

The Clinical Bank of BBMRI.mt

Vella, J., Borg, J., Grech, L., Galdies, R., Scerri, J., Cassar, W., Scerri, C.A., Camilleri, A., Bezzina-Wettinger, S., Farrugia, R., Camilleri, G., Pace, N.P., Zammit, E., Said Conti, V., Grech, G., Saliba, C., Soler, D., Vella, N.R., Borg, I., Said, E., Camilleri Podesta, M.T., Ellul, B., Felice, T., Grima, D., Ebejer, J.P., Felice, A.E.

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The Malta BioBank / BBMRI.mt

The Malta BioBank forms part of the new inter-faculty Centre for Molecular Medicine and BioBanking at the University of Malta and is the BBMRI-ERIC national node of Malta (BBMRI.mt) (Refer to Poster 82). The business model of the Clinical Bank is based on a collaboration between the University of Malta, the Department of Health and Mater Dei Hospital's departments including Pathology, Paediatrics, Neurology and Oncology.

The clinical catalogue holds a number of disease collections including: the Globin Bank; Parkinson's Disease (PD); Diabetes; Multiple Sclerosis; Renal Disorders; various cancers including breast, colon, lung and gastric cancer; and a number of rare diseases.

Biobanking Research

Research at the Centre for Molecular Medicine and BioBanking is supported by various funds: the Research, Innovation and Development Trust (RIDT) through voluntary organisations such as the ALIVE Charity Foundation for breast cancer research and the Life Cycle Organisation for renal diseases; EU funding e.g. Geo-Parkinson project (FP5); ImaGenX project - Italia Malta Genome Breast Cancer Cross Border Risk Surveillance; and fellowships such as the Malta Government Scholarship Scheme (MGSS) and Strategic Education Pathways Scholarships (STEPS).

Globin Bank

The Globin Bank includes a catalogue of samples of patients and carriers with Beta Thalassaemia, Alpha Thalassaemia and haemoglobinopathies including haemoglobin F Malta, haemoglobin St Luke's, haemoglobin Valletta and patient samples with High Persistence of Foetal Haemoglobin having the Kruppel Like Factor 1 (KLF) gene mutation (Refer to Poster 43).

Parkinson's Disease collection

This collection forms part of the Geo-Parkinson project and includes DNA samples from 200 Maltese cases with PD / parkinsonism and 300 age and gender matched controls and associated lifestyle questionnaires.

Genes known to be involved in the onset and progression of PD are being sequenced by NGS. The collection has been tested for a number of SNPs e.g. the common Mediterranean Leucine-Rich Repeat Kinase 2 (LRRK2) and alpha-synuclein mutations, SNPs thought to be involved in chemical detoxification, the local tetrahydrobiopterin (BH4) mutations and other mutations in genes such as the Macrophage Migration Inhibitory Factor (MIF) and Methylenetetrahydrofolate Reductase (MTHFR).

Diabetes Collection

The Diabetes Collection includes a distinctive collection of clinical data and DNA from newly-diagnosed Type 2 Diabetes Mellitus (T2DM) patients that are useful in exploring genotype-phenotype associations where the phenotype is as yet unaltered by drugs and lifestyle changes.

Another collection includes DNA and serum specimens of both Maltese and Libyan T2DM patients with advanced end-organ complications. These are being used to explore the genetics of risk and progression to vascular complications in T2DM. There is an ongoing population-based collection that aims to identify the prevalence of pre-diabetes in the Maltese population.

Rare Disorders

The Clinical Bank holds a number of rare disorders including:

Renal

The rare renal disorder collection includes samples from patients and families with Congenital Nephrotic Syndrome, Congenital Anomalies of the Kidney and Urinary Tract (CAKUT) and Bartter Syndrome.

Mutation profiling of Maltese patients with Finnish-type Congenital Nephrotic Syndrome will determine if any modifier genes are responsible for a milder clinical phenotype seen locally. A homozygous nt3478(C→T) R1160X NPHS1 nonsense mutation in exon 27 has already been identified in Maltese cases. Genetic analysis on CAKUT samples will contribute to a deeper understanding of the disease.

Familial Breast Cancer

High penetrance genes associated with an increased risk to cancer development are exemplified by the BRCA1 or 2 mutations in breast cancer. In western countries, familial cases account for 10% of breast cancer. BRCA1 or 2 mutant genes, are associated with a higher incidence of the disease in close relatives, increasing significantly the risk within a family. Inherited mutations in p53 (Li-Fraumeni Syndrome) and Phosphate and Tensin homolog (PTEN) (Cowden Syndrome) have a high incidence of breast cancer development, but the syndromes are very rare.

121 samples were collected from the Malta BioBank between 2012-2014 of which 72 were cancer patients. 29% of cases had the BRCA1 or 2 mutations. Further samples shall be collected and banked and exome sequencing will help to identify other variants.

Mitochondrial

A total of 17 patients identified with mitochondrial disorders including mitochondrial myopathies, Mitochondrial Encephalomyopathy, Lactic Acidosis and Stroke-Like Episodes (MELAS) and Kaerns Sayre Syndrome were identified from a patient register at Mater Dei Hospital. Genetic analysis will be carried out on patients and their families to increase the scientific understanding of these rare syndromes.

Sudden Cardiac Death

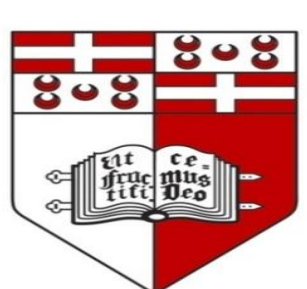
Another project focuses on research into Sudden Cardiac Death (SCD) in subjects under 40 years old. 60 SCD cases over a 10 year period were identified from Mater Dei Hospital's mortuary records. Mitochondrial genome sequencing will help to identify mutations that might be associated with SCD and determine if certain mutations are specific to certain mitochondrial DNA haplogroups. Family pedigrees will also be investigated.

BioBank Networks

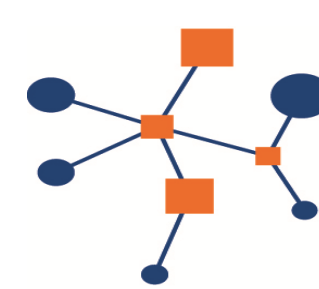
The Malta BioBank is a founding partner in EuroBioBank and the clinical catalogue can be accessed online from:

<http://www.eurobiobank.org/en/services/CatalogueHome.html>

BBMRI.mt is also a partner in RD-Connect with a focus on biobanking and engagement of biobanking in the Euro-Mediterranean region. It also forms part of the Electronic Infrastructure for Thalassaemia Research Network (ithanet). Through BBMRI-ERIC ADOPT, BBMRI.mt will further develop its Clinical Bank.



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