The genetics of running-induced injuries involving tendon and bone – genetic study

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The genetics of running-induced injuries involving tendon and bone – genetic study

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1 Introduction/Background

This is a genome-wide association study (GWAS) aimed at identifying genetic polymorphisms associated with the incidence of Achilles tendinopathy and bone stress injuries in the lower extremities. Due to the high popularity of running in Australia and relative high incidence of overuse injuries during running, recreational runners were selected as the study group. An online questionnaire approach was selected as a method of collecting epidemiological data on Australian runners and to provide a platform for the selection of the appropriate candidates to the GWAS study.

Research question:

1. What are the genetic contributors to the development of overuse injuries and what biological and physiological processes they influence?

Recreational runners were selected as a focus group for recruitment. A questionnaire was delivered using a commercial online survey software package (SurveyGizmo). The questionnaire was designed to collect epidemiological data of recreational runners and enable to select participants for the genetic part of the project. Test-retest reliability study showed that the survey is reliable and meets the recruitment needs for the project (1). The two main criteria for participation in the survey were that the participants were over the age of 18 and ran a distance of at least 15 km per week.

BC Platform software will be used for GWAS data storage and analysis and R software will also be used for data analysis.

2 Key papers/theoretical basis for the method


3 Ethical considerations

This protocol has been approved by the Bond University Human Research Ethics Committee (protocol number RO1688B).

The Participant Information Sheet is in Appendix A, and Ethics of Genetic Research Plan is in Appendix G.

Some of the ethical issues that need to be considered are:

1. **Data storage and coding.** Extensive care should be taken to maintain the participant confidentiality including the use of secure systems for the storage of genetic information, participant details and test results. Identifiers should be kept, however these details will be held separately to the genetic material, with participants given unique participant numbers when entered into the study. These mechanisms will keep the risk of a breach of confidentiality.

2. **Permission to participate in the genetics part of the study.** This should be included in the survey part of the study. Our survey includes the following:
   - I give permission, if I am eligible, to be contacted in the future for related research
     *This question is required.
   - I give permission, if I am eligible, to be contacted in the future to provide a saliva sample for genetic related analysis.
     *This question is required.

3. **Consent form (Appendix B).** An informed consent process in relation to disclosure of health and other information should be ensured, as well as information that the participant may be required to disclose to a third party. By ensuring that the participant is aware of these risks prior to consenting to participate, the risk that this information will impact on the participant is reduced.

4. **Withdrawal of consent form (Appendix C).** The Withdrawal of Consent form should include advice for participants that following withdrawal:
   - their data and if applicable their genetic material will be destroyed; and
   - they will be contacted with a confirmation once that's been done.

4 Facility and equipment

4.1 **Testing facility**

No laboratory required – method is for sample collection only.
4.2 Materials

4.2.1 Consumables

<table>
<thead>
<tr>
<th>Item</th>
<th>Photo/description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Biohazard bag</td>
<td></td>
</tr>
<tr>
<td>Large envelopes (for kits)</td>
<td>Padded bag, 215x280mm (prepaid postage)</td>
</tr>
<tr>
<td>Small envelopes (for samples)</td>
<td>Padded bag, 127x178mm (reply paid), “EXEMPT HUMAN SPECIMENS” written on front.</td>
</tr>
</tbody>
</table>

5 Training/qualifications/competencies

Please indicate (check the box) to indicate which of the following is required prior to undertaking this method:

Yes  No
☐ ☒ Health and Safety training
☐ ☒ Laboratory induction
☐ ☒ Current First Aid / CPR
☐ ☒ Immunisation
☐ ☒ Method-specific training
☐ ☒ Formal qualification required
☐ ☒ Other specific requirements
6 Restricted access

The use of this method does not require specific qualifications.

