



MALTA HUMAN GENOME PROJECT

OBJECTIVES OF THE RESEARCH

The goals of the Malta Human Genome Project were to provide a complete and accurate sequence of a reference Maltese genome making up the DNA of our population. The project involved a random collection of 400 Maltese DNA samples representing 0.1% of the Maltese population. A number of clinical genetic disorders were also examined.

MAIN FINDINGS TO DATE

The research presented pools of mixed DNA from Maltese citizens and analyzed using state of the art equipment capable of reading whole human genomes. This enabled us to conduct studies in Malta that were not possible before and creating the very first Maltese draft genome through sequencing. The Maltese reference genome serves as a platform for comparing clinical genetic disorders and tested against in order to ascertain and distinguish between functional as opposed to innocuous DNA variants.

SOCIO-ECONOMIC IMPACT

This new knowledge will facilitate the creation of a browsable Maltese reference genome. Clinicians can use this database of information for predicting disease susceptibility or for diagnosing and tailoring disease treatment of an individual (personalised medicine). Patients suffering from haemoglobin disorders such as β -thalassaemia and more complex diseases, such as type 2 diabetes mellitus are benefitting.

THE RESEARCH TEAM

The Malta Human Genome Project is being coordinated by Prof. Alex E. Felice and co-PI Prof. Joseph Borg with the participation of Laura Grech, Malcolm Pace and Lidia Ryabova together with other staff currently based at the Laboratory of Molecular Genetics, Department of Physiology and Biochemistry, University of Malta and Thalassaemia Clinic, Mater Dei Hospital, Msida, Malta, Molecular Diagnostics from the Dept. Of Pathology, Mater Dei Hospital, Malta, Erasmus Medical Center, Rotterdam, The Netherlands, and BGI Genomics,

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