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BIORESOURCE PAPER

Urolithiasis Bioresource

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Urolithiasis is frequent and raises significant health care burden in a working-age population. Its prevalence, in the Azores archipelago, is currently unknown but it is thought to be higher than the overall estimated prevalence. The Azores Biobank (AZORBIO) Urolithiasis samples have been collected in accordance with standard operation procedures (SOPs) to ensure high quality. Each donor provided 30 ml whole blood and 9 ml of urine. If possible, we preferred blood collected from fasting individuals and first morning urine samples. Aliquots of plasma, serum, DNA, RNA and urine, are stored at -80°C freezers. This collection of samples, and data, will be used to investigate the genetic and/or environmental risk factors associated with the disease in this geographical area.

Keywords: AZORBIO; biobank; urolithiasis; biological products; samples; SOPs
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(1) Overview

Project description

Biobanks are essential in research, by having collections of samples and data stored in an organized manner. The biobank AZORBIO of the Specialized Service of Epidemiology and Molecular Biology (SEEBMO) has a collection of biological material and associated data of Azorean patients with different pathologies (Breast cancer, Ankylosing Spondylitis, Diffuse Idiopathic Skeletal Hyperostosis, Chondrocalcinosis, Diabetic retinopathy...). The pathology of interest, Urolithiasis, which is characterized by the presence of calculi in the urinary tract, is a multifactorial clinical entity that results from several pathophysiologic mechanisms [1]. In populations of Europeans ancestry, 5 to 9 % of adults experience a painful precipitation of calcium oxalate in their urinary tracts [2].

In order to study Urolithiasis, two departments of HSEIT, SEEBMO and the Urology Service, initiated a research Project in 2012. AZORBIO stored biological samples – DNA, RNA, plasma, serum, urine – and the associated data collected from the Urolithiasis patients.

An informed consent form and a specific Urolithiasis questionnaire were performed and standard operating procedures (SOP) for the entire circuit were established. All procedures were subsequently assessed with the first ten samples received on AZORBIO. Nucleic acid samples were subjected to quality control to verify the concentration, purity, integrity and functionality. The documentation produced (informed consent, informative document, questionnaire and standard operating procedures) contributed for the standardization of the entire circuit for samples of Urolithiasis. With this work the AZORBIO will ensure good Urolithiasis quality samples and data contributing to the improvement of health care and reduce morbidity of this disease.

Classification (1)

Human

Species

N/A

Classification (2)

Biological samples and associated data.

Context

Spatial coverage

Description: Portugal, Azores, Terceira, Angra do Heroísmo.

- Northern boundary: 38.799584/- 27.202835
- Southern boundary: 38.641545/-27.209015
- Eastern boundary: 38.701588/- 27.040787

- Western boundary: 38.742837/- 27.379303

Temporal coverage
The project was initiated in 2012. Sample collection will proceed until 2017.

Temporal coverage for accessibility
N/A

(2) Methods
Steps
All the information regarding this collection is available on www.seebmo.org:

- Selection of Urolithiasis patients (N20-23; ICD-10 classification Version:2008), and call for a medical consultation;
- Patients are invited to participate in the study, and sign the informed consent after understanding the goals of the project;
- Carrying out the questionnaire which assesses a number of factors that may contribute to disease;
- Collection of biological products (30 ml of blood and about 9 ml of urine);
- Transport of the biological products and documentation from the collection center to the biobank;
- Verification of documents and biological products in AZORBIO;
- Processing of biological products to obtain the following final products: DNA, RNA, Plasma Serum and urine;
- Quality assessment of the nucleic acid samples;
- Storage of the samples in ultra-low temperature freezers.

Stabilization/preservation
- EDTAK3 tubes (3.0 ml whole blood)
- PAXgene tubes (2.5 ml whole blood)
- Serum Separator tube (8.5 ml whole blood)
- Vacuette Urine tubes with boric acid (9.5 ml urine)

The tubes are transported at room temperature in a category B packaging (UN3373).

Type of long-term preservation
Frozen

Storage temperature
DNA, RNA, Plasma, Serum and Urine are stored in separate freezers at -80°C (indefinite storage).

Shipping temperature from patient/source to preservation or research use
Room temperature (18 to 25°C) if sent from HSEIT in the pre-determined time interval (30 minutes). If it exceeds this time interval refrigeration of biological products is suggested.

Shipping temperature from storage to research use
Aliquots of DNA, RNA, Serum, Plasma and Urine will be sent frozen (-20°C).

Quality assurance measures
AZORBIO is presently not recognized by the Portuguese authority: Comissão Nacional de Proteção de Dados (CNPD). The submission of this collection to CNPD is currently being prepared.

The subsequent Best Practice Norms are followed: OECD (2007) Best Practice Guidelines for Biological Resource Centres [3] and ISBER (2012) Best Practices for Repositories Collection, Storage, Retrieval, and Distribution of Biological Materials for Research [4].

SEEBMO is currently on a certification process (ISO 15189).