7 Health and safety / Risk Assessment

No risk assessments are applicable to this method. \textit{Error! Bookmark not defined. Error! Bookmark not defined. Error! Bookmark not defined.}
8 Workflow

8.1 Eligibility

This part of the study will not be promoted through media or other channels as the online survey acts as the recruitment tool for the participation in the genetic study.

1. Participants for genetic testing will be recruited directly from the survey participants (see protocol “CRN Workflow AIS epidemiology”).

2. Survey responses in SurveyGizmo are exported to Excel and sorted to establish inclusion and exclusion criteria. The workflow for selection of eligible participants is shown in Appendix D.

3. Participants that meet inclusion criteria are transferred to an Excel spreadsheet and assigned to these groups:
   - Uninjured for the past two years (no injury below the knee due to running that required them to discontinue running for a period of 2 weeks or more)
   - Diagnosed with a below knee bone stress injury caused by running, by a physician or physiotherapist and confirmed by diagnostic imaging, in the last 12 months
   - Diagnosed with Achilles tendinopathy or Achilles tendinitis caused by running, by a physician or physiotherapist in the last 12 months

8.2 Recruitment

1. Participants will receive an email inviting them to participate in the genetic study, with the Participant Information Sheet attached as a pdf document (Appendix A). The invitation requires that they email back their mailing address to enable postage of a saliva kit.

2. Participant replies to email (response recorded) in Excel work sheet.

3. The participant is allocated a barcode number which is recorded in the worksheet and scanned into BC System (details on Appendix E).

4. Participant details are uploaded into the BC System including survey response data, and copy of email return agreeing to participate.

A participation pack, including the saliva collection device, and the participant information sheets and consent forms, will then be mailed out to the participant.

5. Mail package is prepared to contain the following:
   - Oragene kit
   - Instruction sheet (Appendix F).
8.3 Sample collection

The test will be conducted by the participant in their own home. They will follow the kit instructions which are included in the envelope. Once they have collected the sample, they should put this in the provided prepaid envelope and send it back.

1. Sample is received in enclosed package and placed into secure study Pakman dropbox.
2. Samples are retrieved on a bi-weekly basis (Monday and Wednesday) for receipt processing.
3. Receipt processing occurs immediately after retrieval from dropbox; consent forms are checked for matching to sample ID, are signed, and scanned.
4. Samples are barcode scanned, logged and weighed, and recorded on sample receipt form.
5. After receipt processing, signed forms are stored in a locked filing cabinet drawer for the CRN within the restricted-access. Saliva samples are placed into racks/boxes securely into a locked cupboard within the same restricted-access laboratory for storage until batch extraction of DNA is scheduled (after accumulation of 300 saliva samples).

8.1 Cleaning and equipment maintenance

This test involves no cleaning or equipment maintenance.

8.2 Data handling and management

The data handling and managed is summarised in Appendix E.

8.3 Reporting of information

Participants will not have a written report of results.
# 9 Supplier and ordering information

<table>
<thead>
<tr>
<th>Item description</th>
<th>Product code</th>
<th>Supplier</th>
</tr>
</thead>
</table>
| Infinium CoreExome-24 BeadChip 2015. |              | Company name: Illumina  
| Oragene.DNA Barcoded Mailable Tube | OG-500.005   | Company name: DNA Genotek  
Name of contact: Natalie Rickers  
Email: natalie.rickers@dnagenotek.com  
| BC System                         |              | Company name: BC Platforms  
Web: [http://bcplatforms.com/](http://bcplatforms.com/) |
| SurveyGizmo                       |              | Company name: SurveyGizmo  
Name of contact: Seanalee Flaherty  
Email: seanalee.flaherty@surveygizmo.com  
Web: [www.surveygizmo.com](http://www.surveygizmo.com) |
10 Appendices

10.1 Appendix A: Participant information sheet

Participant Information Sheet

A study to investigate the relationship between genes, physical activity and health status.

