PARTICIPANT INFORMATION AND INFORMED CONSENT FORM FOR RESEARCH INVOLVING GENETIC STUDIES

TITLE OF RESEARCH PROJECT: Application of personalised medicine using an integrated service and research approach.

REFERENCE NUMBER: N09/08/224
PRINCIPAL INVESTIGATOR: Professor MJ Kotze
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We would like to invite you and where relevant also other members of your family to participate in a **combined service and research programme** that involves genetic analysis and possible long-term storage of blood or tissue specimens. Please take some time to read the information presented here which will explain the details of this project. Please ask the study staff or doctor any questions about any part of this project that you do not fully understand. It is very important that you are fully satisfied that you clearly understand what this research entails and how you could be involved. Also, your participation is **entirely voluntary** and you are free to decline to participate. If you say no, this will not affect you negatively in any way whatsoever. You are also free to withdraw from the study at any point, even if you do agree to take part initially. This research study has been approved by the ethics Committee for Human Research at Stellenbosch University and it will be conducted according to international and locally accepted ethical guidelines for research, namely the Declaration of Helsinki, and the SA Department of Health's 2004 Guidelines: Ethics in Health Research: Principles, Structures and Processes.

What is Genetic research?
Genetic material, also called DNA or RNA, is usually obtained from a small blood sample, saliva or a cheek swab to study inheritance in families. DNA or RNA can also be extracted from other sources such as liver or tumour biopsies to determine the best treatment strategy for a patient. Genes are found in every cell in the human body. Our genes determine what we look like and sometimes what kind of diseases we may be susceptible to or how we will respond to medication and other environmental exposures. Worldwide, researchers in the field of genetics are continuously discovering new information that may be of great benefit to future generations and also that may benefit people today, who suffer from particular diseases or conditions.

What does this particular research study involve?
If a blood or saliva/cheek swab sample is provided hereditary factors may be tested that determine treatment response and/or **increase the risk** of rare diseases or relatively common medical conditions **in the presence of certain environmental factors**. Based on gene-environment "mismatches" that may be identified and your personal medical and family history, a personalised lifestyle modification plan may be provided after approval by a clinician. This process including genetic testing and generation of a report may take several weeks depending on the initial results that may identify the need for more comprehensive analysis or no further action required. Although study participants may request the lifestyle and diet recommendations for immediate implementation, details on any specific genetic alteration identified will not be provided without genetic counselling or further explanation by a clinician, registered medical scientist or genetic counsellor. The cost for the genetic consultation and reporting including the detailed genetic test report may not be covered by the research and will be discussed with you at entry into this project.

Why have you been invited to participate?

As you are either overweight or a sufferer from a chronic disease or a healthy person who may have a personal or family history of a disease with a genetic component and expressed an interest in risk factor screening at a Wellness Day, during a questionnaire-based survey, or during consultation by a genetic counsellor or clinician, we would like to use the information obtained from you/your family in the research. We are interested to find out how a person's genetic profile may influence the response to the intervention program to be recommended.

What procedures will be involved in this research?
You will be asked to fill in forms with questions pertaining to current health status, family history and use of (chronic) medication, as well as a number of lifestyle factors such as alcohol intake, smoking sport/exercise program, etc. where appropriate. Your length and weight may be measured and up to three small blood bottles of 5 ml each (about three tablespoons), saliva, a blood spot or finger prick, and/or a small amount of tissue or cells (e.g. cheek swab using a sterile cotton bud rubbed a few times in the inside of your cheek) may be taken for laboratory analysis. In addition to genetic studies, biochemical tests (e.g. cholesterol, iron levels) and liver enzyme function may be determined. These pathology tests may indicate altered gene expression that may lead to disease development if left untreated. Collection of blood or other samples may be performed before your genetic test results are provided and possibly again in future to determine your response to the recommended intervention plan. If your specimen has already been used for genetic testing at another laboratory, or is collected as part of a routine service or Wellness Program offered by a Medical Scheme or clinician, you may be approached to

participate in the research in which case the **information obtained as part of your routine clinical work-up will be applied** in the research project. Your clinical care will not be changed due to the inclusion of your information for research purposes.

Genetic material of a study participant may be selected for whole genome sequencing, in which case additional information will be provided to better explain the potential consequences of the results. However, due to the high cost of this procedure, only a few samples will be tested in this way in a family context. It remains the prerogative of the researchers to select the samples most appropriate for the study.

Are there any risks involved in genetic research?
Minor pain or bruising may be experienced at the site where the specimen is taken for laboratory testing. In the event that genetic testing is performed in families, non-paternity may be revealed and it is therefore important that adoption be reported before genetic testing is performed. Based on the family history reported and personal health profile, genetic counselling/consultation may be advisable (the cost for this service is not covered as part of the research). Some insurance companies may mistakenly assume that taking part in genetic research indicates a higher risk for disease. Thus, no information about you or your family will be shared with such companies unless you provided permission. Also, since some of the techniques that will be used may be experimental and thus possibly unreliable, a quality control system will be implemented as part of this project whereby some of the specimens used for the study may be analysed by different laboratories, possibly using different mutation detection methods.

