included in the 2012

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‘Direct-to-consumer DNA genetic testing: An information resource for consumers’ which set up the conversation in the document to discuss the impact to families and how to access further resources).

- Greater detail on the legal and privacy implications is required (along with corrections); please see our response to question 4 above.

Reference List:


