

Information Sheet

Collection of Biological Samples (Blood and Body Fluids)

for DwarnaBio and StudjuDWARNA

at the Centre for Molecular Medicine and Biobanking

University of Malta

Invitation

We would like to invite you to participate in StudjuDWARNA, a study aimed at conducting a comprehensive evaluation of the genomes of Malta. Further information on StudjuDWARNA is available below.

We would also like to invite you to bank your blood/body fluid sample at DwarnaBio, a biobank at the Centre for Molecular Medicine and Biobanking (CMMB), to help us build a repository of human biological samples, which will be used for medical research.

You should only participate if you want to; choosing not to take part will not disadvantage you in any way. To help you decide whether you want to participate, or not, we are providing you with information on the research that could be carried out on the sample/s you provide, what your participation will involve, and how it may affect you. Please take time to read the following information carefully and discuss it with others if you wish. If there is anything that is not clear, or if you would like to receive more information, contact the Biobank Manager on the e-mail or telephone number provided at the end of this document.

What is StudjuDWARNA?

StudjuDWARNA aims to conduct a comprehensive evaluation of the genomes of Malta. This is the first study to draw on a nationally representative cohort of biobanked samples and data, obtained from DwarnaBio, the Maltese national biobank led by the University of Malta. StudjuDWARNA will perform genomic sequencing and analysis of a reference Maltese collection from the general public. It will aim to explore the association between genomic architecture and both rare and common diseases, and eventually to relate genomic findings to clinical outcomes in longitudinal studies. StudjuDWARNA will also pave the way for the construction of a reference Maltese genome. This will be an incredibly powerful tool for future health research with the potential of developing more personalised healthcare and prevention strategies for the Maltese population.

What is a biobank?

A biobank is a research infrastructure, which stores human biological samples, such as blood, DNA, cells, body fluids and human tissues, together with associated personal and medical information, with the main aim of acting as a resource centre for human biological research.

Biobanks collect body fluids and tissues from patients, to store in a Clinical Bank, and from the general population to store in a Population Bank.

Access to samples for research is open to researchers worldwide. Where personal data will be transferred outside the European Economic Area (EEA), this transfer will be carried out in accordance with Chapter V of **Regulation (EU) 2016/679**, the **General Data Protection Regulation (GDPR)** and provided that the biobank is satisfied that ethical approval has been formally obtained from a Research Ethics Committee. Researchers are required to sign a Material Transfer Agreement and a Data Transfer Agreement with the biobank prior to transfer of samples and associated data.

Biobanks are particularly useful for research on rare diseases, where they are the only source providing sufficient samples for research. According to the European Organisation for Rare Disorders (EURORDIS), a rare disease in Europe is classified as any disease that affects fewer than 1 in 2000 Europeans (less than 200 Maltese).

What is DwarnaBio?

DwarnaBio is the national biobank and is based at the Centre for Molecular Medicine and Biobanking (CMMB) at the University of Malta. Governance is provided by the Statute of the CMMB, which states that the Rector of the University, or his delegate, chairs the Board, members of which are appointed by the Council of the University for a duration of one (1) year, renewable for up to four (4) years. Operational aspects of the Biobank are based on a Management Committee, which includes a Quality Management Team and experts in ethical, legal and societal implications (ELSI).

The Population Bank holds samples from adult persons representative of the general population while the Clinical Bank holds samples provided by patients with various diseases, such as blood disorders, diabetes, renal disorders, neurological disorders and various cancers including breast and colon cancer, a number of which are rare diseases.

The Biobank is a founding member of the Biobanking and Biomolecular resources Research Infrastructure-European Research Infrastructure Consortium (BBMRI-ERIC) of the European Union.

Aims of DwarnaBio

DwarnaBio aims to provide biological samples and associated data for biomedical research in Malta and through the online sample catalogue, for research institutions overseas.

Scientific advances on the understanding of the human genome are leading to substantial changes on how to approach the treatment of many diseases and are opening up new perspectives in the field of scientific research centred on the patient. Research can vary from establishing the genome of a specific population, to the study of the genetic basis of specific diseases in affected families, to the discovery of pharmaceuticals and diagnostic biomarker tools, all aiming to improve healthcare.

Current laboratory techniques (and those to be developed in the future), which study genetic pathways, might help us understand better the pathophysiology of disease and consequently lead to improved medical treatment and ultimately to personalised medicine. Having a collection of samples with accompanying data already stored in a biobank will facilitate research, especially for rare diseases.

