TRAINING COURSE ON
DIAGNOSIS AND MANAGEMENT OF
VERY RARE RED CELL AND IRON DISORDERS

Lisbon, Portugal
January 29 - 30, 2016

Chairs: Patricia Aguilar-Martinez, Paola Bianchi, Achille Iolascon,
Richard Van Wijk, Alberto Zanella

Friday, January 29, 2016

08:30–09:00  Introduction to the course:
             An overview on the epidemiology of very rare anaemias
             Data from the ENERCA project – The role of registries
             Michael Angastiniotis (Nicosia)

SESSION I – Disorders of Red Cell Production
Chair: Achille Iolascon (Naples)

09:00-09:25  Congenital dyserythropoietic anaemias (CDA)
             Achille Iolascon (Naples)
09:25-09:30  Discussion

09:30-09:55  Diamond blackfan anaemia
             Lydie Da Costa (Paris)
09:55-10:00  Discussion

10:00-10:25  Aplastic anaemia
             Hubert Schrezenmeier (Ulm)*
10:25-10:30  Discussion

10:30-11:00  Coffee Break

11:00-11:25  Very rare anaemias related to abnormal iron metabolism
             Laura Silvestri (Milan)
11:25-11:30  Discussion

11:30-11:55  Hereditary microcytic anaemia
             Molecular basis, clinical presentation and management.
             Achille Iolascon (Naples)
11:55-12:00  Discussion

12:00-12:25  Hereditary sideroblastic anaemias
             Pathophysiology, genetics, clinical, information,
             Laboratory investigations including rational step of diagnosis.
             Josef Prechal (Salt Lake City)*
12:25-12:30  Discussion

12:30-13:00  Clinical cases with voting boxes
             Achille Iolascon (Naples)
             Patricia Aguilar Martinez (Montpellier)

13:00-14:30  Lunch
SESSION II – Disorders of Red Cell Survival – Hereditary Haemolytic Anaemias
SESSION II.I – Disorders of the Red Cell Membrane
Chair: Alberto Zanella (Milan)

14:30-14:55 Disorders of RBC cytoskeleton: hereditary spherocytosis Paola Bianchi (Milan)
14:55-15:00 Discussion

15:00-15:25 Hereditary elliptocytosis, pyropoikilocytosis and South-east Asian ovalocytosis Leticia Ribeiro (Coimbra)
15:25-15:30 Discussion

15:30-16:00 Clinical cases with voting boxes – (Membrane disorders) Patricia Aguilar Martinez (Montpellier)
Achille Iolascon (Naples)

16:00-16:30 Coffee break

16:30-16:55 Hereditary stomatocytosis Loïc Garçon (Amiens)
16:55-17:00 Discussion

SESSION II.II – Very Rare Disorders of Heme Synthesis
Chair: Béatrice Gulbis (Brussels)

17:00-17:25 Erythropoietic porphyrias Maria D. Cappellini (Milan)
17:25-17:30 Discussion

17:30-18:30 INTERACTIVE DISCUSSIONS IN TWO GROUPS

Group A – Laboratory aspects
Chairs: Paola Bianchi (Milan), Loïc Garçon (Amiens)
Very rare anaemias: diagnostic strategy and tools
Presentation of 3-4 bioclinical cases (brought by the participants and/or the speakers)
and interactive discussions (5 min presentation +10 min discussion for each case)

Group B – Clinical aspects
Chairs: Alberto Zanella (Milan), Achille Iolascon (Naples)
Very rare anaemias: clinical cases
Presentation of 3-4 clinical cases (brought by the participants and/or the speakers)
and interactive discussions (5 min presentation +10 min discussion for each case)

Saturday, January 30, 2016

SESSION II.III – Disorders of Red Cell Metabolism
Chair: Paola Bianchi (Milan)

08:30-09:25 Red blood cell enzymes abnormalities Richard Van Wijk (Utrecht)
09:25-09:30 Discussion

09:30-10:00 Clinical or biological cases with voting boxes Paola Bianchi (Milan)
Richard Van Wijk (Utrecht)

10:00-10:30 Coffee break
SESSION III – Disorders of Red Cell Survival – Acquired Haemolytic Anaemias
Chair: Hubert Schrezenmeier (Ulm)*

10:30-10:55  Autoimmune haemolytic anaemias by warm antibodies  Wilma Barcellini (Milan)
10:55-11:00  Discussion

11:00-11:25  Transient & chronic cold agglutinin disease  Sigbjorn Berentsen (Haugesund)
11:25-11:30  Discussion

11:30-11:55  Microangiopathic haemolytic anaemias TTP, HUS (haemolytic uraemic syndrome)  Flora Peyvandi (Milan)*
11:55-12:00  Discussion

12:00-12:25  Paroxysmal nocturnal haemoglobinuria  Régis Peffault de Latour (Paris)*
12:25-12:30  Discussion

12:30-13:00  Clinical or biological cases with voting boxes  Wilma Barcellini (Milan)
Régis Peffault de Latour (Paris)*

13:00-14:00  Lunch

SESSION IV – Diagnosis and Treatment of Very Rares Anaemias: the example of PK deficiency
Chair: Joan Lluis Vives Corrons (Barcelona)

14:00-14:25  Gene therapy for PKD and other rare disorders  Jose Carlos Segovia (Madrid)
14:25-14:30  Discussion

14:30-14:55  New treatments of PKD  TBA*
14:55-15:00  Discussion

15:00-16:30  INTERACTIVE DISCUSSIONS IN TWO GROUPS:

Group A – Laboratory aspects
Chairs: Patricia Aguilar Martinez (Montpellier), Richard Van Wijk (Utrecht)

(Very) rare anaemias: diagnostic strategy and tools
- The ENERCA telemedicine platform  Béatrice Gulbis (Brussels)
  (10 min +5 min discussion)
- Red cell morphology in very rare anaemias  (15 min+ discussion 5 min)  Lydie Da Costa (Paris)
- Molecular genetics and NGS in the diagnostic process  Richard Van Wijk (Utrecht)
  (10 min + discussion 5 min)

- 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)

Group B – Clinical aspects
Chairs: Maria D. Cappellini (Milan), Achille Iolascon (Naples)

Treatment of (very) rare anaemias
- Splenectomy: why, when and how?  (10 min+ discussion 5 min)  Achille Iolascon (Naples)
- Iron overload and chelation therapy  (10 min+ discussion 5 min)  Maria D. Cappellini (Milan)
- The place of other treatments in the management of rare anaemias  (Blood transfusion, Supportive therapy).  (10 min+ discussion 5 min)  Alberto Zanella (Milan)
- Presentation of 3 clinical cases (brought by the participants and/or the speakers) and interactive discussions (5 min presentation + 10 min discussion for each case)

16:30-16:55  **KEYNOTE LECTURE:**  
New treatments of dyserythropoiesis  
Jean Benoit Arlet (Paris)

16:55-17:00  **Discussion**

17:00-17:05  Diagnosis of (very) rare anaemias: closing remarks  
Joan Lluis Vives Corrons (Barcelona)

**MEETING CLOSURE**

*Pending*