

# DNA Screen Study Participant Information Sheet

This Participant Information Sheet has been provided to help you decide if you would like to participate in the DNA Screen study. Please read it carefully.



# What is the purpose of the DNA Screen study

DNA Screen is a research study. It assesses a new approach of offering preventive genetic testing to young adults. This will identify individuals who have inherited a high-risk DNA variant which increases the risk of certain types of cancer and heart disease. There are proven methods to reduce the risk of and/or prevent these conditions. The aim of the study is to increase access to DNA testing for young people who would not otherwise know they have an increased risk of developing these conditions. Combined, these conditions affect approximately one in 75 people. The study will test 10,000 people between the ages of 18–40 years. Most people tested through this study will not have one of these DNA variants and will not have this increased risk.

## Who is organising and funding the DNA Screen study?

The study is led by Monash University, School of Public Health and Preventive Medicine, in Melbourne. The chief investigator is Associate Professor Paul Lacaze, Head of Public Health Genomics at Monash, who is supported by a team of researchers and healthcare professionals from around Australia. The study is funded by the Australian Government's Medical Research Future Fund, Genomics Health Futures Mission. The study has been approved by the Alfred Hospital Ethics Committee and will be carried out according to the National Statement on Ethical Conduct in Human Research (2007).

## What does the DNA Screen test screen for?

The DNA Screen genetic test detects DNA variants in specific genes, that cause a high genetic risk of developing these three conditions:

**i) Hereditary breast and ovarian cancer (HBOC)** - results in a high risk during a person's life for breast and ovarian cancer in women and prostate cancer in men. The names of the genes DNA Screen will test for are BRCA1, BRCA2, PALB2 and a single variant in ATM. For more information, see [here](#).

**ii) Lynch syndrome (LS)** - results in a high risk during a person's life high for bowel (colorectal) cancer in both men and women, and endometrial cancer (lining of the uterus) and ovarian cancer in women. The names of the genes DNA Screen will test for are MLH1, MSH2, and MSH6. For more information, see [here](#).

**iii) Familial hypercholesterolemia (FH)** - results in an increased risk during a person's life for heart disease or stroke due to high cholesterol from an early age. The genes DNA Screen will test for are *LDLR*, *APOB* and *PCSK9*. For more information, see [here](#).

For these conditions, effective, proven risk-reducing interventions are available for prevention and/or treatment of the condition. Together, these conditions affect one in 75 people. However, they only account for a small proportion of all known cancers and heart disease. Even if your DNA Screen test does not find any of the DNA variants we are testing for, it does not guarantee you a clean bill of health. You can still develop cancer or heart disease.

The one in 75 people identified with the DNA variants are at high risk of disease. However, the DNA variant alone does not guarantee they will develop cancer or heart disease. The DNA variant is a risk factor, not a diagnosis. Various other factors also contribute to risk (such as environmental, lifestyle and other genetic factors). If you have one of these DNA variants, we will provide you with genetic counselling to further explain your risks and preventive options, and to arrange referrals to specialist health professionals as required.

## What are the possible results from the DNA Screen test?

Most people who have testing through DNA Screen will receive a result that they have NOT inherited one of these DNA variants. This result means that the person does not have a high genetic risk of developing the conditions associated with these genes. However, it does not guarantee a clean bill of health. A person can still develop cancer or heart disease.

A small number of people (one in 75) who participate in the DNA Screen study will receive a result that they have inherited one of these DNA variants. Inheriting one of these gene changes does not mean the person will develop the condition. Rather, it increases their risk of developing the condition. For all of these conditions, there are effective proven ways to reduce the increased risk and, in some cases, to prevent the condition.

The DNA Screen test will not predict all future health issues. DNA Screen will not test for paternity or non-health-related information, such as your family ancestry.

If we find you have not inherited a DNA variant, you will be notified by SMS/email. You will receive a report about your results with links to further information, and you can speak with a genetic counsellor from our team about your results if you have further questions.

**Follow-up:** A member of the research team will send you a questionnaire or contact you directly to gather information about your experience. We may also gather information from your medical records for the study or seek medical information from your doctor(s), especially if you have inherited a DNA variant.

## What are the benefits to me?

- DNA results can inform you of important health risks for you and/or your family members. This information can be life-saving, and empower you to take steps to prevent health problems or detect and treat them early.
- DNA testing may also help you access support and plan for the future.
- DNA results may also provide information about the genetic risks for your blood relatives. Where appropriate, they may be used to help inform testing of your family members. Your blood relatives will not be tested as part of this study, but may be eligible for testing through the public healthcare system.

## Are there any risks to me?

There are no significant risks to you if you take part in the study. DNA testing and discussion of the results can be, in the short term, upsetting for some people and their families. There are no studies that show long-term harm from DNA testing. Our study will also provide support and information to you through access to trained genetic counsellors.

Australia. However, DNA results can be used by life insurance companies at the time of underwriting risk-rated policies (e.g. death, income protection, and permanent disability insurance). This is only relevant if you wish to apply for such policies after undertaking DNA testing. There is a partial ban (moratorium) on life insurance companies, where results are not required to be disclosed for policies up to certain financial limits. For more information about this, you can access a fact sheet [here](#) or contact our study team. However, we cannot provide financial advice. If you remain concerned about insurance implications after reviewing this information, you can contact a financial advisor to discuss your options before proceeding with testing.

