NEW SCREENING OPTIONS FOR PATIENTS WITH THALASSEMIA: LEBANON EXPERIENCE

Andrusha A.B., Mohamad B.S.

Харьковский национальный медицинский университет, Харьков 61000, Украина

alina.andrusha@meta.ua

Keywords: thalassemia, laboratory diagnostics, screening

Introduction: Morbidity and mortality in beta thalassemia are caused by anemia and iron overload. The prognosis of patients with thalassemia major depends on the patient's adherence to treatment programs. The early identification of the patients at the preclinical stage contributes to the implementation of relevant recommendations that improve the quality of life and allow manage disease more effectively. Our aim is assess the main criteria for effectiveness of using laboratory screening methods for detecting thalassemia.

Methods: The Chronic Care Center (Beirut, Lebanon) organized screening campaigns in order to identify new carriers within the general population and high risk group. By performing complete blood count, and if mean corpuscular volume (MCV) is decreased, then order hemoglobin electrophoresis with quantitative adult hemoglobin (HbA) and fetal hemoglobin, also iron studies are done (iron and ferritin). The Genetic Laboratory provided prenatal tests: testing of chorionic villous sampling (at 10-12weeks gestation) or amniotic fluid, and Pre-implantation genetic diagnosis for beta-thalassemia

Results: Screening programs showed a carrier rate of around 2.3% in the general population, and 4.0-41.0% in high risk groups. There has been a significant decrease in the number of new cases of thalassemia in patients, reflecting the efforts being made to prevent this disease. In our opinion, two disadvantages of this screening method are obvious. Firstly, some limitations are faced in reaching a complete eradication of the disease, mainly due to the fact that abortion is illegal and pitfalls and incorrect implementation of the premarital law. Secondly, the major pitfall in the law is that only persons with a MCV of >70.0 fL are asked to perform further hemoglobin electrophoresis.

Conclusion: Screening strategies are very important for the prevention of a specific disease especially in high risk population by early identification of the patients and identifying carriers. Although medications for thalassemia are prescribed only for the correction of symptoms and complications, and there is no drug therapy for the disease itself, but diagnosis of thalassemia at the preclinical stage can improve the quality of life and prognosis of patients, as well as effectively manage the disease.