

Article

Human Genetics Society of Australasia Position Statement: Online DNA Testing

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Abstract

Increasingly, consumers have been able to seek DNA testing online to explore their personal genetic information. This increased access to a range of genomic tests has raised concerns among health professionals tasked with providing guidance and support to patients requiring genetic/genomic testing. Individuals will seek genomic testing for a range of purposes; equally, the medical marketplace offers a range of different test types. The Human Genetics Society of Australasia (HGSA) published their first statement on Direct to Consumer Genetic Testing (2012 PS02). This is a revised statement, which considers developments in the field of online DNA testing, including rapid technological changes, diversity of applications and decreasing costs of testing. It draws from the first empirical nationwide study (Genioz – Genomics: National Insights of Australians) and insights from consumers with experience of this technology. The rapid adoption of these tests and the broad range of potential consequences have informed perspectives within this statement. It is the position of the HGSA that both individuals/consumers and health care professionals/providers should be supported to make *informed choices* about online DNA testing. This means *adequate and ongoing education and resources should be available* for individuals/consumers and health care professionals/providers before, during and after testing. Health care professionals/providers should be appropriately trained, have relevant experience and should be able to demonstrate (or provide evidence of) a current certification in their field of practice. This statement was ratified at the 2018 HGSA Council Meeting and was recently reviewed in 2019 for consistency with other HGSA position statements.

Keywords: Online DNA testing; direct-to-consumer genetic testing; genetic/genomic testing; personal genomic testing; consumer genomics; Australia

(Received 14 July 2020; accepted 15 July 2020; First Published online 25 August 2020)

Background

This position statement provides the stance of the Human Genetics Society of Australasia (HGSA) on the role, status and supports needed for consumers/patients who may choose to pursue online DNA testing. For clarity, the following context and definitions will apply to this statement.

Genomic tests¹ are becoming widely available beyond clinical settings. The term *online DNA test* describes a range of different commercially available tests that are marketed to individuals and can be accessed directly, either online or through health care professionals/providers who do not practice within clinical genetics services. The term *online DNA testing* was elected as this is the nomenclature used by those who pursue testing (Metcalfe, Hickerton, Savard, Terrill et al., 2018). Examples are provided in Table 1.

People who seek genomic information may include members of the public, patients, physicians and other health care professionals/providers.

In this Statement, we focus on the genomic information provided by companies offering online DNA testing where the test is one or more of the following: (a) marketed directly to individuals/consumers; (b) available to individuals/consumers outside a clinical context and (c) available to individuals/consumers in a clinical context but in the absence of clinical indications to suggest testing is necessary. This Statement aims to be specific to the legal, regulatory and social context of Australia. However, many of the ideas presented here offer approaches that could be useful in Australasia².

Who Pursues Online DNA Testing and Why?

Individuals may seek online DNA testing for a variety of reasons. These can include:

- to explore health issues (explain past illnesses, present conditions/diagnoses or predispositions to conditions that have not yet manifested)

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Cite this article: Savard J, Terrill B, Dunlop K, Samanek A, and Metcalfe SA. (2020) Human Genetics Society of Australasia Position Statement: Online DNA Testing. *Twin Research and Human Genetics* 23: 256–258, <https://doi.org/10.1017/thg.2020.67>

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Table 1. Alternate terms for ‘online DNA testing’ and types of tests currently available

