



# HAEMOGLOBIN GENE SWITCHING BY *KLF1* REGULATOR

## OBJECTIVES OF THE RESEARCH

The primary objective of this research was to further characterize key the molecules responsible for developmental control and regulation of haemoglobin, an important molecule responsible for carrying oxygen in blood. The project entailed extensive use of genetic engineering, transcriptomic and functional assays in order to elucidate the underlying mechanisms.

## MAIN FINDINGS TO DATE

A number of important DNA mutations have been uncovered through extensive genome sequencing. Their effects were further studied by using advanced techniques based on RNA and protein analysis and allowed us to characterize the variable expression of an important human gene called *KLF1*. This gene was later discovered to be critically important for haemoglobin production, the oxygen carrier molecule in human blood.

## SOCIO-ECONOMIC IMPACT

The results pave the way towards further research in globin expression and towards specialized and tailor-made therapies (personalised medicine). These therapies are aimed at treating conditions such as  $\beta$ -thalassaemia and sickle cell disease worldwide.

## THE RESEARCH TEAM

The research group is being led by Prof. Alex E. Felice and Prof. Joseph Borg, with the active involvement of the following team member; Laura Grech, Jeremy Cutajar, Mary Rose Caruana, Christian A. Scerri, Ruth Galdies and Alex Camilleri. The team is currently based at the Laboratory of Molecular Genetics, Department of Physiology and Biochemistry, University of Malta and Thalassaemia Clinic, Mater Dei Hospital, Msida, Malta and the Department of Applied Biomedical Science, Faculty of Health Sciences, Univeristy of Malta, Msida Malta.

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