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Reply to

# Review of the regulation of certain self-testing in vitro diagnostic medical devices (IVDs) in Australia.

Genetic Alliance would like to thank the TGA for the opportunity to provide for this review.

For the purposes of this reply considerations for genetic self testing be broken into two sections. The first section will address the current availability of DNA testing kits. The second section will address testing for other genetic conditions.

## Background of Genetic Alliance

Genetic Alliance Australia was initiated in 1988 in response to a need for support and information on genetic and rare conditions. Our mission is to facilitate support for those affected directly and indirectly by genetic conditions throughout Australia.

Genetic Alliance provides services at the individual, group support and to state and federal bodies. For individuals, we access information on conditions for individual and referral to support organisations where they exist. In addition, we host meetings for support group leaders for education of current issues or areas of common concern. This presents an opportunity for networking and peer learning. We assist condition specific meetings with venue, sourcing speakers, and promotion of event. Genetic Alliance also attends conferences and seminars so as to be current on new developments and learnings in genetics and the rare disease community. These include the Human Genetics Society of Australia annual conference, Australian Genomic Health Alliance and NSW Cancer Institute. Genetic Alliance has participated in workshops, stakeholder meetings and committees at the state and federal level. These include NSW Dep Health Clinical Genetic Executive Committee, National Bio banking Summit and Medical Services Advisory Committee Stakeholder Meeting on Genetic Testing for Childhood Syndromes. Our principle funding is from the NSW Dept. of Health and we receive enquiries from all Australian states and territories, as well as international enquiries.

## Genetic Testing In Australia

Genetic Tests represent a very broad spectrum of tests from specific gene arrays to whole genome sequencing (WGS). Currently there is a wide range of array testing available. This testing targets specific genes known to be associated with genetic conditions. WGS provides more detail on the whole of an individual's genome. This test may provide information on other genes not covered by array testing.

The National Association of Testing Authorities, NATA, provides accreditation authority for analytical laboratories and testing service providers. This accreditation ensures consistent and reliable testing for health industry, flowing on to the public confidence in the services they receive.

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Genetic testing may be done by an individual to provide information on an inherited condition such as Huntington, Fabry disease or some cancers; or a unique health condition have array genetic testing that confirms the condition. Some diverse health conditions have *de novo* variations that occur in the conception process. Other diverse health conditions are as a result of polygenic changes, have no known identifier and are now collectively called SWAN conditions; Syndrome Without A Name, requiring WGS. All testing for conditions requires training for the necessary skills to provide quality, consistent and meaningful results for the person.

The means to obtain a human sample for genetic testing may be relatively simple such as via a saliva sample or cheek swab. More complex processes involve blood collection, skin scrapings, or by surgery to obtain tissue from an affected organ or from a neoplasms and cancer. Testing for conditions in unborn infants is available prior and during pregnancy.

Testing maybe done at different times during a person's life. Routine newborn heel 'blood spot testing' is conducted for genetic conditions that have treatments available such as galactosaemia or Maple Syrup disease. This testing is optional for parents to consent to. For newborns who display severe or have unknown causes of health conditions, consent for genetic testing my provide information as to a cause and possible treatment.

As some genetic conditions do not present to later in life. This may be in childhood, adolescence, early or late adulthood.

Current data collection for Australian conditions for diagnosis and research is reliant on medical publications and data sharing with national and overseas institutions. There is currently limited data collation for rare genetic conditions for the Australian population. This is likely to change with developments in genetic technology and processing of large data sets.

## Section 1

## 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 evaluation by the TGA for its quality and performance?

The availability in the marketplace of testing kits for ancestry, paternity, athleticism and metabolic processing has been stimulated by client demand as well as active and constant marketing.

As Australia is a country populated mostly by migration, ancestry testing may quell curiosity, provide new or confirm leads for family research. Other demands are to confirm questions of family parentage, within the privacy in the home environment. This is a considered personal decision, with potential long lasting impacts, outside of the health domain. Testing for athleticism or metabolic rate may be curiosity or used as an indicator for more investigation. This demand is from a health conscious public for autonomy in health and lifestyle decisions, despite the lack of need for medications or clinical actions.

However, there is a lack of public knowledge about genetics, partially from urban myths as a result of crime investigation TV series and a view that genetic testing is a definitive, with one type of test.