| Commonly used Internet search terms | Types of tests available ^a |
|--|---|
| At-home genetic/genomic test ^b | Ancestry/family history tracing |
| Ancestry DNA test ^b | Aptitude for sports/activities in children |
| Consumer-directed genetic/genomic test ^b | Cancer predisposition (e.g., breast cancer) |
| Consumer genetic/genomic test ^b | Carrier testing (including reproductive (preconception and early pregnancy) testing/screening) |
| Direct-to-consumer (DTC) genetic/genomic test ^b | Dating compatibility |
| DNA health test ^b | Drug response/pharmacogenetics/pharmacogenomics |
| DNA testing or gene test ^b | Ethnicity-based ancestry testing |
| Family tracing DNA test ^b | Food intolerances/preferences (including wine) |
| Genetic/genomic wellness test ^b | Health-related risk predictions (e.g., Alzheimer’s disease, blood conditions, heart conditions, other inherited/genetic conditions) |
| Health and wellbeing DNA test ^b | Health and wellbeing testing |
| Internet genetic/genomic test ^b | Non-invasive prenatal testing |
| Online DNA test ^b | Nutrition and/or wellness |
| Online genetic/genomic test ^b | Paternity |
| Personal genetic/genomic test ^b | Personality |
| | Skincare |
| | Sporting ability/fitness/aptitude in adults or children |
| | Talents |

^aThis list of test types is likely to change as the field continues to develop.

^bWildcard term, so searching for ‘test*’ would include test, tests and testing.

- to explore or understand family history (health and/or genealogical)
- to explore personal genetics and genomics in more depth, such as to satisfy curiosity
- to guide lifestyle or fitness regimens.

Individuals seeking online DNA testing will do so for different reasons, and they may have more than one reason. In addition, individuals may encourage relatives and friends to undergo testing. It is the position of the HGSA that testing should only be done by individuals who are well informed, aware of the risks and benefits of testing, and able to consent for their DNA to be collected and analyzed.

There is also an emerging market for online DNA testing in children. This includes testing in newborns and young children for a range of health conditions and other characteristics. It is the position of the HGSA that decisions about having a child tested should be well informed and based on good scientific data and that parents should have access to clinical support and professional genetic counseling about this decision or advice, and support for the range of results received. For further information about genetic testing in newborns and children, please refer to the following HGSA position statement on Predictive and Pre-symptomatic Genetic Testing in Adults and Children (Vears et al., 2020) and the Supplemental Online (Direct to Consumer) DNA Newborn Screening Tests (Human Genetics Society of Australasia, 2019).

How do People Access Online DNA Testing?

Online DNA tests can be accessed in a number of ways.

- Individuals can order tests directly from an online DNA testing company—the increasingly common ‘direct-to-consumer’ pathway. In this approach, the test is ordered online, the

individual collects their own DNA sample (or with the assistance of a pathology service) and the DNA samples are sent directly to a laboratory for processing. The laboratory may, *or may not*, be located in Australia (Allyse et al., 2018).

- An online DNA testing company may strongly recommend and/or require that the test is ordered on the individual’s behalf by a trained health care professional/provider or by a health care professional/provider who may be an employee of/on contract with the testing company.
- Individuals may initiate the test through their health care professional/provider, who refers to the online DNA testing company or orders the online DNA test on behalf of the individual.

What Formats Are Available for Online DNA Test Results?

Information from online DNA testing companies can be returned to individuals in a variety of formats. Different approaches to testing and/or interpretation are used by each company. This can range from providing raw genomic data to interpreted reports generated by the company, containing results of analyses for the traits and variants they include in their product. This means individuals could potentially receive different or conflicting results and/or advice from different companies. Currently, there are no regulations or rules governing how samples are collected and processed by online DNA testing companies, or how data should be analyzed or interpreted.

Increasingly, online DNA testing company websites and independent online applications (‘apps’) offer consumers ways to do further analyses on their raw or unprocessed DNA data. These apps enable people to generate results for purposes beyond the remit of the original test. For example, results of online ancestry DNA testing could be used to generate health-related information, independent of health professional support or advice, with potential consequences for the individual, their own and their family’s health care decisions (Nelson et al., 2019).

The HGSA’s Position on Online DNA Testing

The HGSA recognizes that online DNA tests are (and will most likely continue to be) available to people in Australia.