Are there any benefits to your taking part in this study and will you get told your results?

The specimen(s) provided may be stored and tested at a later time when batches of samples are available; this may be necessary to limit testing time and costs involved. This research will benefit other people in the future as they might be in a position to get treatment earlier to prevent or specifically target the cause of many chronic diseases. Health guidelines for lifestyle modification where appropriate will be provided after approval by a clinician but the genetic results obtained (e.g. if additional whole genome sequencing was performed) will be made known to you only if they indicate that you:

- Have a particular disorder or family history relevant to the test results
- Have a condition or predisposition to developing a condition that is treatable or avoidable e.g. by a lifestyle modification
- May need genetic counselling

How long will your blood/specimen be stored and where will it be stored?
Your specimen will be stored at the University of Stellenbosch Medical School in a dedicated fridge or freezer for at least 5 years or at the laboratory that performed the same test(s) as a routine service. A material transfer agreement will be signed by a representative of the university if/when the specimen is shipped from the University of Stellenbosch to another laboratory or country for quality assurance or collaborative research purposes.

If the blood or specimen provided is stored is there a chance that it will be used for other research?

Your specimen will only be used for the genetic research as indicated above and any further testing will only be done after obtaining full written consent. Also, if the researchers wish to use your stored specimen for **additional research in this field** they will be required to apply for permission to do so from the Human Research Ethics Committee at Stellenbosch University that can be contacted at telephone number 021 938 9657. If you do not wish your specimen to be stored after this research study is completed you will have an opportunity to request that it be discarded when you sign the consent form.

How will your confidentiality be protected?
The specimens will be given an ID number and only the researchers, clinicians and laboratory personnel involved in the study will have access to the original questionnaires and assessment forms with identifying information. Specimens sent to other laboratories local or abroad will be shipped only with the ID number attached to them and your name will not be disclosed to the research collaborator. If ever information comes to light that could be important for the individual or their descendants, all possible attempts will be made to contact these participants and counsel them. The results of the study will be presented at congresses/workshops/forums and included in scientific articles and student theses, without revealing the identity of the study participants.

Will you or the researchers benefit financially from this research?
You will not be paid to take part in this study.

Important information: In the event that this research leads to the development of a new commercial application or patent, you or your family will not receive any profits or royalties although the researchers may benefit in this respect

Project N09/08/224 – This section not required in the case of online consent

By signing below, I ……..agree to take part in a research study entitled: **Application of personalised medicine using an integrated service and research approach.**

I declare that:

- I have read the full participant information leaflet or had explained to me this information that is written in a language with which I am fluent and comfortable.
- I understand that I can ask questions any time and all my current questions have been adequately answered.
- I understand that taking part in the research component of this genetic service is voluntary and I have not been pressurised to take part.
- I understand that withdrawing from research participation at any time will not affect my current medical treatment.
- I may be contacted by any of the clinicians and researchers involved in this project in the event that additional information is required

Tick the option you choose:

☐ **STORE SAMPLE:** I agree that my blood or tissue sample can be stored **indefinitely** but I can choose to request at any time that my stored sample be destroyed. My sample will be identified with a special study code that will remain linked to my name and contact details. I have the right to receive confirmation that my request has been carried out. My sample may be shipped to another laboratory in SA or abroad to be used in other research projects in this or a related field without revealing my identity.

OR

☐ **DECODIFY SAMPLE:** I agree that my blood or tissue sample can be stored **indefinitely** after the project is completed but that it is anonymised with all possible links to my identity removed, and that the researchers may then use it for additional research in this or a related field. My sample may be shipped to another laboratory in SA or abroad to be used in other research projects in this or a related field.

OR

☐ **DESTROY SAMPLE:** Please destroy my blood or tissue sample as soon as the current research project has been completed.

Signed at (*place*) ..... on (*date*) .....

..... **.....**

Signature of participant Date of birth

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Declaration by investigator: I…… declare that:

- I explained/provided the information in this document to the study participant
- I encouraged him/her to ask questions and took adequate time to answer them.
- I am satisfied that he/she adequately understands all aspects of the research as described above.

..... Signed at……..……on (date)……..……

Signature of investigator / clinician/ research assistant **(not required in the case of online consent)**

If an interpreter is used then the interpreter must sign the declaration below.

Declaration by Interpreter: I …… declare that:

- I assisted the investigator to explain the information in this document to the participant using the language medium of Afrikaans/Xhosa.
- We encouraged him/her to ask questions and took adequate time to answer them.
- I conveyed a factually correct version of what was related to me.
- I am satisfied that the participant fully understands the content of this informed consent document and has had all his/her question satisfactorily answered.

Signed at (*place*) .....…..……... on (*date*) ……..…….. .

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Signature of interpreter