Thalassemias

Thalassemias are a group of hereditary anaemias which are prevalent in the Mediterranean countries, the Middle East and Asia. This group of diseases can be broadly divided into alpha-thalassemia (α-thal) and beta-thalassemia (β-thal). Depending on the severity of disease, thalassemias can also be classified into thalassemia minor, thalassemia intermedia and thalassemia major. The underlying cause of this group of diseases is deficient synthesis of globin chains in red blood cells.

A normal level of haemoglobin is needed in red blood cells in order to efficiently carry oxygen to all body parts. Each haemoglobin is composed of two alpha-globin chains and two beta-globin chains. The synthesis of alpha-globin chains is dictated by two pairs of genes. These two pairs of genes are inherited, one pair from each of the parents. Individuals with one or two abnormal genes suffer from thalassemia minor and are also known as thalassemia carriers. These patients are asymptomatic and their carrier status is usually ascertained incidentally through routine blood tests. However, when two α-thalassemia carriers get married and reproduce, there is an increased chance for their offspring to carry three or even four abnormal genes. As a result, their offspring would suffer from α-thalassemia intermedia or α-thalassemia major, which are serious anaemias. α-Thalassemia major is usually evident before birth resulting in premature death.

The underlying mechansim of beta-thalassemia is essentially the same and there is a decrease in beta-globin chain synthesis. The synthesis of beta-globin chains is dictated by a pair of genes. Individuals with one abnormal gene are carriers and individuals with two abnormal genes suffer from β-thalassemia intermedia or β-thalassemia major. Children suffering from β-thalassemia major are not able of producing sufficient functional hemoglobin and require regular blood transfusions lifelong.

In Hong Kong, the carrier frequency of α- and β-thalassemia is about one in 12. If both parents are carriers of the same type of thalassemia, there is a 25% chance in each pregnancy to give rise to a baby with thalassemia intermedia or major. At present, thalassemia carriers can be identified through blood testing. Therefore, couples contemplating marriage or pregnancy should undergo testing for thalassemia in order to effectively prevent the birth of affected babies.

Clinical Genetic Service
Department of Health
(2012 Version)