# Who can take part in the DNA Screen study?

The DNA Screen study is designed to be as inclusive as possible. **Registration** is open to any Australian citizen or permanent resident aged 18–40 years who has not previously had a genetic test for the three genetic conditions that we are looking for.

Please note that **registration does not guarantee participation in the study**.

Currently, the study is limited to 10,000 people, meaning not all people who register for the study will be able to take part. We hope to raise more funding to screen more people in the future. If you are selected to participate, we'll send you a separate link via email/SMS to ask you to consent electronically. Once you have completed the consent form, we will send you a saliva DNA collection kit.

# What steps are involved in participating in the study?

1. **Registration**: check your eligibility and register your interest via the Registration webpage.
2. **Read this information document and watch the video**. Contact the study team if you have any questions.
3. **Provide consent**: If you are selected to participate, we will send you an invitation link via email/SMS and ask you to complete an electronic consent form.
4. **Collecting a sample**: After completing the consent form, we will send you (via post) a box containing the saliva collection kit. This kit is approved in Australia for the collection of DNA through a saliva sample and has instructions about how to provide your sample. The box is Reply Paid, so you can post your sample back to our lab at Monash University free of charge. We will then test your DNA and securely store your sample (see Section 8).
5. **Receiving results**: If we find you have inherited a DNA variant, a genetic counsellor will contact you by phone to explain the result and discuss your options. The genetic counsellor will arrange a referral to an appropriate specialist health professional for further advice and ongoing care. A blood test will be required to confirm your initial result. There will be no out-of-pocket costs for tests or appointments associated with the study or any ongoing care.

## Your Consent

If you are selected and agree to participate in DNA Screen, you will be asked to give your consent. This will indicate that you have read and understood this information and that you freely agree to take part in the study as described. This involves giving permission for us to:

- collect and store your personal information in our research database (e.g. contact details, answers to health-related questions).
- undertake DNA screening on DNA from your saliva sample for the DNA variants.
- provide your contact details and DNA result to genetic counsellors from our study team.
- provide you with your results.
- notify your GP or other healthcare professionals of your results.
- collect electronic copies of your past and future health records (e.g. we may send your identifying details to other organisations to obtain this information).
- contact you to answer follow-up research questions

Optional permissions that you can choose to say Yes or No to include:

- permission to re-screen your sample in the future for additional DNA variants that may be relevant for your or your family's health. This might happen if new knowledge becomes available, and if additional funding can be raised, but is not guaranteed.
- undertake other types of research on your sample or data (beyond DNA Screen testing).
- be invited to be part of future research studies.
- destroy your sample and associated data after completion of the DNA Screen project and after the laboratory's legal obligation to retain the sample has lapsed.

If you provide consent (optional), your information and/or sample, which will not include your name or other identifying information, may be made available to other researchers. This may include interstate and overseas researchers, and would only be for studies approved by a Human Research Ethics Committee that is recognised by Monash University.

DNA test results and health data may also be shared with national or international databases. This would help improve our understanding of human genetics by comparing your results to those from other people. All identifying information will be removed from these data.

We will not share or provide access to your sample or data without your express consent.

## How are my data, privacy and confidentiality protected?

We have taken significant steps to prioritise the security and privacy of your sample and data.

Monash University is subject to strict privacy and health records laws which regulate how it collects and handles personal information, including sensitive and health information. All information collected as part of this research study will be stored in secure, controlled-access databases that meet national and international data standards. Security measures will help prevent unauthorised access to or misuse of the data. Information that identifies you, such as your name, date of birth and address, will be stored on a password-protected computer system, and restricted to study staff.

In accordance with applicable Australian health privacy legislation, you have the right to access information about you that is collected and held by us. You also have the right to ask for information to be corrected. Contact the study team on 1800 518 146 or [DNA.Screen@monash.edu](mailto:DNA.Screen@monash.edu) if needed.

## Where will my sample be stored?

Your sample will be securely stored in the [NATA-certified Biobanking Victoria facility at Monash University](#).

## Donating your data and sample(s) for other research studies (optional)

If you provide consent (optional), your information and/or sample, which will not include your name or other identifying information, may be made available to other researchers, including interstate and overseas researchers, for studies that have been approved by a Human Research Ethics Committee recognised by Monash University. DNA test results and health data may also be shared with national or international databases, to improve understanding of human genetics by comparing your results to those from other people. All identifying information will be removed from these data. We will not share or provide access to your sample or data without your express consent.

## Publication of results

The results of this research study may be published and/or presented at scientific and medical meetings. In any publication and/or presentation, data will not identify individual study participants

## What if I no longer want to participate?

You can withdraw from the study at any time, without giving a reason, by contacting the study team on [1800 518 146](tel:1800518146) or email [dna.screen@monash.edu](mailto:dna.screen@monash.edu). If you no longer want your information or samples to be used, all identifying information will be erased and your sample will be physically destroyed.

However, please note that research may have already taken place using your data, and this research and any publications based on it cannot be undone.

If you have any concerns or complaints about the conduct of this survey, you can contact:

### **Reviewing HREC Office/Complaints contact person**

Position: Complaints Officer, Office of Ethics & Research Governance, Alfred Health

Telephone: (03) 9076 3619

Email: [research@alfred.org.au](mailto:research@alfred.org.au)

Please quote the following Project ID number: 80094