Title The genetics of exercise-induced injuries involving tendon and bone

Protocol Number xxxx

Project Sponsor CRN for Advancing Exercise & Sports Science

Principal Investigator xxxx

Associate Investigator(s) xxxx

Location Australian Institute of Sport, Bond University & CRN partner organisations

Introduction

You are invited to take part in this research project because you completed the online survey for the study the genetics of exercise-induced injuries involving tendon and bone, agreeing to follow up genetic analysis and you are:

• A recreational runner,
• Aged 18-50 years of age,
• Run greater than 15km per week, and
• Fall into one of the following categories:
  – Uninjured* for the past two years (*no injury below the knee due to running that required you to discontinue running for a period of 2 weeks or more)
  – Diagnosed with a below knee bone stress injury caused by running, by a physician or physiotherapist and confirmed by diagnostic imaging, in the last 2 years
  – Diagnosed with Achilles tendinopathy or Achilles tendinitis caused by running, by a physician or physiotherapist in the last 2 years

The Participant Information and Consent Forms tell you about the research project. It explains the procedures involved. Knowing what is involved will help you decide if you want to take part in the research.

Please read this information carefully. Ask questions about anything that you don’t understand or want to know more about. Before deciding whether or not to take part, you might want to talk about it with a relative, friend or healthcare worker.

Participation in this research is voluntary. If you don’t wish to take part, you don’t have to.

You will receive the best possible care whether you take part or not. You’re not obliged to participate and if you do, you can withdraw at any time without penalty or prejudice and any samples that you have provided will be disposed of.

If you agree to participate in this study, we would like you to complete the enclosed paperwork and provide a saliva sample according to the enclosed instructions.
All aspects of the study including your personal details and results will be strictly confidential and only the principal researchers above will have access to this information.

If you decide you want to take part in the research project, you will be asked to sign the consent section. By signing it you are telling us that you:

- understand what you have read;
- consent to take part in the research project;
- consent to participate in the research processes that are described;
- consent to the use of your personal and health information as described;
- certify to the best of your knowledge and belief, you have no physical or mental illness or weakness that would increase the risk to of participating in this investigation;
- are participating in this project of your own free will and have not been coerced in any way to participate;

You will be given a copy of this Participant Information and Consent Form to keep.

**What is the purpose of this study?**

Identifying sequences or new mutations in known genes will help researchers to better understand the relationship between lifestyle, health status and genetic profile and we hope that this information will ultimately improve health and quality of life, prevent injury or disability, and prevent or treat chronic diseases. The genetic sequence information will provide new insights into the genetic factors associated with exercise-induced injuries in recreational and elite athletes.

This research is being conducted by a collaboration between xxx, xxx, xxx and xxx.

**What does participation in the research project involve?**

Participation is this portion of the project involves the provision of a saliva sample for genetic analysis. Enclosed are the consent forms to read and sign, a kit that will help you collect the specimen and return envelope to return your sample. This process should take approximately 10-15 minutes.

**What are the possible benefits to participating?**

We cannot guarantee that you will receive any benefits from this research, but your participation in the study may help doctors to better understand the relationship between lifestyle, health status and genetic profile with the hope that this will ultimately improve health and quality of life, prevent injury or disability, and prevent or treat chronic diseases.

**What will happen to my test sample and what are the possible risks to participating in the study?**

Please see the document The Ethics of Genetic Research.

**What if new information arises?**

During the research project, new information about the risks and benefits of the project may become known to the researchers. If this occurs, you will be told about this new information and the researcher will discuss whether this new information affects you.

**Can anyone participate in this study?**

As long as you meet the criteria specified earlier, and have participated in the online survey, you are eligible to take part.

**Do I have to take part in this research project?**
If you do not wish to take part then you don’t have to. If you decide to take part and later change your mind, you are free to withdraw at any stage. All information that you have provided can be destroyed at any time. You can withdraw your consent to participate in this research project by emailing the Principal Investigator at xxxx@xxxx.xxx

Before you make a decision to participate in any follow up studies, a member of the research team will contact you so that you can ask any questions you have about the project. You can ask for any information that you want.

