

Standing Committee on Screening

Genomic Tests in Population Based Screening Programs: Statement

Background to Statement

This Statement is intended to provide detail about the potential application of Australia's *Population Based Screening Framework* (the *Screening Framework*) to proposals for genomic tests in population based screening. This Statement may be of assistance to individuals and organisations developing proposals for such screening; and to government departments and other funding bodies when assessing such proposals.

The *Screening Framework* was developed by the Standing Committee on Screening, based on World Health Organization principles of early disease detection. The *Screening Framework* sets out the principles for the implementation and management of screening programs, and the specific criteria for assessing when population-based screening should be offered or a population based screening program introduced. The *Screening Framework* notes that the Standing Committee on Screening maintains a watching brief on the use of genetic and genomic technologies and their potential for application in screening at the population level.

The COAG Health Council's *National Health Genomics Policy Framework 2018-2021* provides an overview of current and potential use of genomics in the Australian health system. This Statement is consistent with the *National Health Genomics Policy Framework 2018-2021*, and uses the term "genomics" in the same manner. Specifically "genomics" refers to both the study of single genes (genetics) and the study of an individual's entire genetic makeup (genome) and how it interacts with environmental or non-genetic factors. In this Statement it is understood that there are multiple types of genomic testing, including tests involving individual genes, gene panels, clinical exomes and the whole genome.

The Standing Committee on Screening endorses the three core principles that underpin the *National Health Genomics Policy Framework*, which are:

- The application of genomic knowledge is ethically, legally and socially responsible, and community trust is promoted;
- Access and equity are promoted for vulnerable populations; and
- The application of genomic knowledge to health care is supported and informed by evidence and research.

The Standing Committee on Screening is aware of current genomic technologies and overseas genomic screening programs, and the use of commercial genomic tests in Australia. The Standing Committee on Screening is committed to an open dialogue on this topic.

Genomic Tests in Population Based Screening Programs: Statement

The Standing Committee on Screening recommends that any proposal to undertake genomic testing in a current or new population based screening program should demonstrate compliance with *the Population Based Screening Framework* and World Health Organization principles of early disease detection. In this statement, references to screening programs and genomic testing are made in the context of population based screening programs, and should not be extrapolated to non-population based clinical settings. Key considerations relevant to genomic testing are presented below:

1. Condition

- 1.1. The condition is an important health problem and has a recognisable genomic cause or risk.
- 1.2. The clinical significance of a genomic variant/s should be adequately understood. The relationship between the genomic cause or risk, the clinical condition, and the opportunity for intervention must be demonstrated.
- 1.3. The use of genomic technology in screening programs must be in response to a need for early detection of disease, disease risk, or disease carrier status, and not be driven by technological advances.
- 1.4. Conditions screened for in children using genomic tests should be limited to those with childhood onset and (i) acceptable treatment options, or (ii) an understood benefit to the child and/or their family through the disease diagnosis, or through knowledge of increased risk of developing the disease.

2. Test

- 2.1. The “test” refers to the method or procedure used to obtain and analyse a sample of genomic material. For example, a blood test, saliva test, skin scrape, a hair root sample etc. The test must be ordered through a diagnostic laboratory accredited and validated to the appropriate national and international standards of human pathology testing.
- 2.2. The test should be acceptable to the target population and to society.
- 2.3. The test must have clear criteria for positive, negative and indeterminate test results. For screening tests that identify disease or disease risks, there should be an agreed policy on whom to categorise as “screen positive”, “screen negative” and “screen indeterminate”. For screening tests that determine risk of disease, there should be an agreed policy on what determines the degree of risk and how to report or define this.
- 2.4. Screening program data and reports need to be easily understood. Programs should ensure that test result formats and language promote clear and unambiguous interpretation of results, including where results are reported to non-genetic health care professionals, or to consumers.

