



2ND SYMPOSIUM 2º SIMPÓSIO

RECENT ADVANCES & RARE OVERVIEW

THE YEAR IN REVIEW

22/23

O ANO EM REVISTA

22/23

25, 26TH FEBRUARY 2023

Hotel Marina Atlântico, Ponta Delgada

SÃO MIGUEL - AÇORES

HYBRID

PROGRAM



ORGANIZING COMMITTEE MESSAGE

The Rare Diseases Study Group (NEDR) of the Internal Medicine Portuguese Society (SPMI) will hold the 2nd RARO Symposium (Recent Advances & Rare Overview) – the Year in Review 2022/23, on February 25 and 26, 2023, in Ponta Delgada – São Miguel Island – Azores, with concomitant online transmission, using a hybrid format.

This symposium, which arose in a RARE pandemic time in 2021, aims to praise and disseminate the remarkable progress that medicine and science have made (and will continue to make) in the treatment of new, but also “old” diseases.

As has been our organizational practice, this scientific event, intends to provide all health care professionals, without distinction, with the most recent evidence in the area of rare diseases, with a focus on innovations and scientific news of the past year.

The format chosen for this year was the hybrid format. We will thus be present in person in the wonderful city of Ponta Delgada, starting what we intend to be a close partnership with colleagues from the Autonomous Regions and we will maintain the digital transmission, using the enormous projection and participation that sessions in this format have brought us.

We will have the usual presence of national and international speakers and attendees, also optimizing the use of new tools and digital platforms.

We therefor invite you to be present at this hybrid symposium, that is completely innovative in the area of Rare Diseases in Portugal, on the 25th and 26th of February.

On behalf of the 2nd RARO Symposium Organizing Committee, our best regards and RARO greetings. We hope to see you in Ponta Delgada (or online).



Luísa Pereira
On Behalf of the Organizing Committee

2ND SYMPOSIUM

2º SIMPÓSIO

SATURDAY – 25th FEBRUARY

08:45h Registration

09:00-09:30h **Welcome messages**

Luísa Pereira, Paula Macedo e Lèlita Santos 

09:30-10:30h **RARE OVERVIEW NEDR SESSION**

AZORES – STATE OF THE ART IN RARE DISEASES

Chairman: Patrícia Aranha *Divino Espírito Santo Hospital, Ponta Delgada*

Genetics and genomics of rare diseases: The Azores panorama

Luísa Mota Vieira *Divino Espírito Santo Hospital, Ponta Delgada*

Machado-Joseph disease/ Spinocerebellar ataxia type 3: The importance of the Azorean cohort for advances in understanding the disease

Manuela Lima *University of the Azores*

10:30-11:00h Coffee-break

11:00-12:00h **RARE OVERVIEW SPONSORED SYMPOSIUM**

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PENSAR PORFIRIA – SUSPEITE, TESTE, REFERENCIE

Chairman: Patrícia Aranha *Divino Espírito Santo Hospital, Ponta Delgada*

Suspecting Porphyria

Ana Ponciano  *Santo André Hospital, Leiria*

Testing for Porphyria

Filipa Ferreira *INSA Porto, Oporto*

Referring Porphyria

Ana Ponciano  *Santo André Hospital, Leiria*

12:00-13:00h

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RARE OVERVIEW SPONSORED SYMPOSIUM

CASTLEMAN DISEASE: CHALLENGES IN CLINICAL PRACTICE FOR A DIFFERENTIAL DIAGNOSE

Chairman: Luísa Pereira *CUF Tejo Hospital, Lisbon*

Challenging diagnosis in rare hematologic diseases: Between autoimmunity and autoinflammatory – Castleman Disease

Andrés González  *Ramón y Cajal University Hospital, Madrid*

About a clinical case: Patient with idiopathic Multicentric Castleman Disease, challenges in clinical practice

Marta Isabel Pereira *Coimbra University and Hospital Center, Coimbra*

13:00-14:30h

Lunch-break

14:30-15:30h

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RECENT ADVANCES AND RARE OVERVIEW SPONSORED SYMPOSIUM

AT THE FOREFRONT OF TREATMENT OF CUSHING SYNDROME

Chairman: Isabel Sousa *Divino Espírito Santo Hospital, Ponta Delgada*

Speaker: Davide Carvalho *Faculty of Medicine – University of Porto | European NeuroEndocrine Association, Oporto*