There exists concerns about the companies providing the DTC testing of Australian citizens. This includes security and privacy for the data collected. Testing under the current DTC testing does not provide legislative security or protection under Australian Privacy laws. Data may be shared from the commercial entity to law enforcement, or to biomedical companies. This sharing of data may not be in the interests of client and currently does not assist Australian epidemiology.

Further, a company may provide the initial testing, and results with an offer of additional testing at an additional cost, giving rise to claims of misrepresentation of advertised services. At a minimum, the information provided may not be clear, giving rise to further information seeking by the client within the Australian health services. Further, there is anecdotal evidence the information provided is inaccurate. This may lead to delays in health seeking behaviours and additional charges for further clinical testing.

These concerns have been corroborated by enquiries to Genetic Alliance from clients who have used the current available DTC testing services. These enquiries have resulted from unclear communication about the results of the testing, or requesting more information on the results, as they have been uncertain where to obtain more information. Those enquiring did not know the testing is not done to Australian standards. Further, should they pursue genetic testing in a clinical setting to obtain National Association of Testing Authorities (NATA) testing and accurate information, additional costs could be incurred by the client, as not all testing is included in the PBS listing.

This confirms and highlights two aspects as detailed above. First, the clients are unknowing of the standards and full implications of current marketed genetic testing available. Second, the service providers' omissions with regard to testing standards and lack of educational material and information disclosed by the companies providing this service. Third, there is no enquiry or confirmation by the service provider of the client's health literacy or their understanding of the service they are purchasing.

In addition, there are significant concerns with legal, legislative and privacy. This concerns has been voiced by individuals, condition associations, patient advocacy bodies, academia and others. Currently there is a self-imposed moratorium by the insurance industry not to require disclosure of genetic testing. Any testing, past or future, may or may not be required to be disclosed when this moratorium ceases. Australian Privacy Legislation may not provide coverage on testing conducted and data retained outside Australia. This may leave people open to disclosure by companies to and discrimination by insurance agencies or employers. Further, there exists the potential of commercial on selling of genetic data, with or without personal details, and the scale of this practice is unknown.

Of particular concern is disclosure of personal data to other nations' federal bodies, such as military, law enforcement or security agencies.

**Section 2 -**

**Are there any particular genetic tests that should not be available as a self-test?**

**Genetic Testing for Health Conditions.**

Genetic Tests represent a very broad spectrum of tests from specific gene arrays to whole genome sequencing (WGS). Testing maybe done at different times during a persons' life. This services is currently provided by health professionals.

Genetic testing for conditions currently is facilitated, mostly, by Genetic Counsellors. The consultation process identifies the symptoms of the conditions or reproductive status and testing options. This consultation is especially critical for identifying potential inherited conditions and inheritance patterns. This consultation provides information and analysis which would not be available with a self-test.

For an individual, **Diagnostic Testing**, for a condition may be done as an array or WGS performed to identify a specific genetic condition. This may be for a newborn with an acute condition, for a child to diagnose a health condition, or for an adult. Diagnostic testing may be for an inherited condition or *de novo*, a new genetic variation. Genetic conditions may occur at any stage of life, from newborn, to onset of disease in later life.

Genetic testing may be contemplated in adulthood by an individual considering reproduction and pregnancy. Prior to pregnancy, **Carrier Testing**, may be undertaken. This genetic test is performed for a person to identify if they carry a gene change that may be transferred in the conception process. **Predictive and Pre-symptomatic Testing** are genetic tests performed for a person with a family history of a genetic condition, who does not usually have symptoms at the time of testing, to determine if they have inherited that condition. During pregnancy, **Pre-natal Testing,** through different techniques are genetic tests conducted to identify possible genetic conditions in an unborn baby.

All of these tests have benefits of information, but also of limitations and risks from the procedure. The benefit of information assists with decision making and individual autonomy. Risks include reactions or physical damage or infections as a result of testing procedures, especially some types of testing conducted in pregnancy, on the mother and the unborn foetus or infant. These test require skills, training and resources not available with DTC testing.

Diagnosis of any condition, engagement with the health system for services, treatments and management is subject to the severity of the condition and the autonomy of the individual. Physical health and psychological health services may be required as adjustments to education, social, working and daily life are impacted.

Notably, for inherited conditions, discovery of a genetic condition carries psychosocial impacts to identity and emotional stress that may require management and engagement with mental health services. Of particular concern in the counselling process is the discussion on sharing of genetic information to family members. This can require significant consideration and is very individual for each persons' life experience and upbringing. Inherited conditions not only impact the individual but also the generations above, below and genetically beside the person tested. Family relationships, especially those under strain from many other factors, may not be receptive to disclosure or discussion of a health condition. Diagnosis of a condition, or absence of a condition, may produce additional stresses for all parties, with deep and significant impacts. Genetic testing confirming a positive diagnosis may also impact the life partner of the individual in their day to day living, work life and life aspirations.