Is this research project approved?

This project will be carried out according to the National Statement on Ethical Conduct in Research Involving Humans (2007) produced by the National Health and Medical Research Council of Australia. This statement has been developed to protect the interests of people who agree to participate in human research studies.

The ethical aspects of this research project have been approved by Bond University Human Research Ethics Committee; protocol number xxxx, contact xxxx, Research Ethics Manager on (0x) xxxx xxxx.

Will I get paid to participate in this study?

You will not be paid for participating in this study but all costs such as expenses involved with any investigation, posting of forms, questionnaires and samples will be covered

Who can I contact if I have any questions or problems in relation to this study?

If you wish to discuss further the experimental procedure or have any questions, please do not hesitate to contact Dr xxxx phone (0x) xxxx xxxx or email xxxx@xxxx.xxx

If you have any concerns with respect to the conduct of this study, you may contact the Secretary of the Bond University Human Research Ethics Committee Dr xxxx on (0x) xxxx xxxx or by email xxxx@xxxx.xxx
10.2 Appendix B: Consent form

The genetics of exercise-induced injuries involving tendon and bone information sheet

Genetic Sample and Data Consent

Principal Investigator: xxxx

Statement of Informed Consent for Genetic Sample and Data Collection

I have read, or had read to me in a language that I understand, this Participant Information Sheet and I understand the purposes, procedures and risks of this research project as described within it. I have had the opportunity to ask questions and I am satisfied with the answers I have received.

☐ I give permission for my anonymised sample and/or clinical information to be shared by the Investigators of this study with collaborating researchers who have ethically approved studies and are researching the relationship between genes, physical activity and health status.

☐ I give permission for my anonymised sample and/or clinical information to be part of the Biobank, to be shared with other researchers who have ethically approved studies and are researching the relationship between genes, physical activity and health status.

I understand that there is a very small chance that this study could identify a genetic defect that increases my risk for an unrelated condition.

☐ I would like to be informed if a risk factor for a treatable condition is identified

☐ I would NOT like to be informed if a risk factor for a treatable condition is identified

I understand that I will not receive genetic results for conditions where there is no known treatment at the time the result becomes available.

I understand that, due to the type of genetic test being performed, participation in this research project will not provide me with a clearance from any genetic or heritable conditions.

I freely agree to participate in this research project as described.

Name of Participant ___________________________ Date ___________ Signature of Participant ___________________________

Name of Researcher ___________________________ Date ___________ Signature of Researcher ___________________________
10.3 Appendix C: Withdrawal of Consent form

You can withdraw your consent to participate in this study at any time by returning this completed form by email to xxxx@xxxx.xxx or mail to:

xxxx

<table>
<thead>
<tr>
<th>Research Study Title</th>
<th>The genetics of exercise-induced injuries involving tendon and bone</th>
</tr>
</thead>
<tbody>
<tr>
<td>BUHREC Approval Number</td>
<td>xxxx</td>
</tr>
</tbody>
</table>

I hereby wish to **WITHDRAW** my consent to participate in the research proposal described above and understand that such withdrawal **WILL NOT** jeopardise any treatment or my relationship with the Australian Institute of Sport, Bond University & CRN partner organisations.

I understand that all of my data and genetic material (if applicable) will be destroyed and that I will be contacted on completion of this process.