- 2.5. Laboratories using next generation sequencing technology for population screening must limit analyses to only those genes relevant to the screening program. Incidental genomic findings should not be sought or reported without the explicit advance consent of participants.
 - 2.6. Consideration should be made of the **acceptability** of the test in Australia, including the acceptability of the test for culturally and linguistically diverse groups, Aboriginal and Torres Strait Islander peoples, and within vulnerable populations such as those with a disability or those with the condition.
 - 2.7. Consideration should be made of **equity of access** to the genomic screening test for culturally and linguistically diverse groups, Aboriginal and Torres Strait Islander peoples, and within vulnerable populations such as those with a disability or those with the condition.
 - 2.8. The program design, including the test must account for the genetic background of the target population.
3. *Assessment*
- 3.1. There should be a defined assessment process for people with positive and indeterminate test results, following disclosure of screening results.
 - 3.2. Screening programs must make available adequate and appropriate genetic counselling, giving consideration to family and community contexts when applicable, and ensuring cultural appropriateness including interpreters when needed.
4. *Treatment*
- 4.1. There should be an understood benefit to screening for the condition (e.g. prevention, treatment, family planning, or reduction of length of time to diagnosis) that forms part of a coherent management strategy.
5. *Screening program*
- 5.1. The screening program will be conducted to the highest ethical standards, such as those articulated in the National Health and Medical Research Council (NHMRC) *National Statement on Ethical Conduct in Human Research* and the NHMRC *Ethical conduct in research with Aboriginal and Torres Strait Islander Peoples and communities: Guidelines for researchers and stakeholders*. Where screening programs do not contain a research element, the ethical standards set out in the NHMRC documents above should apply. These ethical standards are in conformity with the aims and principles of the United Nations Educational, Scientific and Cultural Organization (UNESCO) *Universal Declaration on the Human Genome*, and the UNESCO *International Declaration on Human Genetic Data*.
 - 5.2. The screening program should target those most likely to benefit, and clearly define the target population. Genomic screening tests used within the

screening program should be suitable for all individuals screened, with a standardised test applied to the target population. The screening program should promote equity among all Australians.

- 5.3. The entire screening program, including the genomic testing component, must be **acceptable** to the target population and society including culturally and linguistically diverse groups, Aboriginal and Torres Strait Islander peoples, and within vulnerable populations such as those with a disability or those with the condition.
- 5.4. Aboriginal and Torres Strait Islander stakeholders must be included at the earliest possible point in new screening program development. It is essential to ensure that Aboriginal and Torres Strait Islander peoples can be part of the planning and development of the screening program. It is also essential that any screening program involving Aboriginal and Torres Strait Islander participants or communities maintains communication with participants throughout the program, provides outcomes from the program back to participants and includes Aboriginal and Torres Strait Islander peoples in program evaluation.
- 5.5. Consideration must be given to ensuring **equity of access** to the screening program, for culturally and linguistically diverse groups, Aboriginal and Torres Strait Islander peoples, and vulnerable populations such as those with a disability or those with the condition.
- 5.6. Any genomic screening should be part of an integrated screening program that includes education of the target population and the health workforce. It should promote equity of access by the target population, have appropriate infrastructure including a register for people screened, access to appropriate clinical services provided by an appropriately skilled and resourced workforce, and program management. When the screening program includes Aboriginal and Torres Strait Islander participants, cultural competency training should be provided for program staff and associated health workforce.
- 5.7. Nationally acceptable program parameters must be available, including: guidelines and educational material on the identification of people suitable for genomic testing, a clear management plan for positive, negative and indeterminate results, appropriate patient information and support including genetic counselling, and on follow up investigation and medical care.
- 5.8. There should be evidence of screening program acceptability, effectiveness and appropriateness before a program is introduced and any benefits should outweigh potential harms, including psychological, physical and social harms.
- 5.9. Evidence of value for money or cost effectiveness for the program should be demonstrated.

- 5.10. “There should be quality assurance incorporated at all levels of the screening program and ongoing program evaluation should be planned from the outset”¹.
- 5.11. Provision must be made for an appropriately skilled and resourced health workforce, including adequate and culturally appropriate support, mental health services and counselling for people undergoing genomic tests, and ongoing education for health professionals on the screening process and outcomes from it. The health workforce should include members of culturally and linguistically diverse groups, Aboriginal and Torres Strait Islander peoples, and vulnerable populations. The health workforce should recognise that Aboriginal and Torres Strait Islander participants may have previous multiple traumas.

Consent

- 5.12. Consent processes must be clear, unambiguous and frank. Consent processes must reflect informed choice, cultural appropriateness for the target population, confidentiality, respect for autonomy and participant understanding of possible discrimination based on genomic data, and participant understanding of the possible outcomes of genomic testing. Consent processes must also acknowledge participants’ ability to consent, and clearly identify when substitute consent has been provided, including in particular, for children.
- 5.13. Consideration should be given to when and how consent is sought from participants, and how participants can decline consent to part or all of the program. For example, whether separate consent processes could be developed for consenting to program participation, for consenting to the use and retention of genomic material or data collected via the program for research purposes, and for consenting to receive future findings; or whether flexible, patient-centred consent processes could be used for obtaining consent from participants as circumstances change.
- 5.14. Consent processes should be developed for screening programs, and should be tailored to the specific program. Consent processes must include protocols for disclosure of sensitive genetic information including familial or community risk, consideration of an individual’s decision not to be informed of specified genomic information, information on the potential inclusion of test results in research, and storage and destruction of genomic material and data. Where a secondary use of data is envisaged, for example, future screening activity and research, this should be clearly articulated in consent protocols. Consent processes should consider the possibility of re-testing samples collected, and establish protocols for re-testing if this is likely, taking into account the cultural

¹ Andermann, A., Blancquaert, I., Beauchamp, S. and Costea, I. (2011) “Guiding policy decisions for genetic screening: developing a systematic and transparent approach.” *Public Health Genomics*, Vol 14, pp9-16.

and religious beliefs of participants on retention and destruction of genomic samples.

