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شبكة المعلومات الجامعية

التوثيق الالكتروني والميكروفيلم

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جامعة عين شمس

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Faculty of medicine
Pediatric department

Some Trace elements in Patients with β thalassemia major

(Essay)

Submitted For Partial Fulfillment of Master Degree In Pediatrics

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B THALASSEMIA

β-Thalassemia

Definition

Thalassemia is a hereditary anemia resulting from defects in hemoglobin production(**Higgs et al.,2001**) . β -Thalassemia, which is caused by a decrease in the production of β -globin chains , affects multiple organs and is associated with considerable morbidity and mortality(**Cunningham et al., 2004) .**

Accordingly, lifelong care is required and financial expenditures for proper treatment are substantial.Thalassemia is among the most common genetic disorders worldwide; 4.83 percent of the world's population carry globin variants, including 1.67 percent of the population who are heterozygous for α-Thalassemia and β -Thalassemia. In addition, 1.92 percent carry sickle hemoglobin, 0.95 percent carry hemoglobin E, and 0.29 percent carry hemoglobin C. Thus, the worldwide birth rate of people who are homozygous or compound heterozygous for symptomatic globin disorders, including α -Thalassemia and β -Thalassemia, is no less than 2.4 per 1000 births, of which 1.96 have sickle cell disease and 0.44 have thalassemias (**Angastiniotis et al ., 1998) .**

Molecular and cellular pathology

β -Thalassemia is caused by any of more than 200 point mutations and, rarely, by deletions(**Angastiniotis et al ., 1998) .**

Thalassemia is clinically heterogeneous because various genetic lesions variably impair globin-chain synthesis. However, genotypic variability at known loci is often insufficient to explain the disparate phenotypes of individual patients with the same genotype. Disparity between genotypes and phenotypes is particularly marked in thalassemia intermedia and hemoglobin E thalassemia . However, the known genetic factors are insufficient to account for the marked variability, and other genetic modifiers may exist **(Weatherall .,2001) .**

Recently, an α-hemoglobin–stabilizing protein was identified that binds to and stabilizes free α- chains, thereby blocking the production of reactive oxygen species and reducing oxidative damage to erythrocytes. This protein appears to modulate the clinical picture of β - thalassemia in a murine model but in human studies has not been found to modify thalassemia. Hemolysis and ineffective erythropoiesis together cause the anemia that occurs in thalassemia. The relative contributions of these two pathologic processes differ in various forms of thalassemia**(Pootrakul et al .,2000) .**

The bone marrow of patients with thalassemia contains five to six times the number of erythroid precursors as does the bone marrow of healthy controls, with 15 times the number of apoptotic cells in the polychromatophilic and orthochromic stages**(Centis et al .,2000).**

Accelerated apoptosis, the major cause of ineffective erythropoiesis, is caused by excess γ -chain deposition in erythroid precursors**(Pootrakul et al ., 2000)** .