15:30-16:30h

RECENT ADVANCES NEDR SESSION

NOVEL TOOLS IN THE DIAGNOSIS OF GENETIC DISORDERS

Chairman: Lúcia Lacerda  *Doutor Jacinto Magalhães Medical Genetics Center, Oporto*

New gene sequencing techniques

Marta Amorim  *Lusíadas Hospital, Lisbon*

Metabolomics helping in the diagnosis

Francisco Laranjeira *Doutor Jacinto Magalhães Medical Genetics Center, Oporto*

16:30-17:00h

Coffee-break

17:00-18:00h

RECENT ADVANCES NEDR SESSION

BIOTECH ADVANCES IN RARE DISEASES

Chairman: Luís Brito Avô *CUF Tejo Hospital, Lisbon*

Speaker: Helder Mota Filipe  *Portuguese Pharmaceutical Board, Lisbon*

18:00-18:30h

Day 1 Symposium highlights and closing remarks

Chairman: Luís Brito Avô *CUF Tejo Hospital, Lisbon*

Speaker: Joana Santos *Cascais Hospital, Cascais*

SUNDAY – 26th FEBRUARY

08:45h Registration

09:00-10:00h **RECENT ADVANCES AND RARE OVERVIEW – WHAT’S NEW?**
PATIENT ORGANISATIONS, UMBRELLA ORGANISATIONS
AND ALLIANCES DEDICATED TO RARE DISEASES

Chairman: Luís Brito Avô *CUF Tejo Hospital, Lisbon*

What’s new in Portugal?

Paulo Gonçalves *RD Portugal, Lisbon*

State of the art Azores

Francisco Carreiro *APL – Associação Portuguesa das Doenças do Lisossoma, Azores*

10:00-11:00h **RECENT ADVANCES AND RARE OVERVIEW SPONSORED SYMPOSIUM**
NEUROFIBROMATOSIS TYPE 1 (NF1)

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Chairman: Diogo Cruz *Cascais Hospital, Cascais*

The clinical challenges of the management of NF1 patients

Speaker: João Passos *IPO Lisboa, Lisbon*

11:00-11:30h Coffee-break

11:30-12:00h **RARE AWARENESS PROGRAM – WHAT’S NEW?**

Chairman: Luísa Pereira *CUF Tejo Hospital, Lisbon*

PORFIRIAS 2022-23

Joana Santos *Cascais Hospital, Cascais*

New Projects 2023-24 – MPS “Find do Adulto”

José Duro *Lisboa Norte University and Hospital Center – Hospital Santa Maria, Lisbon*

12:00-12:30h **RECENT ADVANCES AND RARE OVERVIEW NEDR SESSION**
EDUCATIONAL PROGRAMS IN RARE DISEASES

Chairman: João Eurico da Fonseca  *Faculty of Medicine – University of Lisbon*
| Lisboa Norte University and Hospital Center – Santa Maria Hospital, Lisbon

Speaker: Patrício Aguiar  *Faculty of Medicine – University of Lisbon | Lisboa Norte*
University and Hospital Center – Santa Maria Hospital, Lisbon

12:30-13:00h **Day 2 Symposium highlights and symposium closing remarks**

Chairman: Luísa Pereira *CUF Tejo Hospital, Lisbon*

Speaker: Luís Brito Avô *CUF Tejo Hospital, Lisbon*

ORGANIZATION



ORGANIZING COMMITTEE

Diogo Cruz, Joana Santos, Luís Brito Avô, Luísa Pereira, Patrícia Aranha e Patrício Aguiar

SCIENTIFIC COMMITTEE

Diogo Cruz, Francisco Laranjeiro, Hélder Mota Filipe, Joana Santos, João Eurico da Fonseca, Lèlita Santos, Lúcia Lacerda, Luís Brito Avô, Luís Dias, Luísa Pereira, Marta Amorim, Patrícia Aranha, Patrício Aguiar e Paula Macedo

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