

Training course on Diagnosis and management of very rare anaemias: a challenge

February 3-4, 2012 - Paris, France

Chairs: P. Aguilar-Martinez, H. Heimpel, R. Van Wijk, L. Ribeiro, A. Zanella

Programme

Friday 3rd February 2012

- 08:30-09:00: **What are very rare anaemias ?** B. Gulbis (Brussels)
Epidemiology, Prevalence and incidence in the European countries
Patterns of heredity, Environmental factors and the influence of consanguinity
- Session 1: DISORDERS OF RED CELL PRODUCTION** Chairs: C. Camaschella - H. Heimpel
- 09:00-09:30: **Congenital dyserythropoietic anaemias (CDA)** H. Heimpel (Ulm)
Classification, Genetics, Pathophysiology, including iron overload
Clinical presentation, Laboratory investigations including rational
steps of diagnosis, Therapy of CDA I and CDA II, Lifelong management
and treatment of iron overload
- 09:30-10:00: **Diamond Blackfan Anaemia** I. Dianzani (Novara)
Pathophysiology, Genetics, Clinical information, Laboratory investigations
including rational steps of diagnosis, Pharmacotherapy, Risk of leukaemia
and life long follow up
- 10:00-10:30: **Aplastic anaemia** H. Schrezenmeier (Ulm)
Pathophysiology, Clinical information, Laboratory investigations including
bone marrow pathology, AA and PNH, Differential diagnosis AA and MDS,
Genetics, Therapy including transfusion guidelines
- 10h30-11h00** *Coffee break*
- 11:00-11:30: **Rare inherited anaemias due to abnormal iron absorption/recycling** C. Camaschella (Milan)
Classification, Genetic background, Clinical presentation and management
- 11:30-12:00: **Rare inherited anaemias due to defects in the erythroblast iron acquisition pathway** C. Beaumont (Paris)
Molecular basis, Clinical diagnosis, Management
- 12:00-12:30: **Hereditary sideroblastic anaemias** A. May (Cardiff)
Pathophysiology, genetics, clinical information, Laboratory investigations
including rational step of diagnosis
- 12:30-13:00: **Clinical cases** (brought by the participants and/or the speakers) C. Camaschella (Milan)
or clinical cases with voting boxes & H. Heimpel (Ulm)
- 13:00-14:30:** *Lunch*

Session 2.1: DISORDERS OF RED CELL SURVIVAL - HEREDITARY HAEMOLYTIC ANAEMIAS

➤ DISORDERS OF THE RED CELL MEMBRANE - Chair: J. Delaunay

- 14:30-15:00: **Hereditary spherocytosis** A. Zanella (Milan)
Pathophysiology, Molecular and clinical aspects, Laboratory investigation
- 15:00-15:30: **Hereditary elliptocytosis, pyropoikilocytosis and South-East Asian Ovalocytosis** L. Ribeiro (Coimbra)
Pathophysiology, Molecular and clinical aspects, Laboratory investigation
- 15:30-16:00: **Clinical cases of membrane disorders (with voting boxes)** L. Ribeiro (Coimbra) & A. Zanella (Milan)
- 16:00-16:30: Coffee break**
- 16:30-17:00: **Hereditary stomatocytosis and other very rare membrane disorders** A. Iolascon (Naples)
Pathophysiology, Molecular and clinical aspects, Laboratory investigation

➤ VERY RARE DISORDERS OF HEME SYNTHESIS - Chair: S.L. Thein

- 17:00-17:30: **Erythropoietic porphyrias** J.C. Deybach (Paris)
Pathophysiology, Molecular and clinical aspects, Laboratory investigation
- 17:30-18:00: **Closing talk of Day 1 - ENERCA 3** J.L. Vives-Corróns (Barcelona)
*Results in the domain of very rare anaemias : national networks and registries
Expert centres and supranational provision of diagnostic methods in Europe*