Infrastructure and facilities
AZORBIO has sufficient air conditioning that provides air circulation to maintain ambient temperature ≤22°C in order to prevent excess freezer wear and early failure. SEEBMO has continuous electric power supply. In case of power failure, 2 UPS (Uninterruptible Power Supply) and 1 Electric Generator will be able to maintain the electric supply for 3 and 72 hours, respectively. There is temperature control software which sends a local and remote alarm, email and message, in case of temperature failure. Adequate backup capacity for low temperature units is always maintained. Stored biospecimens are split into two sets of aliquots, each set stored at different locations.

Standards operating procedures (SOPs)
Procedures in a standardized written format were developed to function as Manuals, readily available to all laboratory personnel, including specimen handling, laboratory procedures for specimen processing, procedures for shipping and receiving specimens, quality control of biospecimens, documentation for reporting accidents, procedures for disposal of wastes.

SOPs are not publicly available but can be provided, if requested.

Information and communications technology (ICT)
A laboratory information management system (LIMS) software, recently acquired, is used for the sample management as well as storage of all the associated data.

Source of associated data
Associated data was collected with a specific questionnaire (IMP.EBM.002), available on www.seebmo.org. The questionnaire was produced based on previous research studies on Urolithiasis risk factors. Sociodemographic and socioeconomic characteristics, anthropometrics measurements, physical activities, personal habits, nutrition and health information were included. It is a physician assisted paper questionnaire which is filled just before the collection of biological specimens.

Ethics statement
This project was planned according to the regulations stated in the Portuguese Law for Genetic Information and Health Information act in 2005 (Law 12/2005 of 26 January). This Law states that patients must be informed by their doctors and grant written consent before any

samples are collected. As a result, the Ethics Committee at Hospital de Santo Espírito da Ilha Terceira approved the Informed consent form, which is provided to patients as well as the information sheet and the questionnaire. The authorization for this study was granted by the Ethics Committee on the 3rd February of 2012.

The informed consent form (available on www.seebmo.org) allows the use of samples and related data in specific Urolithiasis research and in future investigations directly or indirectly associated with them. If participants wish to withdraw their samples and associated data they can do so, after filling the appropriate document, available on www.seebmo.org.

Participant confidentiality: all samples and data are codified and can only be linked back to the participant in HSEIT through the physician.

Constraints
AZORBIO is located in Terceira Island, in the Azores archipelago.

(3) Bioresource description

Object name
LU (Lithiasis Urinary)

Bioresource name
Urolithiasis

Bioresource location
Serviço Especializado de Epidemiologia e Biologia Molecular, Hospital de Santo Espírito da Ilha Terceira, Vinha Brava (Edifícios dos Serviços de Desenvolvimento Agrário). 9701-880 Angra do Heroísmo, Terceira, Azores, Portugal.

Urolithiasis samples are collected in HSEIT and then sent to SEEBMO for storage in AZORBIO.

Bioresource contact
- Tel./Fax: +351 295 216263
- E-mail: azorbio@seebmo.org

Bioresource URL
http://www.seebmo.org/

Identifier used
N/A

Bioresource type
- Pathology
- Genetic variant
- Population based

Type of sampling
Disease based (cross-sectional).

Anatomical site
N/A

Disease status of patients/source
Urolithiasis patients

Clinical characteristics of patients/source
Urolithiasis patients of different ages and genders. The inclusion criteria is N20-23; ICD-10 diagnostic.

Vital state of patients/source
In vivo

Clinical diagnosis of patients/source
Confirmed Urolithiasis by biochemical analysis (blood and urine).

Pathology diagnosis
Urolithiasis (N20-23; ICD-10)

Control samples
N/A

Biospecimen type
AZORBIO stores 2 x 500 μl aliquots of DNA obtained by "Salting- Out" method and 1 aliquot of 200 μl extracted by EZ1 (Quiagen). Plasma and serum are stored in 4 x 800 μl aliquots of each. Urine is stored in three types of aliquots that will permit a large range of future testing: a low speed centrifugation pellet (2x100μl), the cleared supernatant after centrifugation (2x800 μl), and an additional aliquot of the pre-centrifuged urine (2x800 μl).

Size of the bioresource
At the moment the AZORBIO have Urolithiasis samples from 40 patients however we expect to achieve about 200 individuals, by the end of the project.

Release date
The samples will become available from 2018 onwards.

Access criteria
Research projects that wish to have access to AZORBIO samples must submit the project to the Ethics Committee of the HSEIT for approval.

AZORBIO must be referred in all published articles which have used AZORBIO samples for the studies.

(4) Reuse potential

The risk of Urolithiasis is higher in individuals with a family history of Urolithiasis [5]. It is estimated to be more than 2.5 times greater in those individuals [6]. A combination of genetic predisposition as well as similar environmental exposures may be the cause for it. A polygenic inheritance has been proposed to account for the familial tendency [7]. However, there is limited information on the genes that contribute to the risk of the common forms of Urolithiasis.

The samples and data stored in AZORBIO may be used for biochemical, genetic and epidemiological studies, to investigate the disease. In conclusion, the Urolithiasis samples and data contribute to the improvement of health care and reduction of morbidity of this disease.

Author roles
- Mónica Seidi, Raul Rodrigues, Maria Ribeiro: Collector of data or samples

- Bruna Parreira: Bioresource technical implementation
- Ana Rita Couto: Bioresource manager
- Raquel Meneses: Bioresource technician
- Manuela Lima: Curator
- Jácome Bruges-Armas: Director and creator

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