

Editorial

Using NGS to diagnose thalassemia

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Thalassemia is a congenital hemolytic anemia syndrome caused by an imbalance of globin synthesis. By genetic classification, the α -globin gene mutation usually involves deletions removing one or more entire genes and β -globin gene mutation will decrease the production of the corresponding protein.^{1,2}

The incidence of thalassemia in Thailand is high or we could say that thalassemia is the most common congenital hemolytic anemia in Thailand. We are sufficiently familiar to diagnose thalassemia among patients presenting clinical signs of anemia, jaundice, hepatosplenomegaly, symptoms those with thalassemia face. With clinical anemia, jaundice and thalassemia diagnosis suspected, patients with thalassemia usually present bone complications and need chronic blood transfusion which may be followed by transfusion reaction or other complications followed by iron overload.⁴ We usually perform hemoglobin electrophoresis or chromatography (Hb typing), which will detect abnormal hemoglobins to confirm the diagnosis of thalassemia. Nevertheless, patients with thalassemia carrier status or thalassemia traits may present no anemia, no jaundice and no hepatosplenomegaly, but we can screen using red cell index (MCV, MCH), *osmotic fragility* (OF) or dichlorophenol indophenol precipitation (DCIP) and then can confirm by Hb electrophoresis. To identify α -thalassemia carrier status, we can use multiplex-Gap-PCR for large deletion or Amplification Refractory Mutation System PCR (ARMS-PCR) for point mutation, but for those cases revealing few uncommon abnormal hemoglobin signs, sometimes we cannot identify what hemoglobin type they represent.⁵ Currently, advances in analyzing patient's

DNA test results provide another option to diagnose genetic diseases like thalassemia. Gene diagnosis like Next Generation Sequencing (NGS) will help physicians to correctly diagnose; and further, provide accurate, specific treatment, decrease disease complications, or even offer family counselling for thalassemia couples. Similar to this article, the authors showed detection of rare mutations for α -thalassemia gene by NGS.⁶ However, the advance diagnosis test is now expensive and not many patients or hospitals can access this test. In the future, we may develop or modify this test to be more affordable or combine with other method to diagnose uncommon thalassemia types. It may be possible to study the gene editing treatment for thalassemia, constituting the most common congenital hemolytic anemia syndrome in Thailand.

Reference

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