Barossa Family Heart Study

PATIENT INFORMATION SHEET
for genetic testing (18yrs & older)

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You are participating in the BAROSSA FAMILY HEART STUDY because you have, or someone in your family has, been diagnosed with familial hypercholesterolaemia (FH). Before you decide whether or not you agree to the DNA analysis part of this program, you should read this Information Sheet. This document contains information about the purpose of the DNA analysis part of the BAROSSA FAMILY HEART STUDY.

Please read the information carefully. If you have any questions about the information provided please contact the health professionals listed at the end of this document. If you decide to have DNA taken, you will be asked to sign the separate Informed Consent Form. By signing the Consent Form, you indicate that you fully understand the information provided and that you consent to participating in this part of the program. You will be given a copy of the Information Sheet and Consent Form to keep for your records.

WHAT IS FH?

FH is an important and under-diagnosed health problem that affects 1 in 500 people leading to high blood cholesterol levels and premature heart disease. It is known that a gene (DNA) for the LDL receptor plays a major role in FH. This means that the risk of developing premature heart disease from FH occurs frequently in a family and may be passed from parents to on average half of their children. Recent advances in science allow doctors to study the LDL Receptor gene in the families of people with FH. Faulty genes may be identified thereby allowing early detection and effective treatment of FH before symptoms develop. DNA tests may also identify other genetic factors that increase the risk of developing heart disease. Laboratory investigations may also lead to the development of better methods of detection, treatment and prevention of FH. Please see the back page for an explanation of terms.

WHAT IS REQUIRED?

If you agree to the DNA test for FH, we will take about 20 mL (about 4 teaspoons) of blood from your arm. The sample is processed so that DNA can be separated from your blood, stored, and

used for laboratory analysis. Once your DNA is analysed for FH and reported, your blood sample, or materials derived from it will not be labelled with any part of your name and cannot be directly linked to you in any way (de-identified). Your blood, DNA samples and the information you give us will be stored separately using a code, without your name and other identifying information. However, the study doctor and staff involved in this study at Prince of Wales Hospital will be able to identify you by your code.

CONFIDENTIALITY

All medical and genetic information is kept confidential to the extent the law allows. However, absolute confidentiality cannot be guaranteed. Information about you **will not** be given to insurance companies, your (future) employers, or be used for any purposes other than those described in this agreement. Information may be made available to clinicians, clinical researchers, general practitioners and scientists. Published information will not divulge your identity.

The Griffith University Research Ethics Committee may have access to your records to ensure that your rights are being properly protected.

Because other family members will be invited to participate, it is possible that we may find out personal information that you or your relatives do not know or do not want others to know. For example, if we find out that someone's mother or father is not their biological parent, we will not tell the person concerned or any other family member under any circumstances. It is important to note however, as with all health information kept about you, that there may be circumstances where disclosure of your health information will be required by law, for example, as a result of a court order.

RESULTS

Knowing or not knowing your results may be relevant to questions asked about your health by your insurance company or your (future) employer. In those circumstances you would need to decide whether you disclose the information.

OWNERSHIP & RIGHT OF WITHDRAWAL

When you provide a blood sample, you will no longer own it or the genetic materials obtained from it. However, you have the right to request at any time that your blood sample and genetic materials be destroyed. You can do this by writing to the BAROSSA FAMILY HEART STUDY and to the doctor or staff who asked you to participate in the study.

Participation in this part of the BAROSSA FAMILY HEART STUDY is entirely voluntary. Your decision not to participate at any time will be respected. You may withdraw your consent at any time. Doing so will not affect your treatment at Prince of Wales Hospital, or your relationship with Prince of Wales Hospital.

It is possible that information or materials from this program might be used to develop products that have commercial value. If this happens, you will not receive any share of the profits.

POSSIBLE BENEFITS

You and your family's participation in this program will assist in the investigation of the underlying genetic defect causing FH. By participating in this program, you may be able to help doctors to

develop better detection methods or treatment thereby preventing the premature heart disease known to be associated with FH.

POSSIBLE HARM

Blood sampling will be performed by experienced professional laboratory staff. It carries a small risk of discomfort and bruising at the site of blood withdrawal. DNA testing carries potential risks of policy loading by insurance companies if the subject is felt to be at higher risk, and paternity issues. The former is dealt with by negotiation with insurance companies on the basis of controlling cholesterol levels, which reduces risk accordingly. Paternity issues are not relevant to the BAROSSA FAMILY HEART STUDY as DNA will not be available for this purpose. Statutory or contractual duties may require participants to disclose the results of genetic tests or analysis to third parties (for example, insurance companies, employers, financial and educational institutions), particularly where results provide information about health prospects. Our research is designed to minimise any resultant risk that participants will be deprived of benefits available to others in the community.

FUTURE RESEARCH

You will also be asked to indicate on the Consent Form if you agree to allow your DNA to be used for future research projects, subject to approval by the Griffith University Research Ethics Committee. With your permission your blood samples may also be sent to participating laboratories both in Australia and overseas so that they too can assess the genetics of FH, its diagnosis and most effective treatment. Because of the sensitive nature of genetic materials, guidelines have been developed to protect your privacy.

