



# Workshop on Classical Homocystinuria (CH)

- **Audience:** all medical specialties (30 participants)
- **When:** 23<sup>rd</sup> February 2024
- **Where:** Lux Lisboa Park Hotel
- **Registrations available at** <https://www.spdm.org.pt/formacoes/em-agenda/>
- Lunch is included and a participation diploma will be distributed

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**Reference Center for the Treatment of Inborn Errors of Metabolism,  
Lisbon, Portugal**



**Sponsored by:**



**Program      Lisboa, 23<sup>rd</sup> February 2023**

Hour	Subject	Speaker
10.00–10:30	Registration and Coffee	
10:30-10:40	Welcome and Introduction Coordinator of IEM Reference Center at CHULN	Ana Gaspar – CHULN, Lisboa, PT
10:40-11:05	How biochemistry impacts the diagnosis and treatment options.	Rita Castro – FFUL, Lisboa, PT and Pennsylvania State University, USA
11:05-11:25	The clinical picture from newborn to adult life.	Patrícia Janeiro – CHULN, Lisboa, PT
11:25-11:45	Cerebrovascular manifestations in young adults	Leonor Guedes – CHULN, Lisboa, PT
11:45-12:15	Therapeutic approaches: which are the options?	Viktor Kozich – Charles University, Prague, CZ
12:15- 12:30	Discussion	
12:30-13:45	Lunch	
14:00-14:15	Nutritional management: a continuous challenge for diet compliance	Rita Loureiro – CHULN, Lisboa, PT
14:15-15:30	<b>Round Table</b>  Introductory lecture Patients management along life  Portuguese Reference Centers experience:  Treatment interventions vs clinical and biochemical outcome: from early onset to lifelong. Complications of Cystathionine $\beta$ -synthetase (CBS) deficiency: how to face them. The usefulness of the guideline in patients 'management: does the daily practice dictate adjustments?. Questions and Answers (QA). Wrap up and conclusions.	Viktor Kozich – Charles University, Prague, CZ   Moderators: Daniel Gomes, Patrícia Janeiro, SPDM
	CHULN, Hospital de Santa Maria, Lisboa, PT	Ana Gaspar and Anabela Oliveira
	CHULC, Hospital Dona Estefânia, Lisboa, PT	Ana Cristina Ferreira
	CHUSJ, Hospital de São João, Porto, PT	Esmeralda Rodrigues and M. Teresa Cardoso
	CHUP, Hospital Santo António, Porto, PT	Esmeralda Martins and Arlindo Guimas
	CHUC, Hospital Pediátrico de Coimbra, PT	Luisa Diogo and Sónia Moreira



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