Beta-Thalassemia and HB lepore heterozygotes: phenotype-genotype correlation

STELLINGEN

behorende bij het proefschrift

ß-THALASSEMIA AND IIB LEPORE HETEROZYGOTES: PHENOTYPE-GENOTYPE CORRELATION

van

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1. Hb H disease when associated with the heterozygous state for Hb New York, has a phenotype which is more severe while the number of erythrocytes with Hb H inclusion bodies is lower.

2. The coinheritance of a δ- and ß-thalassemia allele should be considered in the presence of a hypochromic mycrocytic anemia with normal Hb A_2 levels in Mediterranean populations.

3. The phenotypic differences associated with point mutations in the proximal and distal CACCC boxes of the promoter region of the ß-globin gene depend on the different binding specificities and affinities of these two motifs for erythroid factors.

4. The single strand conformation polymorphism (SSCP) procedure is a useful technique for the screening of mutations in hemoglobinopathies.

5. Prenatal determination of the fetal RhD type by DNA amplification is a reliable and rapid method for the management of Rh alloimmunization.

6. The homozygous state for the dominant form of Hereditary Spherocytosis, Band 3 Coimbra mutation (488 Val→Met), is associated with hydrops fetalis, and a severe, transfusion dependent, phenotype with metabolic acidosis.

7. The association of a homozygosity for a Pyruvate Kinase deficiency and a heterozygosity for Hb Lepore Baltimore (δ^{68Leu}-β^{84Thr}) creates a phenotype of mild chronic hemolytic anemia with hypochromia, microcytosis and splenomegaly.

8. The implementation of molecular technology in the developing countries is essential for the prevention and control of emerging and endemic infectious diseases.

9. The use of erythropoietin to treat anemia in premature infants reduces their transfusion needs.

10. In infants with neuroblastoma, the hyperdiploid tumor DNA is associated with a favorable prognosis, while N-myc amplification is associated with a poor prognosis.

11. It is easier to climb a mountain than to come down.

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