A Population Bank with a large database will make it possible to carry out long-term (longitudinal) studies, primarily to establish the epidemiology (distribution, patterns, risk factors) of health and disease. Studies focus on the ways that the social and physical environments might interrelate with genetic factors in the onset and development of particular diseases. The ultimate aim is to understand the genetic variation in normal individuals, so that disease prevention measures can be developed and administered to maintain a healthy population.

Why was I invited to participate?

You have been contacted to participate in:

a. the Population Bank

because you are currently a healthy person and have shown an interest in participating in health research, by taking the initiative to contact us through the Dwarna portal/NGO/Patient Advocacy Group/health promotion event or any other way. We thank you for your interest.

b. the Clinical Bank

because you have already provided blood and/or a tissue sample for diagnostic purposes or for management of one of the following diseases: haemoglobin disorders; genetic diseases (e.g., disorders of coagulation, cystic fibrosis, gangliosidosis, haemochromatosis, phenylketonuria, Y chromosome deletion); or cancer (e.g., breast cancer, thyroid cancer, gastric cancer, colon cancer). The Biobank already has collections of samples from patients with these conditions and is keen to increase the database.

Your participation is being actively sought to donate blood and/or saliva and/or urine, from which we will extract DNA. You will also be required to provide your demographic data (age, gender, ethnicity, level of education, occupation) and your medical data.

Demographic data is useful to study patterns of health/disease over time in the population. These studies will help us analyse ways that your socio-economic status impacts on long term health.

All samples will be stored and preserved in appropriate conditions in the Biobank until they need to be used for medical research in Malta. Research must be approved by the Centre Research Ethics Committee (CREC) at the University of Malta, or the Health Ethics Committee (HEC) at the Ministry for Health or another recognised Research Ethics Committee. Samples and/or data requested by internationally recognised research institutions will only be transferred overseas if research ethics approval of the proposed research project is provided by the applicant research institution, to the satisfaction of the Biobank governance.

All researchers, from Malta and from overseas, enter into a legal agreement with the Biobank to ensure that your personal data is fully protected in line with national and European standards and legislation.

What will happen to me if I decide to participate?

If you agree to participate in this initiative, you will be invited to attend at a Genetics Clinic, either at Mater Dei Hospital, or at the Centre of Molecular Medicine and Biobanking, where you will be first asked to sign the attached Consent Form.

Then a sample will be taken in one/or more of these three (3) ways:

- 1) a saliva sample will be taken by swabbing the inside of the cheeks with a buccal swab; and/or
- 2) a sample of blood (20ml) will be taken from your arm (venepuncture) by a qualified and experienced nurse or a phlebotomist; and/or
- 3) a urine sample provided by the research participant at the clinic.

The saliva and/or blood sample will be processed at the Biobank and both the remaining samples and/or the extracted DNA samples will be stored in the quality approved DwarnaBio facilities.

During the same visit, you will also be asked to provide demographic data, which includes your personal details (age, gender, ethnicity) and some information about your socio-economic status (level of education, occupation). You will be asked to provide medical information by answering some health-related questions in the form of a questionnaire. The demographic data and the medical data is called **sample associated data**.

We are also requesting your permission to authorise DwarnaBio to request access to your health records from the Director of the Directorate for Health Information and Research at the Ministry for Health and from the Data Controller at Mater Dei Hospital. Your consent will allow DwarnaBio to obtain your medical information from your hospital admissions, diagnostic tests, discharge letters and from data in disease registries.

You will also be asked to provide personal **contact data** (name, Identity Card or Passport number, address, e-mail and mobile number) for the **sole** purpose of contacting you in the future. We will contact you only if, on the Consent Form, you opt to be re-contacted by the Biobank to be provided with:

- a) important information before deciding to consent to the future use of your samples and associated data in a research project; and/or
- b) specific results and information which may be important for your health and/or your medical management. In this case, you will be referred to a medical doctor for further evaluation.

Your **contact data** will be **stored separately from your coded demographic and medical data**. It will be retained by the Biobank and **will not be shared** with researchers or any other third parties, unless this is legally required or if otherwise ordered by the Maltese Courts of Justice.

It is in your interest to keep the Biobank informed of a change in your contact details.

We are therefore requesting your signed explicit consent to donate your samples, and provide associated data, which will in future be used in research, only according to the specific options you choose on the Consent Form. You will be able to change these consent conditions at any time by contacting the Biobank. Once you provide us with your consent, you will be considered as a research participant.

What will happen to my sample?

Processing and storage

Samples will be transferred to DwarnaBio for processing to extract DNA prior to storage at low temperatures at secure facilities at the Biobank, in keeping with accredited laboratory guidelines to ensure quality assurance and quality control, crucial for obtaining optimum samples suitable for use in research studies.

