THALASSEMIA

"Thalassaemia" refers to a genetic disorder of an extremely heterogenous group which is characterised by a reduced or erroneous production of haemoglobin. The disease was originally widespread in the Mediterranean Basin, South-East Asia and various countries in equatorial Africa. However, following its persistent migration over planet it is today verifiable in almost all regions of the globe.

Until now correct transfusion therapy has been the only available treatment for patients affected by Thalassaemia Major. It is only possible to cure the disease by bone marrow transplant and in this way replacing diseased cells with healthy ones.

The probability of a child being born affected by Thalassaemia Major is 25% if both parents are carriers. Present method of diagnosing Thalassaemia relies on detection of DNA defect of the fetus and procurement of fetal tissue.

A less severe disease than Thalassaemia Major is Haemoglobin H disease. Since most people affected by Haemoglobin H disease have only moderate anemia, in such cases invasive prenatal diagnosis and/or termination of pregnancy is usually not needed but non-invasive prenatal prediction is desirable. The Department of Obstetrics and Gynaecology, Faculty of Medicine, HKU is now developing a non-invasive prediction test to reduce the pain and suffering of fetus and mothers.