C18. THE USE OF DNA TECHNIQUES FOR DIAGNOSIS IN A CASE OF HIPERSIDEREMIC ANEMIA.

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Is widely recognized that the diagnosis of anemia can be quite tricky and difficult in some cases. An accurate and reliable diagnositic is however crucial for treatment and monitoring of patients. In recent years, molecular biology and genetics of the various syndromes have been described in detail, revealing the wide range of mutations encountered in each type of inherited genetical disorders. That leaded to improve considerable the diagnosis of those syndromes.

Case report: male, 39 years old, diagnosed with Thalassemia minor in 1983 in our institute (HbF 11%) without any transfusion until now. After the diagnosis has not made any control until recently when he made a biliary colic which were admitted to gastroenterology service. Clinical and laboratory parameters as pallor, jaundice, hepato-splenomegaly, Hb 9 g / dL and ferritin 1700 ng / ml., raised the suspicion of hemochromatosis. The DNA tests for 11 of the most frequent mutation for hemochromatosis were negative.

Hematologic reevaluation raised suspicion of Thalassemia intermedia. A number of 22 of the most common β thalassemia mutations specific to the Mediterranean have been tested by molecular biology techniques. Testing showed double heterozygosis: -101 [C> T] and Codon 8[-AA]

In INHT, the principal tools in thalassemia diagnosis were, for many years, classical Hb electrophoresis combined with peripheric blood examination on microscope. Since 2010, the new method - molecular biology was introduced. In our opinion, the classical methods remain the basis for routine diagnosis, but molecular biology allow to clarify ambiguous and difficult cases.