___________________________________________________________
Research Participant Name *(Print)*

___________________________________________________________
Research Participant Signature  Date

Research Participant contact details (email, phone or mailing address acceptable)
10.4 Appendix D: Workflow for selection of eligible participants

- The Order of Selection Criteria for Runners:
  1. I give permission, if I am eligible, to be contacted in the future for related research: Please enter your personal details here (include Yes)
  2. I give permission, if I am eligible, to be contacted in the future to provide a saliva sample for genetic related analysis: Please enter your personal details here (include Yes)
  3. What year were you born?: Please enter your personal details here (include between 18 and 50)
  4. Maternal grandmother: What is the ethnic background of your biological grandparents? Maternal grandfather: What is the ethnic background of your biological grandparents?
     Paternal grandmother: What is the ethnic background of your biological grandparents?
     Paternal grandfather: What is the ethnic background of your biological grandparents? (include only “Caucasian Europeans”)
  5. Km: On average, how many km per week would you run? (exclude <15)
  6. In the last 2 years have you had any injuries of the lower limbs, which have forced you to discontinue running for a period of 2 weeks or more? (separate Controls and Cases)

- Uninjured controls:
  1. Do you currently smoke? (Exclude smokers)
  2. If No, at what age did you quit? (Exclude those, who gave up smoking less than 5 years ago)
  3. Rheumatoid arthritis: Have you ever been diagnosed with any of the following conditions/disorders?
     Osteoarthritis: Have you ever been diagnosed with any of the following conditions/disorders?
     Osteoporosis: Have you ever been diagnosed with any of the following conditions/disorders?
     Cystic fibrosis: Have you ever been diagnosed with any of the following conditions/disorders? (if “Yes”, exclude)
  4. Undergone chemotherapy?: To your knowledge, have you ever (If “Yes”, exclude)

- Cases:
  1. You indicated that you have been diagnosed with a lower limb injury within the past 2 years. How did this injury occur? (include ONLY “while running”)
  2. Doctor?: Was this injury diagnosed by a professional:
     Physical therapist?: Was this injury diagnosed by a professional:
     Was this injury diagnosed by imaging (x-ray/ultrasound/bone scan/CT scan/MRI)? (at least one “Yes”)
  3. Apply 1 and 2 for all injuries, which are described by a runner.
  4. Search for ‘Shin Splint’ in Other injuries - What type of injury was it? And include, applying all other criteria.
  5. Do you currently smoke? (Exclude smokers)
  6. If No, at what age did you quit? (Exclude those, who gave up smoking less than 5 years ago)
7. Rheumatoid arthritis: Have you ever been diagnosed with any of the following conditions/disorders?
   Osteoarthritis: Have you ever been diagnosed with any of the following conditions/disorders?
   Osteoporosis: Have you ever been diagnosed with any of the following conditions/disorders?
   Cystic fibrosis: Have you ever been diagnosed with any of the following conditions/disorders?
   (if “Yes”, exclude)

8. Undergone chemotherapy?: To your knowledge, have you ever (If Yes, exclude).

9. Have you ever had a fracture of any bone?
   Where was the fracture?: If yes:
   When did it occur?: If yes:
   If a low limb fracture happened during past two years – Ask additional information by email.

10. If someone had a steroid injection – ask if it was before the injury.
10.5 Appendix E: Workflow to send out kits

- **Package preparation**
  1. Open BC, select folder AIS INJURY STUDY/ Recreational Runners/REC restricted and click on the dataset **Participants to send kits**.
  2. Click on **Count subset rows**, you can see how many kits you have to send and compare the number to the number of the emails in the folder Confirmed address.
  3. Click on **export** on the left in the Datasets options, then click **Next** (don’t change anything here)
  4. In the multiple choice question encoding choose **Export as text**
  5. Select variables – click Select and click on FIRSTNAME, LASTNAME, STREETNAME, SUBURBNAME STATEPOSTCODE, then click OK and **Export data**
  6. In the Tools options on the left click on **result archive** and click on the latest job and then on the green arrow on the right. You are saving a text document with the list of runners to send out kits
  7. Open the file and copy all rows with the participant information
  8. Open folder ‘My Labels’ and then folder ‘AIS labels’ and open text file ‘INJURY STUDY RESPONSE id LABELS’ and paste the data, then save the file.
  9. In the same folder open file ‘INJURY STUDY RESPONSE id LABELS.lbl’ and click on Print, then Select records and tick each box of the response ID you wish to print. Click OK and then Print. (make sure that the printer is on and connected to the computer)
  10. Open folder ‘Labels form for envelopes’, open ‘AIS envelope labelling spreadsheet’ and paste copied data on the first page ‘Exported data’. Yellow cells will adjust data in the form for printing the labels (everything will be in capitals). Then copy data from yellow cells and paste as Values on the second page ‘LINKED SHEET’. Save and close the file.
  11. Open file ‘AIS envelope labelling template_wider sides’, click Ctrl+A and then F9 – the form should be updated with the names and addresses from the ‘LINKED SHEET’. Now you can print the labels on the stickers, check that in the printer properties you selected Bypass tray as a paper tray and put the stickers there.
  12. In the same folder you have a file ‘Labels for small envelopes’ which you can use to print labels for small envelopes.
  13. To pack the kits you’ll need printed consent forms with sticker ‘sign here’ and the 6 page, small envelopes, biohazard bags and stamped on both sides big envelopes (stickers and stamps are at my desk under the second screen)

