HGSA / EESIC Committee Submission to TGA Regarding Regulation of Certain Self-Testing (IVDs) in Australia: particularly in relation to Direct to Consumer Genetic Testing

This submission is made by members of the Education, Ethics and Social Issues Committee (EESIC) on behalf of the Human Genetics Society of Australasia (HGSA). Several committee members were involved in developing the HGSA Position Statement about Online DNA Testing (available here: https://www.hgsa.org.au/documents/item/18).

Should direct to consumer genetic tests be allowed?

Should Direct to Consumer Genetic Tests be permitted in Australia (following evaluation by the TGA) to provide consumers with an alternative to overseas testing which has not been evaluated by the TGA for its quality and performance?

From research conducted by Savard et al (2014)[1], Critchley et al (2014)[2] and the Genomics: National Insights of Australians (*Genioz*) study (www.genioz.net.au), Australians' preferences are for an option to access testing from an Australian company. For example, Genioz interview participants' views can be broadly encapsulated as: *"No option here so I go with the options available to me now"*

However, to sanction regulated tests in this space would require investment in an infrastructure to ensure appropriate monitoring, accreditation and regulation of these tests and develop pathways for consumer access, information, and support.

While regulation aims to reduce harm, there are many ways to interpret and define harm, as it results from direct to consumer genetic testing. Direct to consumer genetic testing is a process with several points where harm can be introduced. For example:

a. Access – there are different considerations when testing is sought onshore or offshore. For some consumers, offshore testing provides a sense of anonymity. For others, offshore access is a rate-limiting step, as the price, inconvenience and longer turn-around times can make testing prohibitive.

b. Privacy – privacy and security of information is a common concern regarding genetics, genetic information and discrimination. Concerns around privacy can include data breaches or unexpected access by law enforcement, third party researchers and/or insurers. In the insurance context, some of these concerns may be mitigated, such as advice from specific health care professionals encouraging patients to seek and secure certain forms of insurance before proceeding with genetic testing.

c. Interpretation of results:

i. While analytic validity (accurate and reproducible testing procedures in a laboratory) are generally reliable, the interpretation of test results by direct to consumer genetic testing companies can vary. Currently, there are regulatory requirements for interpretation in consumer genomics or an agreed-to set of standards that all companies aim to satisfy. This has led to consumers receiving confusing and differing, if not conflicting, reports when their sample is tested by multiple companies[3].

ii. An individual or organisation may interpret results from different perspectives. Most genetic tests will offer a risk result, rather than a definitive answer. A consumer may interpret, and place value on, results differently to a clinician or a larger institution (such as a health care system). Results from online tests may affect an individual's perception of their health and/or wellbeing and may lead to unnecessary testing and follow-up, placing additional stresses on patients, clinicians and systems. Consultation across *all* these levels will be needed.

iii. Importantly, genetic test results can provide sensitive, personal information that is highly emotive and ethically challenging for some consumers; for example, challenges to family communication and potential breakdown in relationships. As a result, harms may be delayed or only emerge after the testing process has been completed.

It is important the TGA is clear about how it might characterise "harm" and how it proposes that regulation to allow onshore testing will minimise it. As suggested in the consultation document, an outcome of testing regulation could be to include a network of support pathways for informed decision-making, including genetic counselling, health referrals and accessible information. However, these will all need resources to be invested in them.

Are there any particular genetic tests that should not be available as a self-test? Please provide reasons why not.

[In this question, we also address the question further in the consultation document related to "should self-tests for serious diseases be able to be supplied in Australia following evaluation by the TGA to determine their safety and performance?"]

The HGSA position is that when individuals are concerned about their health, they should consult an appropriate health care professional to decide whether a genetic test (direct to consumer or other) is appropriate. The HGSA appreciates and supports the existing regulatory and accreditation standards for genetic tests and processes and acknowledges the premises of health and social care in Australia; that is, care should be person-centred and enable meaningful engagement between consumers and health providers.

With regards to self-testing (and direct to consumer genetic testing), medically-relevant testing – including diagnostic testing and risk score testing for severe disease, such as heart disease or cancer – needs to be regulated and supported with appropriate information, counselling and access to referrals. The HGSA recently developed a supplemental position statement recommending against direct to consumer genetic testing for newborn babies, as parents may be confused by this type of testing. The HGSA was particularly concerned that this form of testing may be perceived as a substitute for government-funded newborn screening.

