

Inborn errors of metabolism in neonatology: clues for successful diagnosis and treatment

Santiago de Compostela, Spain, 13-15 June 2019

Course description

Early diagnosis and management of newborns and infants with inherited metabolic disorders is essential for the affected children's outcome. However, the quick and correct recognition of specific inherited metabolic disease in neonates is a challenge for neonatologists. It may be difficult, especially in critically ill neonates, to distinguish between the primary genetically encoded metabolic disorder and the secondary metabolic disturbances resulting in similar clinical picture.

This practical course is run by an experienced team of paediatricians, neonatologists, molecular biologists and biochemists specialised in metabolic medicine, who already contributed substantially to the understanding of metabolic disorders in childhood. The two days course includes lectures and seminars for 35 participants. The course is aimed at paediatricians with about 2-3 years clinical experience in the neonatology field.

Candidates are expected to register with an abstract reflecting a case with solved or unsolved diagnostic or therapeutic dilemmas related to the programme.

Learning objectives

- Appreciation of clinical situations leading to the suspicion of an inborn error of metabolism
- Awareness of emergency investigations and the use of specialised laboratory services
- Awareness of metabolic causes of neonatal encephalopathy, cardiomyopathy, liver disease, hypoglycaemia, lactic acidosis and dysmorphic syndromes
- Understand treatment and management options of critically ill newborns with inborn errors
- Understanding and optimal diagnostic use of neonatal screening and next generation sequencing
- Practical application of theory - participants are strongly encouraged to submit a case study from their own experience

Scientific organising committee

- Prof. Ivo Barić (Zagreb, Croatia)
- Prof. Maria-Luz Couce (Santiago de Compostela, Spain)

Target audience and participant profile

The course is restricted to about 35 participants and designed primarily for neonatologists with about 2-3 years clinical experience in the neonatology field, but also paediatricians of other profiles and paediatric residents who may benefit from the course.

PROGRAMME

Thursday 13th June:

13.30 **Registration and lunch**

14.10 **Welcome**

Maria-Luz Couce, Santiago de Compostela & Ivo Barić, Zagreb

14.20 **Pre-course quiz**

14.40 **Inborn errors of metabolism in the neonatal period: presentation patterns and importance of early diagnosis and intervention**

Domingo González-Lamuño, Santander, Spain

15.10 Discussion

15.20 **Dysmorphic baby: think of inborn errors of metabolism**

Stephanie Grünewald, London

15.50 Discussion

16.00 Coffee break

16.30 **Pregnancy and inherited metabolic disorders**

Elaine Murphy, London

17.00 Discussion

17.10 **Emergencies in neonatology due to inborn errors of metabolism**

John Walter, Manchester

17.40 Discussion

17.50 **Hyperammonaemia in newborns**

Johannes Häberle, Zurich

18.20 Discussion and **case report 1**

Paula Sánchez- Pintos, Santiago de Compostela

19.30 **Dinner**

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Friday 14th June

08.30 **Newborn screening for metabolic diseases. Present and future.**

Giancarlo La Marca, Florence

09.00 Discussion

09.10 **Next-generation sequencing in the neonatal diagnosis of inborn errors of metabolism**

Holger Prokisch, Munich

09.40 Discussion

9.50 Coffee break

10.20 **The floppy newborn: could it be a metabolic disease?**

Barbara Plecko, Graz

10.50 Discussion and **case report 2**

11.10 **Movement disorders in neonates**

Angeles García-Cazorla, Barcelona

11.40 Discussion and **case report 3**

12.00 **Metabolic approach to cardiomyopathy in neonates**

Manuel Schiff, Paris

12.30 Discussion and **case report 4**

12.50 Lunch

14.00 **Approach to lactic acidosis in the neonatal period**

Angeles García-Cazorla, Barcelona

14.30 Discussion and **case report 5**

María José de Castro, Santiago de Compostela

14.50 Coffee break

15.20 **Gallery of illustrative images and video clips**

Contributions from all lecturers and participants

15.50 **Meet the clinical expert** - parallel sessions

Carlo Dionisi-Vici, Rome; John Walter, Manchester; Stephanie Grünwald, London

17.30 **Guided tour through the old city & dinner**

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Saturday 15th June

08:45 **Neonatal epileptic encephalopathy**

Barbara Plecko, Graz

09.15 Discussion and **case report 6**

09.35 **How to recognize and treat inborn errors of metabolism presenting with neonatal liver disease**

Carlo Dionisi-Vici, Rome

10.05 Discussion and **case report 7**

10.25 Coffee break

10.55 **Neonatal hypoglycemia and inborn errors of metabolism**

René Santer, Hamburg

11.35 Discussion and **case report 8**

11.55 **Ethics in neonatology and inborn errors of metabolism**

Participation of all participants and lecturers

Lead: Peta Coulson-Smith, Southampton

12.40 **Experiences and expectations from associations of families of patients with metabolic diseases**

Monica Da Silva, Santiago de Compostela & Aitor Calero, Madrid

13.10 **Discussion**

13.20 **Post-course quiz & final discussion**

Maria-Luz Couce, Santiago de Compostela & Ivo Barić, Zagreb

13.50 **Lunch and departure**