- **Registration of the kits**
  14. Open BC, select folder AIS INJURY STUDY/ Recreational Runners/REC restricted and click on the dataset **Injury Study – RECREATIONAL – Restricted database**
  15. Under Data input on the left click on the add entry and scan with the scanner (it should be connected to the laptop) first printed label of the response IDs (or type in), click OK
  16. Check the name and the address, stick the label on the consent form on the same page where the ‘sign here’ sticker is, take the kit you are going to allocate to the response and attach barcode stickers from the backside of the kit to the consent form on 6 and 9 pages.
  17. Select Yes/No in the question ‘Did participant request their genetic data?’
18. Select ‘Yes’ in the question ‘Has the kit been sent out?’, then select the date you are sending out the kit and scan the barcode from the tube (no need to open the box).

19. Look up in the Excel spreadsheet of the selected participants in what group they should be (Uninjured, BSI, AT etc) and choose the group in the line ‘What phenotypic group are they?’ And also select Yes/No in the following 4 lines.

20. Now save the changes and continue with the next response ID.

21. Put the kit, consent form, small envelope and biohazard bag in the big envelope and it’s ready to be sent.
10.6 Appendix F: Instruction Sheet

Thank you for agreeing to participate in *The genetics of exercise induced injuries involving tendon and bone.*

Please set aside 5 minutes to collect the sample and fill out the paperwork.

Please follow the steps below, **skipping a step could mean that your sample is unable to be included in the research:**

1. You must **wait 30 minutes after eating, drinking or smoking** before providing a saliva sample.
2. Read the Participant Information Sheet and the document entitled *The Ethics of Genetic Research - ethically defensible plan.* Contact the researchers using the email/phone number provided if you have any questions.
3. Once the information is read and understood, sign the consent form (the form marked COPY is for you to keep for your own reference).
4. Open the Oragene – DNA kit and carefully read the instructions provided on the next page, follow the five steps carefully. Please ensure that the sample fills the tube to the line without any bubbles
5. Place the **collection tube** (but not the funnel) **into the biohazard bag**.
6. Place the following in the reply paid envelope:
   a. **Tube containing saliva sample in the biohazard bag**
   b. **The signed consent form (single page only)**

then seal and place the envelope in the post.

Please do not hesitate to contact the AIS if you have any questions or issues.

Kind regards,

xxxx
0x xxxx xxxx or 04xxxxxxx
injurystudy@ausport.gov.au
10.7 Appendix G: The Ethics of Genetic Research

The Ethics and Risks Associated with Genetic Research

A study to investigate the relationship between genes, physical activity and health status.

What is genetic research?

Genes are made of DNA – the chemical structure carrying your genetic information that determines many human characteristics such as the colour of your eyes. Researchers study how genes are expressed in order to understand why some people differ in their response to exercise and to develop, improved and individualised approaches to exercise prescription. This will potentially provide new ways in which to reduce disease within the wider community.