It is the position of the HGSA that both individuals/consumers and health care professionals/providers should be supported to make *informed choices* about online DNA testing. This means *adequate and ongoing education and resources should be available* for individuals/consumers and health care professionals/providers before, during and after testing (Metcalf, Hickerton, Savard, Stackpoole et al., 2018). Health care professionals/providers should be appropriately trained, have relevant experience and should be able to demonstrate (or provide evidence of) a current certification in their field of practice (Brett et al., 2012; Horton et al., 2019; Middleton et al., 2017).

Results from online DNA tests may have an impact on an individual’s perception of their health and/or wellbeing, or the relationship between an individual/consumer and a health care professional/provider and/or the wider health system. To best support and engage with individuals considering (or who have already undergone) online DNA testing, discussions about the following key issues are encouraged:

- **Choosing to test and choosing an appropriate test:** Online DNA tests and their results can be used for a range of purposes; it is important to consider whether testing is appropriate and how to choose a test that will provide the information sought.

- **Expectations:** Individuals can overestimate the utility of results from online DNA tests; it is important to have realistic expectations of the actions that can be taken.
- **Support and information:** Online DNA tests and results can be hard to interpret and understand. It is important to know where, and with whom, help can be obtained (Savard *et al.*, 2019).
- **Actionability:** Online DNA test results may be more useful personally than medically. Individuals should consult with their health care professional/provider if they are concerned about their health and to seek advice and confirm any health-related online DNA test results from an accredited provider before acting on the results.
- **Family implications:** Online DNA tests generate information not only about the individual being tested but also about their family members. Family members may not want this information generated and may not want to know this information. Discussions within families about these issues are encouraged.
- **Evidence:** Some online DNA test results and related associations may not be supported by strong scientific evidence.
- **Rapid change:** Online DNA testing is still a developing field where the evidence base is incomplete. An individual's results may differ between companies and/or change over time as new information becomes available.
- **Regulation:** Online DNA tests may be offered by Australian and overseas companies. The availability of these tests and the techniques used may differ from laboratory to laboratory, and the level of regulation applied to testing companies differs in each country. This makes it difficult to ensure that results are analyzed consistently by different testing laboratories, and this may raise questions about reliability of results. It is also difficult to determine what legislation or regulation will be applied to how DNA can, or has been, stored, shared and used; this may depend on the country in which the company and laboratory are based—as that could be in different locations (Kalokairinou *et al.*, 2020; Savard, 2013).
- **Privacy:** Data generated from online DNA testing may be shared or sold to other research groups or companies. It is important to be aware of how much the Australian law can protect an individual's data, and when it cannot.
- **Commercial interests and/or conflicts of interest:** Consumers should be aware that some companies provide training that they require health care professionals to undertake before they can order a test for a patient. This may present a conflict of interest.
- **Discrimination:** Online DNA test results may affect an individual's ability to obtain risk-rated insurance products, such as life insurance.

Within the Australian health care system, there are regulatory and accreditation standards for genomic tests and processes. When individuals are concerned about their health, they should consult an appropriate health care professional to decide whether an online DNA test is appropriate and discuss how useful test results could be to make health-related decisions.

Acknowledgments. The authors would like to thank and acknowledge the work of the Genioz Study Team: Sylvia A. Metcalfe, Ainsley Newson, Kathleen Gray, Bronwyn Terrill, Clara Gaff, Anna Middleton, Brenda Wilson, Jacqueline Savard and Chriselle Hickerton. Empirical data from this study underpin and inform the ideas and perspectives outlined in this statement. The authors would also like to thank the additional members of the Education, Ethics and Social Issues Committee of the Human Genetics

Society of Australasia: Sam Ayres, Jackie Boyle, Michael Gabbett, Julia Mansour, Aideen McInerney-Leo, Ainsley Newson, Amy Nisselle and Danya Vears.

Financial Support. The open access publication of this Position Statement has been supported by the Human Genetics Society of Australasia.

Conflict of Interest. Three of the named authors (Jacqueline Savard, Bronwyn Terrill and Sylvia Metcalfe) and a member of the EEISC committee (Ainsley Newson) were also research team members on the Genioz Study.

