BBMRI.mt participates in collaborative BBMRI-LPC project on Mitochondrial Disorders

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The Malta BioBank (BBMRI.mt) participated in the BBMRI-LPC WES call. This was a collaborative research initiative jointly organised by BBMRI-LPC, EuroBioBank, RD-Connect and Centro Nacional de Análisis Genómico (CNAG-CRG). The goal was to sequence 50 exomes from patients with genetically undiagnosed mitochondrial disorders whose samples were banked within EuroBioBank.

A total of 50 patients from Malta and Turkey consented to participate in the study.The Maltese cohort included 13 probands. WES and bioinformatics analysis were carried out at CNAG-CRG. Phenotypic data of each participant was recorded on PhenoTips. Exome data was analysed on the RD-Connect Genome-Phenome Analysis Platform.

A comparative analysis of rare autosomal recessive mutations shows that some patients share the same variants. Rare missense mutations in the mitochondrial cytochrome B gene (MT-CYB) at positions 14766 and 15326 were present in 7 and 12 probands respectively. The mtDNA mutation at position 15326 (rs2853508) was not present in the reference Maltese Exome database, whereas that at position 14766 (rs193302980; rs57236041) had a frequency of 59%.

The infrastructure for data sharing in rare disease research set up through RD-Connect will aid in establishing a genetic diagnosis for these rare disease patients. This initiative was supported by the National Alliance for Rare Diseases Support – Malta.