What will happen to my test sample?

Saliva samples will be used immediately or stored for future studies. Saliva samples will be used to extract your DNA. DNA sequence and other information obtained in this research project will be stored securely, with each participant sample allocated a specific code number. All data processing will occur by use of this number and not the participant’s name. However all samples are potentially re-identifiable and must remain so to satisfy the aims of the research and also to identify participants if required for the return of genetic information.

The samples will be tested using genome-wide association studies. These studies are increasingly being used to identify biological pathways and networks underlying complex diseases. This type of genetic test is very unlikely to identify mutations associated with heritable diseases as it will not provide information about the whole genome, only portions of the genome which are unlikely to contain genes relating to disease.

Researchers from the Australian Institute of Sport (Canberra), University of Queensland (Brisbane), the University of Sydney and Bond University (Gold Coast, Queensland) are collaborating to study your DNA samples for research only. We may also share some information about your clinical information in our research group but all identifiers will be removed. All medical information is stored in password protected databases.

We will also be establishing a biobank – this is a way of collecting and storing anonymous biological material and information for further research. If you consent, your DNA will be stored and then can be used by other researchers for future projects.

What are the possible risks of participating in the study?

All medical information is stored in password protected databases. It is possible, though highly unlikely, that someone could get access to this database without permission. All samples and participant data will be stored securely in locked facilities.

This type of genetic test is very unlikely to identify mutations associated with heritable diseases. However, as part of this study there is a small chance that we might coincidentally find a defect in a gene which, in the duration of the project, may be identified as being associated with an increased risk for a genetic condition. If the condition is treatable or preventable, you can specify on the consent form if you want to be informed about such a finding. Learning this information may be upsetting. It could also affect your ability to get life insurance. The information could be used against you in the work setting.

You may be asked to give us health information about your relatives. Any information you give us will be kept confidential. We will not contact your relatives without your permission. We may discuss with you the possibility of including your relatives in the research project in the future. You may learn information from your test result about inherited diseases or disorders that may affect others, such as your brothers or sisters. This could interfere with family relationships. You may be faced
with the decision to make the family aware of the existence of genetic information. Family members may or may not wish to know this information. Therefore, although we think that there are benefits in having that information, individuals have to weigh up the risks and decide whether or not to receive that information. In the unlikely event that a risk for an unrelated genetic condition is identified we will help to put you in contact with a local genetics service.

If we identify that you could be at risk for a disorder, which is currently untreatable and unpreventable, we will not disclose this information.

If you become upset or distressed as a result of your participation in the research, the researcher is able to arrange for counselling or other appropriate support. Any counselling or support will be provided by staff, who are not members of the research team. In addition, you may prefer to suspend or end your participation in the research if distress occurs.

What if new information arises?
During the research project, new information about the risks and benefits of the project may become known to the researchers. If this occurs, you will be told about this new information and the researcher will discuss whether this new information affects you.

How will I be informed of results from of this research project?
In relation to any genetic information generated from use of your DNA, you may elect to:

1. Have no information returned to you.
2. If a risk factor for a treatable condition is identified, have information returned to you to provide to your medical practitioner for consideration, verification testing and possible clinical or other action.

If your preference is not to receive genetic information, a researcher will contact you following the completion of genetic testing to ensure that this preference still remains.

What will happen to information about me?
In accordance with relevant Australian privacy and other relevant laws, you have the right to access the information collected and stored by the researchers about you. You also have the right to request that any information, with which you disagree, be corrected. Please contact one of the researchers named at the end of this document if you would like to access your information.

If you wish to discuss further the experimental procedure or have any questions, please do not hesitate to contact Dr xxx phone (0x) xxxx xxxx or email xxxx@xxxx.xxx

If you have any concerns with respect to the conduct of this study, you may contact the Secretary of the Bond University Human Research Ethics Committee Dr xxxx on (0x) xxxx xxxx or by email xxxx@xxxx.xxx