Ethical Standards. Not applicable.

Notes

1 Although the term *genetics* has been used in the past, these tests look at more than one gene to include a range of genomic variants. Thus, throughout this statement, we will use the term genomic to include both genetic (single gene) and genomic (more than one gene) tests.

2 This statement is specific to Australia and does not try to provide recommendations applicable to New Zealand or the Australasian region due to different regulatory frameworks around direct-to-consumer advertising and health system diversity.

References

- Allyse, M. A., Robinson, D. H., Ferber, M. J., & Sharp, R. R. (2018). Direct-to-consumer testing 2.0: Emerging models of direct-to-consumer genetic testing. *Mayo Clinic Proceedings*, 39, 113120.
- Brett, G. R., Metcalfe, S. A., Amor, D. J., & Halliday, J. L. (2012). An exploration of genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice. *European Journal of Human Genetics*, 20, 825830.
- Horton, R., Crawford, G., Freeman, L., Fenwick, A., Wright, C. F., & Lucassen, A. (2019). Direct-to-consumer genetic testing. *British Medical Journal*, 367, 15688.
- Human Genetics Society of Australasia. (2019). Supplemental, Online (Direct to Consumer) DNA Newborn Screening Tests. <https://www.hgsa.org.au/documents/item/9582>
- Kalokairinou, L., Borry, P., & Howard, H. C. (2020). 'It's much more grey than black and white': Clinical geneticists' views on the oversight of consumer genomics in Europe. *Future Medicine*, 17, 129140.
- Metcalfe, S. A., Hickerton, C., Savard, J., Terrill, B., Turbitt, E., Gaff, C., ... Newson, A. J. (2018a). Australians' views on personal genomic testing: Focus group findings from the Genioz study. *European Journal of Human Genetics*, 26, 11011112.
- Metcalfe, S. A., Hickerton, C., Savard, J., Stackpoole, E., Tytherleigh, R., Tutty, E., ... Middleton, A. (2018b). Australians' perspectives on support around use of personal genomic testing: Findings from the Genioz study. *European Journal of Medical Genetics*, 62, 290299.
- Middleton, A., Mednes, A., Benjamin, C. M., & Howard, H. C. (2017). Direct-to-consumer genetic testing: Where and how does genetic counselling fit? *Personalized Medicine*, 14, 249257.
- Nelson, S. C., Bowen, D. J., & Fullerton, S. M. (2019). Third-party genetic interpretation tool: A mixed-methods study of consumer motivation and behaviour. *The American Journal of Human Genetics*, 105, 122131.
- Savard, J. (2013). Epidemiology, quality control and consumer access in the medical marketplace: The changing landscape of human genetic technology regulation in Australia. *Journal of Law and Medicine*, 20, 595603.
- Savard, J., Hickerton, C., Tytherleigh, R., Terrill, B., Turbitt, E., Newson, A. J., ... Metcalfe, S. A. (2019). Australians' views and experience of personal genomic testing: Survey findings from the Genioz study. *European Journal of Human Genetics*, 27, 711720.
- Vears, D. F., Ayres, S., Boyle, J., Mansour, J., Newson, A. J., on behalf of the Education, Ethics and Social Issues Committee of the Human Genetics Society of Australasia. (2020). Human Genetics Society of Australasia Position Statement: Predictive and Presymptomatic Genetic Testing in Adults and Children. *Twin Research and Human Genetics*, 23, 184–189.



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Title:

Human Genetics Society of Australasia Position Statement: Online DNA Testing

Date:

2020-08-01

Citation:

Savard, J., Terrill, B., Dunlop, K., Samanek, A. & Metcalfe, S. A. (2020). Human Genetics Society of Australasia Position Statement: Online DNA Testing. *TWIN RESEARCH AND HUMAN GENETICS*, 23 (4), pp.256-258. <https://doi.org/10.1017/thg.2020.67>.

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