However, the HGSA also acknowledges that prohibition has not stopped the proliferation of direct to consumer genetic tests being accessed through other channels. For example, through limited point of care avenues, e.g., complementary practitioners, or the proliferation of tests now offered for children.

An approach to current standards and measures for oversight

To oversee and restrict testing, a coordinated effort to establish, monitor and enforce oversight would be needed from multiple agencies (for example, Therapeutic Goods Administration, Australian Border Force, National Association of Testing Authorities, Australian Consumer Complaints Commission and Office of the Australian Information Commissioner, Human Genetics Society of Australasia, Royal Australian College of General Practitioners). These (and other organisations) would need to collaborate to: monitor test access; develop standards of validity and utility for approval; and assess support and counselling measures.

The current commercial sector has invested in models that meet current standards by providing limited point of care/health professional interaction, but these are consumer driven, subsequent support is mainly available at an additional expense, and is not guaranteed to be free from conflicting interests. Through these pathways, tests that currently fall outside the Medical Devices Excluded

Purposes Specification are available, but perhaps should not be. For example, direct to consumer genetic testing can provide incidental information about severe disease (such as metabolic genetic tests that include Alzheimer's risk via testing for APOE variants).

One genetic test does not equate to one use of genetic information

The HGSA would also like to note that the nature of genomic testing and information makes the question of test restriction more complicated than it suggests. If the TGA continues to restrict the supply of direct to consumer self-tests, it should consider extending that restriction to include tools that interpret raw data for health purposes.

A consumer can seek their genomic information for 'benign'/ non-health purposes (such as fitness or family/ancestry testing), then download their raw genomic data. This data may then reveal information about more 'severe' or health-relevant information. In recent years, online applications and portals have emerged for consumers to re-submit their data for further analysis (for example, GED match, Promethease and Livewello) – known as third party interpretation. Current regulation would not cover the initial pursuit or form of testing (as ancestry and fitness testing are not currently included in existing regulation), but the latter consequences (the generation of health-specific data) could be within the scope of TGA oversight.

The HGSA suggests that the TGA carefully consider not just current forms of testing available, but the possible uses and harms that can result from the generation and consumer-focused interpretation of genetic information. To best address this mobility of information, we would suggest developing standards that address the context where consumers might seek a test from a provider; this would include standards that tests must meet in order to be available in Australia (including analytic validity, appropriate provision of support services and follow up for consumers) and a level of consumer protection according to laws in Australia (which would address privacy and insurance concerns).

A final point we would like to articulate is the challenge to regulation by direct to consumer genetic testing models that are outside the scope of TGA IVD regulation. As noted in the document, these include point of care-sought direct to consumer genetic tests. As these forms of tests can fall both under clinical and non-clinical review, it is important for the TGA to be clear moving forward how these forms of tests will be classified, reviewed and (potentially) regulated.

Do you have any suggestions on how potential risks to consumers could be mitigated if genetic self-tests were allowed to be supplied in Australia?

In the HGSA Online DNA testing statement[4], we state that 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. Ideally, these resources would not be produced by testing companies themselves.

Additionally, the TGA needs to consider how the broader consumer genomics market may evolve in the future and how and why patients/consumers will access such tests. We suggest that there should be pathways and structures to provide support to individuals who seek these tests and unbiased materials that support them when making decisions.

Key issues that need to be addressed in any conversation about direct to consumer genetic tests include: choice of test; potential harms; expectations of test results; support and information; actionability; family implications; and the potential for discrimination. We also suggest that any

information needs to address the strength of the evidence base supporting a test, variability in regulation, the rapid change in the field of genomics, privacy and commercial interests.

References

1. Savard J, Mooney-Somers J, Newson A, Kerridge I. Australians' knowledge and perceptions of direct-to-consumer personal genome testing. *Internal Medicine Journal.* 2014;44(1):27-31.
2. Critchley C, Nicol D, Otlowski M, Chalmers D. Public reaction to direct-to-consumer online genetic tests: Comparing attitudes, trust and intentions across commercial and conventional providers. *Public Understanding of Science.* 2014:1-20.
3. Shuren J. Statement of Jeffery Shuren, M.D., Director, Centre for Devices and Radiological Health, Food and Drug Administration, Department of Health and Human Services. Washington, DC: House Energy and Commerce Committee's Subcommittee of Oversight and Investigations; 2010.
4. Education Ethics and Social Issues Committee, Human Genetics Society of Australasia. Online DNA Testing Position Statement. 2019; https://www.hgsa.org.au/documents/item/18, 2019.