Further, genetic testing may provide information that is not congruent with paternity and family narratives. This additional stress for all parties, and may require careful discussion assisted by trained counsellors. This is compounded if inherent conditions are either present or absent.

Complex genetic conditions such as mosaicism and chimerism require additional testing and may alter treatments to provide the best care and health for the individual

Consultation with trained health professionals in genetic variation provides the individual with information on the condition and treatment choices. Other options may be medications available both on or off the PBS listing or for participation in trial for new treatments. This enables autonomy in decision making and ensure the best fit for the person and their life.

## Genetic Testing and DTC Services

The Review does not cover the specific concerns with any medical testing, notably consent and understanding. It is presumed with DTC testing there is literacy to be able to follow instructions and perform the test, read and interpret the results. It does not cover significant concerns of genetic testing with regard to consent, health literacy, CALD communities or Indigenous, and different socio-health norms and practices.

Genetic testing currently is supported by trained health professionals. This provides assurance that testing is understood and consent is received with regard to the long term impacts of genetic information. Genetic information has the potential to be used not only to guide medical treatment for the individual, but also for data collection for health epidemiology. Conditions caused by polygenetic changes or diverse, complex genetic conditions, such as mosaicism and chimerism, cannot be fully or correctly investigated by DTC testing methodology.

Current testing is done under clinical supervision has a mandate of beneficence, autonomy and non-maleficence. These ethical considerations are not in place, nor enforceable with DTC testing. Inappropriate or incorrect testing may be sought in an uncontrolled DTC environment, leading to requests for incorrect medications or misuse of medications. Of special concern is possible abuses in adult/ guardianship to child/ adult minor relationships utilising testing for unethical, immoral or duplicitous purposes.

Provision of professional genetic testing services also enables socio-health concerns to be addressed or remedies sought. Stigma and lack of understanding of conditions can be addressed in a clinical setting. This is especially a concern of health literacy and for CALD communities. Australia's multicultural community is unique and increasing in diversity. Our Indigenous and Torres Strait Islander peoples' health needs have been a priority for Federal government with their 'Closing the GAP' programs. Differing social practices, beliefs and priorities with regard tissue use and disposal to western health norms need to be considered and provided for in Australia's health services. These considerations and needs are not provided in a 'one size fits all' DTC service.

Genetic testing in a clinical setting provides opportunities to collect data, and for data to be retained for analysis and epidemiological studies to address large scale health concerns within Australia. Data on specific rare conditions may be individually low but collectively high, suggesting economies of scale for health services could be introduced. Academia and peak medical bodies are anticipating

personalised treatments, illness preventions and screening programs to be developed from the information genetic technologies can bring. Therefore, genetic data collection for the Australian health context should be encouraged and facilitated. This would not be possible with DTC testing models currently available.

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

As outlined in Section 1, DTC testing is actively sought by the public for a variety of reasons. Genetic Alliance Australia recommends that DTC testing continues with the current available range of ancestry, paternity, athleticism and metabolism with the following recommendations-

These kits are to be sourced from both Australian producers, have NATA testing quality assurance and for data to be retained in Australia.

Marketing of the DTC testing is to branded with NATA accreditation and contain information advising customers of the accreditation, legal provisions for data storage and information of clinical genetic services. This will distinguish product quality from competitors.

If the customer would like a copy of the test results to be included on their e-health record, a suitable report is to be provided for this purpose. Participants may be able to provide their Medicare number for this report to be sent to the Federal Dept. of health and be included in their record.

For the reasons in Section 2, Genetic Alliance recommends DTC testing should not be provided for diagnosis of health conditions requiring prescribed medication, clinical treatment, monitoring and supervision. This is as potential risks for the individual, as yet unborn and families are significant, require complex interventions and support.

Testing for diagnosis, carrier testing, predictive and pre-symptomatic testing are to continue to be provided in a clinical setting. This is to provide treatments and medications, psychosocial supports and opportunities for research participation. Data collection is enhanced with appropriate security and legislative protection.

Prenatal testing for diagnosis while the foetus is in utero, should not be available as over the counter dispersal or online, especially in light of potential risks to the health of the mother and foetus.

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