LEPORE HEMOGLOBINOPATHY

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Introduction: Hb Lepore represents a small group of structurally abnormal Hbs that result from in-frame fusion between the 5′ end of the δ-globin gene and the 3′ end of the β-globin gene is defined, formed by unequal crossover or gene conversion events during meiosis. The βδ fusion (hybrid) chain is synthesized inefficiently and normal β and δ chain production is abolished. The homozygotes show thalassaemia intermediate and heterozygotes show thalassaemia trait.

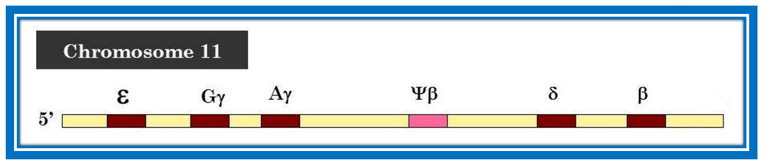

Fig.1. Hemoglobin genes [1]

In Hb Lepore, transcription is from the δ globin promoter so the hybrid gene is expressed at the same low level as δ (~2% of the β). Hb Lepore is a β-thalassaemia allele, anti-Lepore is essentially normal.

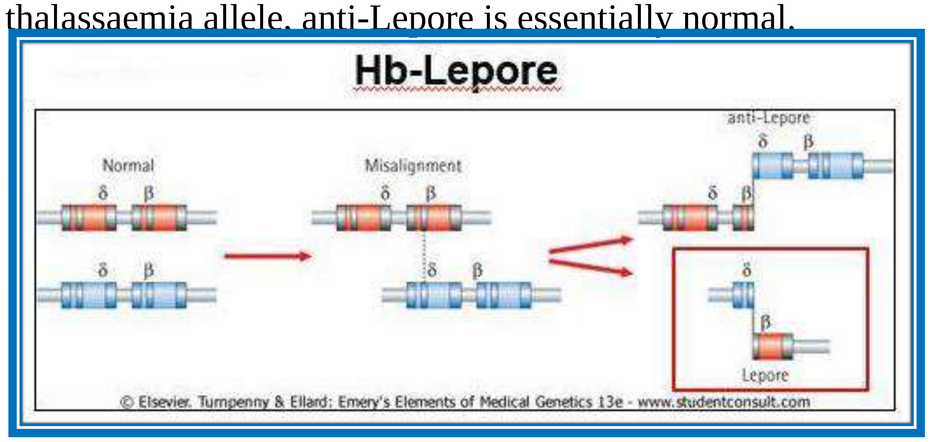

Fig.2. Mechanism of unequal crossing over which generates Hb Lepore and anti-Lepore. [2]

Keywords: Hemolytic anemia, hemoglobinopathy, Lepore.

Purpose: Presentation of the first case of Lepore hemoglobinopathy from the activity of Hematology and General Pediatrics Department, Institute of Mother and the Child.

Bibliography:
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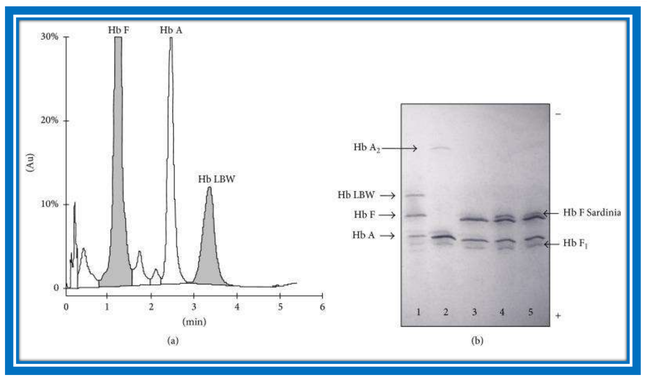
Material and methods: Paediatric patient, girl, age 11 years presents moderate anemia, pronounced pallor of skin, mucous and refracterity to the previous antianemic therapy. On the basis of the complete blood count, the clinical findings, and the electrophoretic studies it was possible to establish the diagnosis of Lepore hemoglobinopathy.

Anemic syndrome: General pallor and pale conjunctivae and fingernails, Tachycardia, tachypnea, and hypotension. Lab test – complete blood count: Hb – 99g/l, RBC – 3.0 x 10^6u/l

Lab test: total bilirubin 13.8 μmol/l, indirect bilirubin – 12.4 μmol/l . Ultrasound exam: moderate hepatomegaly.

Hemoglobin electrophoresis: Hb A – 85.3 %, Hb A2 – 2.4%, HbF – 2.2% (normal values – till 0.5%)Hb Lepore – 10.1%



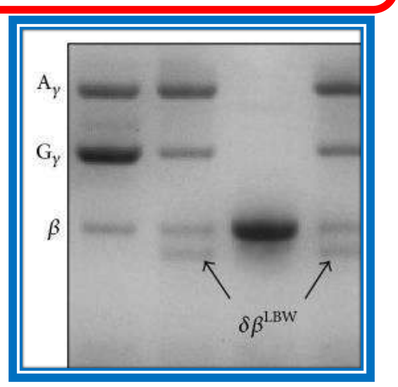


Fig. 3. Hemoglobin electrophoresis. Hb Lepore. (LBW – Lepore-Boston-Washington the most common Lepore hemoglobinopathy) [3]

Results: Clinical and paraclinical tests established moderate anemia, hepatomegaly and a minor hemolytic syndrome. Only the specific test - hemoglobin electrophoresis, highlighted the abnormal fraction - Lepore hemoglobin - 10.1%.

Conclusions: Anemic syndrome is common in children, but usually it is a real challenge when it comes to establish a certain diagnosis. In the Republic of Moldova, this case is unique in that it creates difficulties confirming the diagnosis.