

MITOCHONDRIAL DNA TYPING OF THE MALTESE (O)

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The Maltese Archipelago has an area of 316km² and over 475,000 residents. The islands have a rich demographic history. Although historical records trace population origins to the Temple people, contemporary Maltese are descendants from those who re-populated the islands at the turn of the first millennium AD.

Maltese mitochondrial DNA (mtDNA) data is not publicly available. A new high-quality mtDNA control region (CR) dataset was set up to evaluate Maltese maternal lineages. A total of 798 samples were collected randomly with associated ancestry data from Malta and Gozo. This population collection is archived in the Malta BioBank (BBMRI.mt). The EMPOP protocol was used to amplify and sequence a subset of 300 samples with a minimum of four EMPOP sequencing primers according to forensic quality guidelines.

So far, 284 full CR sequences were typed of which 190 were unique (67%). mtDNA haplotypes were checked on EMPOP and Phylotree and haplogroup frequencies were calculated. The majority of the observed Maltese mtDNAs (77%) could be attributed to West Eurasian haplogroups: H (35%), T (18%), K (12%), J (5%), U (5%), X (1%), W (1%). African mtDNA lineages were also present: L1 (0.4%), L2 (10%), L3 (1%), M1 (0.4%). Other lineages were observed at frequencies of 0.7% - 6%. The frequency of haplogroup H in Malta was similar to that in Sicily and Southern Italy (35%). Sub-Saharan L2 and L3 lineages were present at higher frequencies than in Sicily and Southern Italy. The observed sub-clade L2a1c6 was previously reported in Spain (Galicia) and Portugal and is estimated to be 1,700 years old. The presence of L lineages in Malta represents the sub-Saharan gene flow into Europe.

This population genetics research provides a first insight into the matrilineal origins of the Maltese. The dataset can be used as the first national reference database for mtDNA applications in forensic and missing persons casework and population genetic studies.