gsnv
Genetic Support
Network Victoria

22 November 2019

RE: REVIEW OF THE REGULATION OF CERTAIN SELF-TESTING IN VITRO DIAGNOSTIC MEDICAL DEVICES (IVD's) IN AUSTRALIA

SHOULD DIRECT TO CONSUMER GENETIC TEST BE ALLOWED

Thank you for the opportunity to respond to the TGA review.

The Genetic Support Network of Victoria (GSNV) is Victoria's peak body supporting people with genetic, undiagnosed and rare conditions and those who support them.

Established more than 20 years and funded by the Victorian State Government, our vision is for a Victoria where everyone can flourish. We are committed to accomplishing this by empowering through education, advocacy and support, representing, listening, engaging, understanding and facilitating a collective community voice that is clear and focussed on people being able to live their 'best' lives.

As a direct result of our area of expertise and experience, the GSNV has chosen to respond only to the question of *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?

Genetic Tests represent a very broad spectrum of tests including:

Carrier Testing: a genetic test performed on a person to identify if they carry a gene change.

Diagnostic Testing: a genetic test performed on a person to identify a specific genetic condition.

Predictive and Pre-symptomatic Testing: a genetic test performed on 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.

Pre-natal Testing: a genetic test to identify possible genetic conditions in an unborn baby.

Testing may also be carried out using a range of technical application involving single to multiple or all genetic information.

This is a highly complex area with the technology providing significantly more certainty that we used to have but still in many cases predictive rather than absolute results. This is an evolving technology and must be considered so.

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As a result of our experience with health consumers and patient organisations, the GSNV would support the current TGA position with genetic testing excluded from self-testing IVD's under the Excluded Purposes Specifications.

The reasons for this are outlined below:

The efficacy of the Self Tests

Current Direct to Consumer 'genetic testing' such as 23andMe, ancestry.com and others who offer health information options as part of their services are already increasing the burden on GP's, Clinical Geneticists and other health professionals as they present 'test' results that lack efficacy because of the process, are questionable because of the result analysis and interpretation, and over-promise on the accuracy of results.

The choice of genetic testing over other tests and the type of genetic test are the result of consultation between health professionals and the individual and take other factors such as phenotype, utility of the test and likelihood of consequences. Self - sourced testing could not include such a comprehensive process and yet will provide a result.

The consequences of a self-test that offers a result which may over-estimate, be incorrect – offering a positive or negative interpretation can send an individual or a family down a path from which it can have many unintended consequences; psychologically, financially and on family relationships.

Genetic tests impact people's sense of who they are and how they are put together. It is a much more personalised and intimate relationship than a part of the body not working so well or contracting a condition or disease because of an external influence. Access to testing which may impact at such a deep level must be as accurate as possible and supported.

With increasing efficacy of predictive testing for common multifactorial conditions, such as diabetes, obesity and heart disease improves, this may be one area which can be included in self-testing availability in Australia in the future. At this time, results are one factor and genetic data is less predictive than lifestyle and life stage.

Genomic and Genetic Literacy

There is recognition across the health, community service and government sector that genomic and genetic literacy is low across the Australian general public. This is a relatively new and constantly changing technology and practice.

We know that people who find themselves in the health system quickly develop literacy in the specific areas pertaining to themselves. This generally grows from self-driven internet searches, access to health professionals and support organisations and connection to others facing similar or the same experience.

Each of these sources of information can be inconsistent, inadequate or simply not current. It is usually a combination which assists an individual or family to understand, find their way and become empowered to make their own health decisions.

Self-testing would put significant pressure on these information channels from tests that are presented as conclusive and yet are not.

There is already a tendency for the Australian public to view genomic and genetic technology to always and in every case, provide an absolute truth. This is true of ancestry.com and others as well as results from an accredited laboratory. The public does not tend to discriminate. This may damage the credibility of genetic and genomic testing over time.

The future implications of receiving a result from a genetic test that is predictive of future health conditions may include access to life insurance. Whilst there is a moratorium regarding the use of genetic test results to determine life insurance access and premiums, this may not continue. The Australian consumer is not generally aware and often not made aware of these potential unintended consequences.

