In Collaboration with the Laboratory of Molecular Genetics, Department of Physiology and Biochemistry;

The Effects of Trans Regulatory Loci on Globin Gene Control

Important clinical models such as people with hereditary persistence of foetal haemoglobin (HPFH) or others such as carriers of foetal/adult haemoglobin variants (that include Hb F Malta1 and Hb Valletta) can serve to identify what and how Hb F is regulated and controlled. A large collection of Hb F Malta 1/Hb Valletta heterozygotes are being tested both at the Hb F profiling level, and genetic level. Both cord and adult blood is already available and additional patients are being collected prospectively. Additional patient material from β thalassaemia trait patients are also being identified by testing that is conducted at the Laboratory of Molecular Genetics, University of Malta. It is anticipated that by comparing the different Hb F expression profiles of adult β thalassaemia trait patients as well as the haemoglobin chain profiles of cord Hb F Malta 1/ Hb Valletta vs. the genetic profiles obtained from KLF1, BCL11A and MYB will highlight important features that these genes exert on the globin gene locus itself and to what extent are they modifying the phenotype. Furthermore, adult patients with Hb F Malta 1 / Hb Valletta are tested by real time PCR to quantify the foetal mRNA globin transcripts, and including also a number of re-sequencing of genes in the large Maltese family that exhibit HPFH. Together this should summarize the role of important genes and their protein products in well-defined models.