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* Scientific Program

**Scientific Program**

Please click on the day to see the full scientific program of that day.

Thursday, February 2

14:00 - 14:15 Opening of conference

14:15 - 15:45 SCIENTIFIC SESSION

**Update on erythropoiesis**

Chair: C Camaschella (Italy)

* Erythropoiesis control  
  S Rivella (USA)
* Congenital dyserythropoietic defects  
  A Iolascon (Italy)
* Erythropoietic transcription factor defects and consequences  
  D Higgs (United Kingdom)

15:45 - 16:00 Coffee break

16:10 - 17:30 CLINICAL CASE SESSION

**Case presentations by delegates**

Moderators: A Iolascon (Italy), MD Cappellini (Italy)

* Diagnosis of anemias by tomographic digital holography  
  P Ferraro
* Atypical hereditary spherocytosis associated with mitochondrial cytopathy diagnosed by whole-exome sequencing  
  L Couronne
* Mutating heme oxygenase-1 into a peroxidase causes a defect in bilirubin synthesis associated with microcytic anemia and severe hyperinflammation  
  J Greil
* Adult onset isolated severe sideroblastic anemia attributed to an amino-acylation defect  
  D Swinkels
* Hemosiderosis, severe anemia and erythrocyte morphology aberrations explained by dna analysis  
  A van Gammeren
* Whole genome sequencing (WGS) for rare inherited anaemias- filling in the gaps  
  N Roy
* XPO1 regulates erythroid differentiation by exporting HSP70, and is a potential target for the treatment of B-thalassemia  
  F Guillem

17:00 - 18:30 SCIENTIFIC SESSION

**Failure of Erythropoiesis**

Chair: S Rivella (USA)

* Blackfan Diamond anemia  
  L Da Costa (France)
* Acquired failures of erythropoiesis  
  C Dufour (Italy)

18:30 - 19:00 POSTER PITCHES

**Short case presentations by delegates**

Moderator: A Iolascon (Italy)

* Iron refractory iron deficiency anemia (IRIDA): A heterogeneous disease that is not always iron refractory   
  A Donker
* When something bad runs in the family: The power of modern molecular techniques in explaining  a very rare clinical syndrome  
  A Ermens
* Diagnosis and molecular characterization of a novel alpha(0)-thalassemia deletion found in a Greek child with unexplained microcytic hypochromic anemia  
  A Makis
* Fyn plays a novel key role in erythropoiesis as oxidative sensor  
  E Beneduce
* Pancytopenia in a fifteen years old girl previously diagnosed with immune thrombocytopenia  
  V Campuzano
* Acute hepatic crisis in a 4-year-old girl with sickle cell anemia: Favourable outcome after exchange transfusion  
  H Khalifeh

Diagnosis of anemias by tomographic digital holography

P Ferraro

Atypical hereditary spherocytosis associated with mitochondrial cytopathy diagnosed by whole-exome sequencing

L Couronne

Mutating heme oxygenase-1 into a peroxidase causes a defect in bilirubin synthesis associated with microcytic anemia and severe hyperinflammation

J Greil

Adult onset isolated severe sideroblastic anemia attributed to an amino-acylation defect

D Swinkels

Hemosiderosis, severe anemia and erythrocyte morphology aberrations explained by dna analysis

A van Gammeren

Whole genome sequencing (WGS) for rare inherited anaemias- filling in the gaps

N Roy

XPO1 regulates erythroid differentiation by exporting HSP70, and is a potential target for the treatment of B-thalassemia

F Guillem

19:00 - 20:00 Welcome reception

Friday, February 3

09:00 - 10:30 SCIENTIFIC SESSION

**Next generation sequencing**

Chair: M Muckenthaler (Germany)

* General approach to NGS  
  V Benes (Germany)
* Use of NGS in the diagnosis of anemia  
  I Roberts (United Kingdom)
* Clinical cases solved by NGS  
  M Sanchez (Spain)

10:30 - 11:00 Coffee break

11:00 - 12:00 SCIENTIFIC SESSION

**Proteomics of anemias**

Chair: L De Franceschi (Italy)

* General approach to Proteomics of erythropoiesis  
  G Bosman (the Netherlands)
* Proteomics of red cells and applications  
  AM Toye (United Kingdom)

12:00 - 13:00 ROUND TABLE SESSION

**Round table: Rare anemias and European Network for rare disease from patient’s perspective**

Chair: JL Vives-Corrons (Spain)

* Point of view of ERN   
  JL Vives-Corrons (Spain)
* Point of view of patients association   
  E Androulla (Cyprus)
* Point of view of a HCP   
  MD Cappellini (Italy)

13:00 - 14:30 Lunch

14:30 - 16:00 INTERACTIVE SESSION

**Interactive case presentations by faculty (I)**

Moderators: A Iolascon (Italy), MD Cappellini (Italy)

* Interactive clinical cases of red cell membrane defects  
  P Aguilar-Martinez (France)
* Interactive clinical cases of immunoemolytic anemia  
  W Barcellini (Italy)
* Interactive clinical cases of stomatocytoses  
  I Andolfo (Italy)

16:00 - 16:30 Coffee break

16:30 - 18:00 INTERACTIVE SESSION

**Interactive case presentations by faculty (II)**

Moderators: P Aguilar-Martinez (France), JL Vives-Corrons (Spain)

* Clinical cases of enzymatic defects  
  R van Wijck (the Netherlands)
* Clinical cases of Thal interacting with other defects  
  A Taher (Lebanon)
* Clinical Cases of CDA-s  
  R Russo (Italy)

Saturday, February 4

09:00 - 10:30 SCIENTIFIC SESSION

**Iron and Erythropoiesis**

Chair: P Aguilar-Martinez (France)

* General concept on Iron and Erythropoiesis  
  M Muckenthaler (Germany)
* Iron overload in Thalassemias and rare anemias  
  A Taher (Lebanon)
* Iron restricted erythropoiesis  
  C Camaschella (Italy)

10:30 - 11:00 Coffee break

11:00 - 13:00 SCIENTIFIC SESSION

**New therapies for iron overload and deficiency**

Chair: S Rivella (USA)

* New drugs in Thalassemias and rare anemias  
  MD Cappellini (Italy)
* New drugs for iron deficiency  
  D Girelli (Italy)
* New drugs for enzymatic defects  
  M Layton (United Kingdom)
* Modulation of DNA expression  
  L De Franceschi (Italy)

13:00 - 13:15 Concluding remarks

13:15 - 14:45 Lunch