

Transition from childhood to adulthood in inherited metabolic diseases
SSIEM-MetabERN collaboration
First webinar

On the European commission webinar platform
25th April 2019

PROGRAMME

14h30 Nadia Belmatoug, France

Introduction

14h40 Sarah-Jayne Blakemore, UK

Development of the mature brain during adolescence and young adulthood

15h Allyson Terry, UK

Case 1: PKU

- Pregnancy planning, teenage, unplanned pregnancy
- Moving out of home, university, etc.

15h20 Nadia Belmatoug, France

Case 2: Gaucher disease, long-term care, monitoring with ERT

15h40 Andrew Morris, UK

Case 3: Patient with learning difficulties, neurodegeneration

- Capacity
- Guardianship
- End of life care

16h Elaine Murphy, UK

Case 4: Patient at risk of acute decompensation

- Specific triggers – alcohol, drugs, peri-menstrual, music festivals
- Disordered eating
- Emergency guidelines and hospital admissions

16h20 Patient interview

A female patient with symptomatic OTC deficiency, now a young adult: perimenstrual decompensation, adherence, travel, work, relationships, etc.

16h40 Elaine Murphy, UK

Conclusion, Questions/Answers