- 5.15. There are Commonwealth, State and Territory laws, that apply to stakeholders, that specify how stakeholders can collect, use, store and disclose participants' personal information including health information. Stakeholders must abide by these laws, including the requirement to provide participants with privacy notices. Privacy notices address issues such as identifying those circumstances where stakeholders intend to disclose participants' personal information without obtaining participant consent. For example, where the stakeholder is authorised by law to disclose personal information, including under court order. These circumstances may vary with each program and the privacy obligations imposed upon stakeholders can vary across jurisdictions.
- 5.16. Informed consent to participate in the screening program would usually require information about the availability of appropriate treatment and/or condition management pathways.

Management of data

- 5.17. Planning should consider the possibility of re-testing stored genomic material and/or re-analysis of genomic data in light of advances in technology or knowledge, including how future findings will be notified to participants and/or other family members or descendants, and how program data could be used in research.
- 5.18. Screening program data management planning must reflect the informed consent of participants, and take into account participants' cultural and religious beliefs on retention and destruction of genomic samples. Informed consent must be obtained for analysis of data, storage and re-testing of stored materials and mechanisms to contact parties with information on the findings.
- 5.19. Consideration of data ownership, ownership of genomic material, and equity of ownership should be made, including interests and rights of Aboriginal and Torres Strait Islander peoples, and whether individuals screened can access data for other purposes. Data access and management should be culturally appropriate.
- 5.20. The program must ensure suitable data and sample storage with a high degree of security. Policies and procedures for access and sharing to data and samples must demonstrate compliance with national standards. Data sharing and reuse must align with Aboriginal and Torres Strait Islander cultural values as relevant to Aboriginal and Torres Strait Islander program participants.
- 5.21. Genomic population screening registers will need protocols for management of data and samples, relating to storage, sharing, access rights, research requests, and possible notification of new findings to participants in population health screening programs.

6. Ongoing management

- 6.1. Policies must be in place for potential future recall of participants affected by new testing or research findings, including transient populations. Note that this may require consideration to gain additional informed consent by participants.
- 6.2. Ideally, post-diagnosis support and advice to participants on the management of the genomic condition/s should be consistent across jurisdictions.

7. *Carrier screening*

All the above guidance (1-6) applies to carrier screening. Additional considerations also apply, described below.

- 7.1. Carrier screening is appropriate for family planning, to assist in identifying the risk of having offspring with inherited genetic diseases. Individuals who are carriers of some inherited diseases may also have a higher risk of some health conditions, and carrier screening is also appropriate in those circumstances.
- 7.2. For carrier screening, consideration should be made of the potential implications for people other than the individual screened, including consideration of incidental findings. Consideration of the risks and benefits for family members and community members should be undertaken.
- 7.3. Decisions to screen individuals for carrier status of inheritable diseases should be made on the basis that screening will provide actionable information for individuals, families or communities that assists them in making informed choices.

The NHMRC National Statement on Ethical Conduct in Human Research

The Standing Committee on Screening endorses and acknowledges the NHMRC *National Statement on Ethical Conduct in Human Research* (2007 updated 2018), and the provisions relevant to consumers participating in genomic research as part of genomic screening.

UNESCO Universal Declaration on the Human Genome

The Standing Committee on Screening notes that the ethical principles set out in the UNESCO Universal Declaration on the Human Genome and Human Rights are in conformity with the NHMRC *National Statement on Ethical Conduct in Human Research*.

UNESCO International Declaration on Human Genetic Data

The Standing Committee on Screening notes that the principles set out in the UNESCO International Declaration on Human Genetic Data are in conformity with the NHMRC *National Statement on Ethical Conduct in Human Research*.

NHMRC Information for consumers

The Standing Committee on Screening endorses the National Health and Medical Research Council information resources for consumers about genetic testing:

- *Medical Genetic Testing: Health information for you and your family* (2012).
- *Personalised medicine and genetics* (2013).
- *Understanding Direct-to-Consumer Genetic DNA Testing: An Information Resource for Consumers* (2014).

Individualised advice can be obtained through discussion with a general practitioner or referral to a clinical genetics service. A list of genetics services can be found at the Centre for Genetics Education webpage: www.genetics.edu.au/genetic-services