Saturday 4th February 2012

Session 2.2: DISORDERS OF RED CELL SURVIVAL - HEREDITARY HAEMOLYTIC ANAEMIAS

➤ DISORDERS OF RED CELL METABOLISM - Chairs: E. Ristoff* - R. Van Wijk

- 08:30-09:00: **Normal and defective generation of energy in red blood cells abnormalities of glycolysis and pyrimidine metabolism** R. Van Wijk (Utrecht)
Pathophysiology, molecular and clinical aspects, Laboratory investigation
- 09:00-09:30: **Defective anti-oxidant defence** E. Ristoff (Stockholm)*
(abnormalities of HMP-shunt and glutathione metabolism)
Pathophysiology, molecular and clinical aspects, Laboratory investigation
- 09:30-10:00: **Clinical or biological cases with voting boxes** R. Van Wijk (Utrecht) & P. Aguilar-Martinez (Montpellier)
(brought by the participants and/or the speakers)
- 10:00-10:30: Coffee break**

Session 3: DISORDERS OF RED CELL SURVIVAL - ACQUIRED HAEMOLYTIC ANAEMIAS

Chairs: H. Schrezenmeier - G. Socié*

- 10:30-11:00: **Autoimmune haemolytic anaemias by warm antibodies** N. Ahrens (Regensburg)
*Red cell antigens and antibodies, Methods to detect cell bound and free anti red cell antibodies
Anti-bodies, specificity, Warm/cold agglutinins, Pathophysiology
Classification according to underlying condition, Antibody type, Rh specificity
Clinical presentation, Laboratory investigation including rational steps of diagnosis
The problem of Coombs negative AIHA*
- 11:00-11:30: **Transient & chronic cold agglutinin disease** S. Berentsen (Haugesund)
*Pathophysiology, Classification according to underlying condition
Clinical presentation, Laboratory investigation including including rational steps of diagnosis, Management therapy*

11:30-12:00: **Microangiopathic haemolytic anaemias** P. Coppo (Paris)
TTP, HUS (haemolytic uraemic syndrome)
Pathophysiology, genetics, Clinical presentation, Laboratory investigation including rational steps of diagnosis, Paraneoplastic microangiopathies and mechanical of red cell fragmentation

12:00-12:30: **Paroxysmal Nocturnal Haemoglobinuria** G. Socié (Paris)*
Clinical picture, underlying mechanisms, Laboratory diagnosis
Therapeutic management: transfusions, Eculizumab, stem cell transplantation
PNH patient registry

12:30-14:00: Lunch

Session 4: DIAGNOSIS AND TREATMENT OF VERY RARE ANAEMIAS Chair: D. Loukopoulos

14:00-14:30: **Quality controls for the diagnosis of rare red cell disorders** A. Mosca (Milan)

14:30-15:00: **Stem cell transplant in the context of very rare anaemias** F. Locatelli (Rome)

15:00-15:30: Coffee Break

INTERACTIVE DISCUSSIONS IN TWO GROUPS

15:30-17:00: Groupe A - Laboratory aspects - **Chairs: P. Aguilar-Martinez, R. Van Wijk**
Very rare anaemias: diagnostic strategy and tools
- Flowcharts for the biological diagnosis of very rare anaemias (15min + discussion 5 min) A. Zanella (Milan)
- Red cell morphology in very rare anaemias (15min + discussion 5 min) O. Fenneteau (Paris) & E. Lainey (Paris)
- What is the added value of molecular genetics in the stepwise diagnostic process ? (15 min + discussion 5 min) A. Iolascon (Naples)
- Presentation of 3 clinical cases (brought by the participants and/or the speakers) and interactive discussions (5 min presentation + 5 min discussion for each case)

15:30-17:00: Groupe B - Clinical - **Chairs: H. Heimpel, I. Dianzani**
Treatment of very rare anaemias
- Splenectomy: Why, when and how ? (15min + discussion 5 min) H. Heimpel (Ulm)
- Iron overload and chelation therapy (15min + discussion 5 min) I. Dianzani (Novara)
- Role of EPO in the management of rare anaemias (15min + discussion 5 min) N. Casadevall (Paris)
- Presentation of 3 clinical cases (brought by the participants and/or the speakers) and interactive discussions (5 min presentation + 5 min discussion for each case)

KEY NOTE LECTURES Chair: Y. Beuzard

17:00-17:25: **Cord blood transplant and very rare anaemias** TBA *

17:25-17:30: **Diagnosis of very rare anaemias : a challenge !** J.L. Vives-Corróns (Barcelona)

17:30: **End of the course**

* speakers to be confirmed