DNA analys efter blod transfusion eller organ transplantation

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Fall 1

- Man, 48 år, har fått höga ferritinvärden efter benmärgstransplantation. Tacksam för HFE genotypning.
- Analys resultat: HFE 187 C/C; HFE 845 G/A
- Tolkning?
- Åtgärd?
Fall 2

- 2-årig flicka i invandrarfamilj har svår transfusionskrävande mikrocytär anemi.
- Thalassemi ? Hemoglobininopati ?
- Analys ?
- Tolkning ?
- Åtgärd ?
• Number and maturation of reticulocytes in various genotypes of thalassaemia as assessed by flow cytometry.
• Khuhapinant A, Bunyaratvej A, Tatsumi N, Pribwai M, Fucharoen S.
• Source
• Department of Pathobiology, Faculty of Science, Mahidol University, Bangkok, Thailand.
• Abstract
• Ineffective erythropoiesis is a prominent defect leading to anaemic status in thalassaemic patients. Reticulocyte enumeration in the peripheral blood is a non-aggressive method of measuring bone marrow erythropoietic activity. We used an automated reticulocyte counter (Sysmex R-3000) to determine the number and maturation level of circulating reticulocytes among various types of thalassaemia: non-splenectomized beta-thalassaemia/haemoglobin E (beta E) and splenectomized cases (beta E-S), classical haemoglobin H disease (H), haemoglobin H disease with haemoglobin Constant Spring (H/CS), homozygous haemoglobin Constant Spring (CS/CS), homozygous haemoglobin E (EE), heterozygous thalassaemics and other rare combinations. Haemoglobin H disease has a higher absolute count than beta-thalassaemia (beta E), indicating relatively better compensatory erythropoiesis in haemoglobin H disease. Those with CS genes (H/CS and CS/CS) have poorer reticulocyte maturation than any other type of thalassaemia with rather high absolute numbers, especially in H/CS. This indicates a severer degree of ineffective erythropoiesis in beta-thalassaemia (beta E), which reflects an insufficient rise in reticulocyte number in comparison with alpha-thalassaemia (H). The presence of haemoglobin Constant Spring is associated with abnormally low reticulocyte maturation due to enhanced erythrocyte production or direct effect of Constant Spring globin itself, both still unexplained with the current information. The splenectomized beta E has increased reticulocyte number and cells with high DNA content, probably nucleated red cells, designated as the upper particle count parameter. However, there is the same degree of reticulocyte maturation in non-splenectomized and splenectomized beta E patients, suggesting a role for splenic pooling of reticulocytes.
Provtagningsföreskrifter inför DNA test

• (Multi-)transfunderad patient
• Att begrunda:
  – Innehåller transfunderat blod leukocyter?
  – Retikulocyter?
  – Antal retikulocyter jfr patientens egna leukocyter?
  – Skillnader i analysstrategi?
Provtagningsföreskrifter inför DNA test

• (Multi-)transfunderad patient
• Benmärgstransplanterad patient
• Patient med solid organtransplantation