Indicating "yes" now does not affect your choice to say "no" at a later date. All contact with you will be made by BAROSSA FAMILY HEART STUDY staff and at no time will your details be given to other agencies without your consent.

The conduct of this research involves the collection, access and / or use of your identified personal information. The information collected is confidential and will not be disclosed to third parties without your consent, except to meet government, legal or other regulatory authority requirements. A de-identified copy of this data may be used for other research purposes. However, your anonymity will at all times be safeguarded. For further information consult the University's Privacy Plan at http://www.griffith.edu.au/about-griffith/plans-publications/griffith-university-privacy-plan or telephone (07) 3735 5585.

All research on de-identified DNA samples collected from this program will be carried out according to the Declaration of Helsinki. The Griffith University Research Ethics Committee will determine whether or not such future research may proceed.

COSTS AND PAYMENT

No costs will be incurred by participants, including DNA testing, other than usual costs associated with travel to pathology laboratories or to GP surgeries, which will not be reimbursed. Costs may be involved if your doctor refers you elsewhere for further investigations or treatment.

No payment will be made for participation; it is for the benefit of the community and the participant.

OVERALL RESULTS

Participants who wish to receive the overall results of the overall research will be sent a letter, fax or email with an appropriate and timely summary. This will take place after publication of the results of the overall study in a peer-reviewed medical journal (usually within 12 months after completion of the study).

FUNDING

The BAROSSA FAMILY HEART STUDY is funded jointly by Griffith University (Queensland), the University of Western Australia, and Merck Sharp and Dohme (Australia).

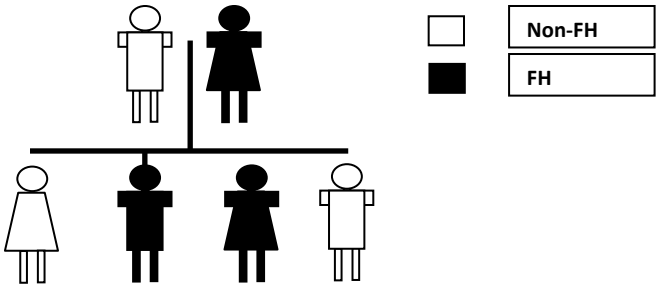
QUESTIONS

Griffith University conducts research in accordance with the National Statement on Ethical Conduct in Human Research. If potential participants have any concerns or complaints about the ethical conduct of the research project they should contact the Senior Manager, Research Ethics and Integrity on 3735 5585 or research-ethics@griffith.edu.au.

PRIVACY STATEMENT

The conduct of this research involves the collection, access and / or use of your identified personal information. The information collected is confidential and will not be disclosed to third parties without your consent, except to meet government, legal or other regulatory authority requirements. A de-identified copy of this data may be used for other research purposes. However, your anonymity will at all times be safeguarded. For further information consult the University's Privacy Plan at http://www.griffith.edu.au/about-griffith/plans-publications/griffith-university-privacy-plan or telephone (07) 3735 5585.

YOUR TEST RESULT EXPLAINED



- A **POSITIVE** test result indicates that I have a gene change (mutation). This means that I have an increased risk of FH and my child has a 50% chance of inheriting the mutation from me.
- A **NEGATIVE** test result indicates that if a mutation **HAS** already been found in an affected family member, I have not inherited this particular gene change and cannot pass it on to my children.

- An **INCONCLUSIVE** result indicates that a mutation **HAS NOT** been found in an affected family member and is therefore not informative. This is because the test can not detect all possible mutations.
- The test result cannot accurately predict the age of onset or severity of FH that may develop.
- Tests of one individual can change the estimation of risk for other family members.
- The test result may affect the ability to obtain some types of insurance, for example, life.
- Genetic counselling will be available for me and other family members during the testing process and after the test result has been given.

Explanation of Terms:

- Hypercholesterolaemia: is the clinical description of the disorder where only cholesterol and not other types of fat are present at above normal levels in the bloodstream.
- Familial hypercholesterolaemia: is inherited and passed from generation to the next.
- Genes associated with familial hypercholesterolaemia: Specific genes in which changes (mutations) are associated with an increased risk of high blood cholesterol levels.
- Gene tests involve analysis of one or more of those genes to determine whether a mutation is present.
- Mutation: Change in the normal DNA code which may cause disease.
- DNA (Deoxyribonucleic acid): The chemical compound of which genes are made.
- LDLR gene (NM 000527.2): a gene coding for a protein that clears cholesterol from the bloodstream.
- APOB gene (NM 174930): a gene coding for a protein particles that carry cholesterol in the blood stream (also commonly referred to as “bad cholesterol”).
- PCSK9 gene (NM 0003841.1): a gene coding for a protein involved in the metabolism of cholesterol in the liver.

Please keep this information for future reference