The sample and the sample associated data (demographic and medical data) will be coded, as explained in the next section. The processing and storage of data is managed through secure computerised systems at the University of Malta.

Your participation in StudjuDWARNA will be for ten (10) years.

Your sample and/or extracted DNA, and the associated data, will be retained for ten (10) years, according to common research practices within the European Union. You will then be re-contacted to renew your consent to storage. However, you have the right to withdraw your consent at any time and if that happens, the sample/s and associated data, including your personal contact data that you provided, will be destroyed.

The Population Bank sample collection is under the responsibility of a designated curator, the Biobank Manager. The Clinical Bank sample collection is under the joint responsibility of the Biobank Manager and a designated curator, a member of the team who organises the collection.

Pseudonymisation

The sample Collection will be an “Identifiable Collection” as the biological samples will only be partially anonymised (pseudonymised) by coding, that is, they are assigned a random number as a label. The samples with their associated data, including the medical information archived and referenced in this way, can only be linked to the personal contact data through this code, securely held by the Controller.

DwarnaBio has decided not to have complete anonymisation because interpretation of research results depends on the associated medical data. Also, it might be in your interest to be re-contacted with specific results and information, which may be important for your health and/or your medical management. However, this will only occur if you consent to be re-contacted by the Biobank.

Return of results from StudjuDWARNA

If in the course of genetic analysis of your samples, there is identification of clinically significant findings that are directly relevant to your health and/or your medical management, you will be re-contacted to be provided with results and/or information, but **only** if on the Consent Form, (point 7), you choose to be re-contacted by DwarnaBio. In this case, you will be referred to a medical doctor for further evaluation.

Future use of samples and associated data

It is important to emphasise that when samples and their data are used in research, approved by the Centre Research Ethics Committee (CREC) at the University of Malta, or the Health Ethics Committee (HEC) at the Ministry for Health, or any other recognised Research Ethics Committee, only the pseudonymised samples and associated coded data will be made available to the main researcher, in line with the approved protocol and the consent of the participant.

Your samples and associated data will **never** be used for research on human embryos or for human stem cell research.

As explained above, your biological samples are collected, and stored, and used in future research, only with your consent. Coded samples may be used in various research projects, following ethical approval of the new research project by the appropriate Research Ethics Committee.

Each future research project will require approval from a Research Ethics Committee, as appropriate, before the researcher applies for use of samples and associated data from DwarnaBio. If you consent to be re-contacted via our *portal* www.dwarna.mt, we intend to keep you, as the research participant, aware of studies in which the Biobank is involved, including an explanation of any new technology which may be developed for research. The Information Technology (IT) system for this portal (dwarna.mt) is completely separate from the Biobank's IT system.

Dwarna portal will access your contact data only for the purpose of facilitating dynamic consent, allowing you to modify your consent options. Dwarna portal will not have access to your demographic and medical data.

Your consent will also authorise the Biobank to share data in an aggregate form through the online biobank sample catalogue.

Data obtained from research studies will only be published in an anonymous and aggregated way, such as percentages or numerical data, without identification of the research participant. Under no circumstances will data be provided in an individualised way. In this way, your data protection rights will be ensured and respected.

Coded (pseudonymised) samples and associated data may only be shared with researchers from countries outside Malta when there is approval by DwarnaBio, which is legally bound to ensure that necessary data protection safeguards are in place prior to effecting such transfer, in line with local legislation, including adherence with the **GDPR**.

What are the benefits and risks if I agree to take part?

Benefits

You may not be able to benefit personally from the research process. However, the research results obtained in future studies will enable us to learn more about common and rare diseases, which may help future patients, including possibly yourself and/or your family. You may also benefit from any disease prevention measures that are introduced through new knowledge arising from future research.

Your participation is an altruistic donation. You will not gain financially by participating, even if research leads to commercial development of a new treatment or a diagnostic tool or a test/s that may someday be used for genetic diagnosis. If such a test/tool/treatment is developed, you have no right for any payment, even if a third party, including a commercial entity, might benefit from such development.

Risks

There is no significant physical risk to you if you participate. Although the drawing of blood causes no serious problems for most people, it can cause some bleeding, bruising, and/or discomfort at the injection site.

Taking a buccal swab requires a gentle scraping of the lining of the mouth, which may rarely result in bleeding. You will have to abstain from eating and drinking for at least one hour prior to the test.

The storage of samples and data involves minimal risk because the Biobank has taken necessary and appropriate security measures, including but not limited to, secure storage facilities for samples, the implementation of a strict coding technique for pseudonymisation, and the use of a robust IT environment for the purpose of ensuring the security of data processing.