Interpretation of Test Results

We are already experiencing members of the public who have purchased self-testing kits and received results from overseas seeking to access health professionals to interpret results, to explain terminology, to tell them what they need to do next.

The skill and capability to analyse test data and interpret test results is highly valued and developed through specialised training and experience. This is acknowledged by policy makers and health systems alike across global health systems. By restricting how tests are accessed, we are also ensuring that interpretation is consistent, underpinned by quality, validated by other experts where relevant and provided in an accessible and actionable format. These controls would be difficult to enforce through a self-testing regime.

Support for individuals and families

The clinical process for genetic testing includes counselling options through all steps of the process. Prior to testing, a consent process taking individuals through the risks, benefits and potential outcomes and consequences is completed. While this may be completed online in some cases, options for access to a health professional for clarification or discussion is offered. This is critical to manage the expectations of the individuals or families. No genetic test can occur without a clear and guided consent process.

Self-testing may provide the same opportunity for an individual to review risks, benefits and potential outcomes and consequence but this then places the onus firmly with the individual to do so rather than a health practitioner who understands the importance of being familiar with these areas.

Results from genetic tests – self testing or not can have impacts on individuals, families and extended families. Results can range from inconsequential to life-changing. A no result can be as impactful as a clear result. Individuals and families need support from individuals that can include health, mental health, services, financial, peer connection and more. These support decisions are most often made in partnership between the health professional explaining the test results, individuals and families. They are critical to the wellbeing of the individual and their family.

Pressure on the health system

This is another of the reasons that testing cannot be made available as a self-administered test. Any result that provides uncertainly will cause individuals to seek further information. We have evidence that individuals will seek out assistance from already overburdened health services where interpretation from self-tests is not clear to them or they don't know what to do with results.

People approach health services anticipating an easy solution and are often greeted with options that involve taking a new test (at a much higher cost usually) or disappointment that clarity cannot be provided due to the efficacy of the test undertaken. Significant stress and anxiety have usually been experienced by the individual already at this point. This places further pressure on the health practitioner to find a solution for the individual.

The generation of data, data security and ownership

Self-testing already is generating large amounts of data and databases which can be used for commercial, research and population based purposes. Australia is yet to have a meaningful conversation about the ownership of such data generated in Australia, ownership of how it may be used and its security.

Whilst this is a 'user-pays' risk, the implications include a much wider risk where Australian consumers expectation standards that if not met, will impact the view of health consumers in the wider research and clinical trial arena and the credibility of health data security.

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

The GSNV recommends that the current status is maintained.

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?

It is difficult to see how the risks to consumers could be mitigated as much of the risk stems from the science of genomics and genetics itself.

The risk is in creating consumer expectations that cannot be met with certainty at this time and then in that environment, not providing access to support to assist

people to find the best pathway and services for them to navigate through the uncertainty.

Conclusion

The significant difference between genetic tests and the other tests identified as potentially having their status changed by this review are the benefits that can be assumed for the others including:

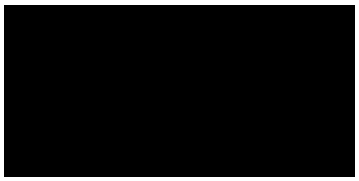
- that self-testing will take pressure off the health system because people will rule themselves out of having conditions
- people where self-testing confirms a condition or the potential for a condition will seek assistance and treatment earlier
- earlier treatment may influence the progression of the condition and alleviate costs in the health system in servicing chronic conditions

None of these are true for genetic testing or where they are, the support required pre and post testing is significant. For example, a test for a single gene condition where the efficacy is strong will require significant support in the case of a positive result.

Predictive genetic testing for common multifactorial conditions, such as diabetes, obesity and heart disease will improve over time and this may be one area which can be included in self-testing availability in Australia in the future. At this time, in these areas, genetic data is less predictive than lifestyle and life stage.

I would welcome the opportunity for further discussion, thank you for the opportunity to contribute.

Yours sincerely,



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