

Malta BioBank (BBMRI.mt) in RD-Connect

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The Malta BioBank (BBMRI.mt); Centre of Molecular Medicine and Biobanking, University of Malta is a founding partner in RD-Connect. It is registered on the RD-Connect Registry and Biobank Finder. More details in www.um.edu.mt/biobank.

BBMRI.mt participated in the BBMRI-LPC whole exome sequencing (WES) call with two collaborative research projects: undiagnosed mitochondrial disorders and undiagnosed neuropathies. WES and bioinformatics analysis were conducted at CNAG-CRG. Phenotypic data were recorded on PhenoTips. Exome data was analysed on the RD-Connect Genome-Phenome Analysis Platform.

Biobank-led research in globin gene disorders focused on strategies to re-activate foetal haemoglobin to therapeutic levels in thalassaemia patients. DNA variants in the human Kruppel Like-Factor 1 (KLF1) gene and others that regulate gamma- to beta globin gene switching were characterised. 420 subjects with HbA2 between 3.1 and 4.5% were retrospectively collected from the biobank. More than half of these subjects had a mutation in KLF1.

An increasing number of studies point to genetic changes as the cause of congenital renal malformations. 10 patient samples were subjected to whole-genome sequencing. The coding and splice-site regions of 96 genes implicated in non-syndromic CAKUT and involved in kidney development are being studied.

Another project focuses on rare monogenic subtypes of diabetes e.g. MODY. 30 probands and their families with suspected MODY were recruited. Preliminary analysis has identified rare and novel mutations in several genes, including GCK, HNF1 α and HNF1 β , WFS1 with evidence for several founder effects.

BBMRI.mt was instrumental in placing Rare Diseases (RD) on the agenda of Malta's Presidency of the EU. High level meetings about RD were organised in parallel to the Informal meeting of EU Health Ministers. BBMRI.mt works closely with the National Alliance for Rare Diseases Support – Malta to create awareness about RD. The biobank is seeking to reach into community health professionals to strengthen Stakeholder Engagement, Education and Dialogue (SEED).

Words 301 max 300