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SOCIEDADE PORTUGUESA  
DE DOENÇAS METABÓLICAS

**Reference Center for the Treatment of Inborn Errors of Metabolism,  
Unidade Local de Saúde Santo António (ULSSA), Porto**

**&**

**Portuguese Society of Inherited Metabolic Diseases (SPDM)**

**Disorders of B vitamins metabolism and transport | Post-graduate education  
Porto, 21 February 2025**



**Audience:** Clinicians, Clinical Laboratory Geneticists, Pharmacists, Nutritionists, Nurses (30 participants)

**When:** 21st February 2025

**Where:** Hospital de Santo António, Auditório Alexandre  
Moreira, Training rooms

**Registrations available at** [Formação Disorders of B vitamins metabolism and transport/](#)  
Lunch is included and a participation diploma will be distributed



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**09:45 | Registration with Welcome Coffee**

**10:00 | Opening**

Arlindo Guimas, Course coordinator, ULS Santo António, Porto, PT

**10:05 | 13:00**

**From clinics to genetics, knowledge update**

Chairs: Dulce Quelhas, Anabela Bandeira, ULS Santo António, Porto, PT

10:05 Disorders of Cobalamin from neuropathophysiology to clinical manifestations

*David Coelho, Nancy, FR*

10:50 Disorders of Pyridoxine and Riboflavin

*Andrew Morris, Manchester, UK*

11:35 Thiamine: from pathophysiology to clinical manifestations

*Margarida Coelho, ULSSA, Porto, PT*

11:55 Biotin: from pathophysiology to clinical manifestations

*Joana Correia, ULSSA, Porto, PT*

12:15 Other B vitamins: folate, pantothenic acid, nicotinamide

*Andrew Morris, Manchester, UK*

**13:00 | 14:00 | Lunch**

**14:30 | 16:20**

**Clinical Workshop**

**Diagnostic Challenges**

Chairs: Andrew Morris and David Coelho

14:05 Case 1 – Thiamine Deficiency: A Case Report

*Rui Diogo - CRDHM ULS Coimbra, Coimbra, PT*

14:25 Case 2 – Riboflavin: Family challenge

*Joana Tenente- CRDHM ULS São João, Porto, PT*

*Rute Martins - CRDHM ULS Santo António, Porto, PT*

14:45 Case 3 – Uncovering Biotin: A Case Report Analysis

*Inês Aires Martins - CRDHM ULS Santo António, Porto, PT*

15:05 Case 4 – B vitamin deficiency - case reports

*Patrícia Lipari Pinto - CRDHM ULS Santa Maria, Lisboa, PT*

15:25 Case 5 – Exploring CBL F/G: A Comprehensive Case Report

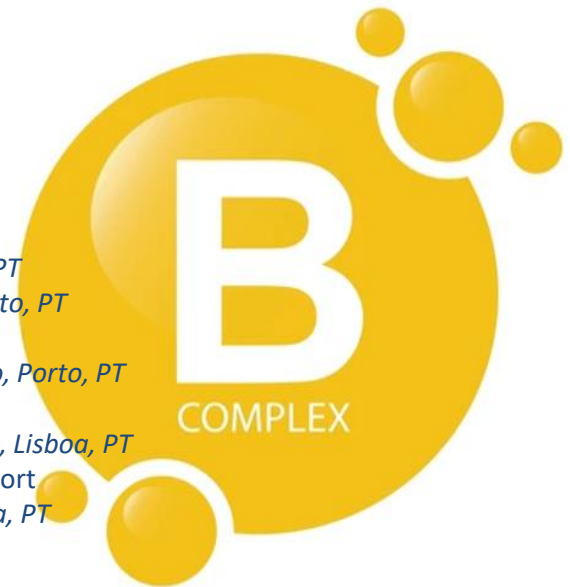
*Gonçalo Padeira - CRDHM ULS São José, Lisboa, PT*

15:45 Discussion

**16:20 | Take home messages**

Sara Rocha, ULSSA

**16:35 | Closing Remarks and Farewell Coffee**



For more information contact:

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Inborn Errors of Metabolism Unit of your Hospital

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