Data Protection

Processing and storage of data

The Controller responsible for the processing of your personal data is DwarnaBio, established within the Centre of Molecular Medicine and Biobanking at the University of Malta.

The purpose of data collection is to build a repository of human biological samples with associated data, to be used for medical research, only according to the specific options you choose on the Consent Form.

Your data will be treated in accordance with national law on data protection, namely the **Data Protection Act**, Chapter 586 of the Laws of Malta, which implements **Regulation (EU) 2016/679**, the **General Data Protection Regulation (GDPR)**.

Your signed explicit consent will serve as the legal basis for processing your data, in line with Article 6(1)(a) of **Regulation (EU) 2016/679**, the **GDPR**.

The signed Consent Forms (paper documents) will be stored at DwarnaBio in a fireproof safe in a secure office accessible only to the Biobank Manager and authorised delegates.

Biological samples will be pseudonymised by coding. The demographic data (age, gender, ethnicity, level of education, occupation) and medical data associated with the sample will be coded with the sample number and stored at DwarnaBio in secure computers, maintained by IT experts. The key for the code will be stored separately at DwarnaBio in a separate secure computer and only the Controller, will have access to the key for the coding.

Contact data will be stored separately from your coded demographic and medical data. Your sample and/or extracted DNA, and the associated data, will be retained for ten (10) years. You will then be re-contacted to renew your consent. However, you have the right to withdraw your consent at any time and if that happens, the sample/s and associated data, including your personal contact data that you provided, will be destroyed.

Access to samples and data

Samples and/or associated data, which may be made available to researchers outside the EEA, will be transferred in accordance with Chapter V of the **GDPR** and provided that the Controller is satisfied with the ethical approval formally obtained from a Research Ethics Committee. Researchers are required to sign a Material Transfer Agreement and a Data Transfer Agreement and will only receive pseudonymised samples and associated coded data.

Your consent will also authorise the Biobank to share data in an aggregate form through the online biobank sample catalogue.

Contact data will be retained by the Biobank and will not be shared with researchers or any other third parties, unless this is legally required or otherwise if ordered by the Maltese Courts of Justice.

Dwarna portal will access your contact data only for the purpose of facilitating dynamic consent, allowing you to modify your consent options. Dwarna portal will not have access to your demographic and medical data. The IT system for this portal (dwarna.mt) is completely separate from the Biobank's IT system.

Access to data will be under the responsibility of the Biobank Manager, Dr Lidia Ryabova and only authorised delegates. The University of Malta is represented by the Rector, Professor Alfred J. Vella, or his delegate, Professor Charles V Sammut.

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Your rights

You are free to decide whether you agree to become a research participant by donating your samples and data to the Biobank. Participation in the Biobank is voluntary and you have the right to withdraw any consent at any time without giving a reason and without repercussions.

You have the right to request information about any personal data that the Biobank may hold about you, in line with the **Data Protection Act** and the **GDPR**, which grants you the right to access, rectify or erase your personal data. Should you opt to withdraw your participation at any stage, you have the right to decide whether the material already collected, and the data already processed, can remain in use by the Biobank and the scientific collaborators. However, data that would have already been used in research cannot be removed or destroyed. Destruction of your sample will be accompanied by deletion of your associated demographic and medical data and of the code linking them to your personal contact data, so that no further information about you is collected or retained. The contact data will also be destroyed.

Should you wish to exercise your data protection rights, as well as withdrawing your samples from the CMMB Biobank, please contact the Biobank Manager, Dr Lidia Ryabova, on 2340 3272 during office hours or by e-mail at lidia.ryabova@um.edu.mt and at contact@dwarna.mt

Any query in relation to data protection should be directed to the University Data Protection Officer, Dr Luisa Spiteri Baluci, who may be contacted on 2340 3233 during office hours or by e-mail at luisa.spiteri-baluci@um.edu.mt and at dpo@um.edu.mt

You also have the right to lodge a complaint with the Office of the Information and Data Protection Commissioner, by accessing <https://idpc.org.mt/file-a-complaint/> and submitting the online form.

Final words

Please take your time to read this Information Sheet. It is up to you to decide whether or not you take part. If you do decide to take part, and become a research participant, you will be asked to sign the Consent Form. You must consent to the donation of both the biological sample and the associated demographic and medical data. You will be given a copy of the signed Consent Form to retain together with this Information Sheet.

Should you need additional information or wish to communicate any change of address you can contact the Biobank Manager, Dr Lidia Ryabova at DwarnaBio during office hours on 2340 3272 or by e-mail at lidia.ryabova@um.edu.mt and at contact@dwarna.mt