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 ?** *Epidemiology, Prevalence and incidence in the European countries* *Patterns of heredity, Environmental factors and the influence of consanguinity*	B. Gulbis (Brussels)

Session 1: DISORDERS OF RED CELL PRODUCTION **Chairs: C. Camaschella - H. Heimpel**

09:00-09:30:	**Congenital dyserythropoietic anaemias (CDA)** *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*	H. Heimpel (Ulm)
09:30-10:00:	**Diamond Blackfan Anaemia** *Pathophysiology, Genetics, Clinical information, Laboratory investigations including rational steps of diagnosis, Pharmacotherapy, Risk of leukaemia and life long follow up*	I. Dianzani (Novara)
10:00-10:30:	**Aplastic anaemia** *Pathophysiology, Clinical information, Laboratory investigations including bone marrow pathology, AA and PNH, Differential diagnosis AA and MDS, Genetics, Therapy including transfusion guidelines*	H. Schrezenmeier (Ulm)
10h30-11h00	***Coffee break***	
11:00-11:30:	**Rare inherited anaemias due to abnormal iron absorption/recycling** *Classification, Genetic background, Clinical presentation and management*	C. Camaschella (Milan)
11:30-12:00:	**Rare inherited anaemias due to defects in the erythroblast iron acquisition pathway** *Molecular basis, Clinical diagnosis, Management*	C. Beaumont (Paris)
12:00-12:30:	**Hereditary sideroblastic anaemias** *Pathophysiology, genetics, clinical information, Laboratory investigations including rational step of diagnosis*	A. May (Cardiff)
12:30-13:00:	**Clinical cases** (brought by the participants and/or the speakers) or clinical cases with voting boxes	C. Camaschella (Milan) & 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** *Pathophysiology, Molecular and clinical aspects*, *Laboratory investigation*	A. Zanella (Milan)
15:00-15:30:	**Hereditary elliptocytosis, pyropoikilocytosis and South-East Asian Ovalocytosis** *Pathophysiology, Molecular and clinical aspects, Laboratory investigation*	L. Ribeiro (Coimbra)
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** *Pathophysiology, Molecular and clinical aspects, Laboratory investigation*	A. Iolascon (Naples)

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

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

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** *Pathophysiology, molecular and clinical aspects*, *Laboratory investigation*	R. Van Wijk (Utrecht)
09:00-09:30:	**Defective anti-oxidant defence** (abnormalities of HMP-shunt and glutathione metabolism) *Pathophysiology, molecular and clinical aspects*, *Laboratory investigation*	E. Ristoff (Stockholm)*
09:30-10:00:	**Clinical or biological cases with boting boxes** (brought by the participants and/or the speakers)	R. Van Wijk (Utrecht) & P. Aguilar-Martinez (Montpellier)
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** *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*	N. Ahrens (Regensburg)
11:00-11:30:	**Transient & chronic cold agglutinin disease** *Pathophysiology, Classification according to underlying condition* *Clinical presentation, Laboratory investigation including including rational steps of diagnosis, Management therapy*	S. Berentsen (Haugesund)

11:30-12:00:	**Microangiopathic haemolytic anaemias** **TTP, HUS (haemolytic uraemic syndrome)** *Pathophysiology, genetics, Clinical presentation*, *Laboratory investigation including rational steps of diagnosis***,** *Paraneoplastic microangiopathies and mechanical of red cell fragmentation*	P. Coppo (Paris)
12:00-12:30:	**Paroxysmal Nocturnal Haemoglobinuria** *Clinical picture, underlying mechanisms*, *Laboratory diagnosis* *Therapeutic management: transfusions, Eculizumab, stem cell transplantation* *PNH patient registry*	G. Socié (Paris)*
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-Corrons (Barcelona)
17:30:	**End of the course**	

*** speakers to be confirmed**