

10TH JULY / 10 DE JULHO

2021

ONLINE

1ST SYMPOSIUM
1º SIMPOSIO

RECENT
ADVANCES
&
RRARE
OVERVIEW

THE YEAR IN REVIEW 20/21
O ANO EM REVISTA 20/21

FINAL
Program



Scientific Sponsorship:



1st RARO Symposium

ORGANIZING COMMITTEE MESSAGE

The Rare Diseases Study Group (NEDR) of the Internal Medicine Portuguese Society (SPMI) will hold the 1st RARO Symposium (Recent Advances & Rare Overview) – the Year in Review 20/21, on July 10, 2021, in an 100% digital format.

In a RARE time where vaccines are developed in record time for a devastating pandemic and we all found ourselves, somehow deprived of access to innovation in areas other than SARS Cov2, we felt the need to start what we want to be the first of many RARO (Recent Advances and Rare Overview) Symposia.

This scientific event, as has been our organizational practice, 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.

We also intend, and taking advantage of what we have learned in a positive way to use in this pandemic *momentum*, to give a new dimension with the maximum range of themes, using the enormous projection and participation that the sessions in digital format brought us.

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

We invite you to be present at this online symposium, which will be completely innovative in the area of Rare Diseases in Portugal, on the 10th of July from 9 am to 6.30 pm.

On behalf of the 1st RARO Symposium Organizing Committee, our best regards and RARO greetings. Please take care of yourselves and your families. We hope to see you (virtually!) in July.

Luísa Pereira

Deputy Coordinator NEDR

1st RARO Symposium

TITLE:

The year in review 20/21

DATE:

10th of July 2021

ORGANIZING COMMITTEE:

Diogo Cruz, Luísa Pereira, Luís Brito Avô

SCIENTIFIC COMMITTEE:

Diogo Cruz, Lèlita Santos, Luísa Pereira, Luís Brito Avô, Patrício Aguiar

FACULTY MEMBERS:

The contribution from the nacional and the international scientific community:

- Carlos Aguiar, Lisbon, Portugal
- Derralynn Hughes, London, UK
- Diogo Cruz, Lisbon Portugal
- Eric Crombez, Boston, USA
- Lèlita Santos, Coimbra, Portugal
- Lígia Peixoto, Lisbon, Portugal
- Luís Brito Avô, Lisbon, Portugal
- Luísa Pereira, Lisbon, Portugal
- Manuel Praga, Madrid, Spain
- Maria Luisa Lozano, Múrcia, Spain
- Marta Leal Bento, Lisbon, Portugal
- Olga Azevedo, Guimarães, Portugal
- Patrício Aguiar, Lisbon Portugal
- Salil Patel, Cambridge, Massachusetts, USA
- Teresa Cardoso, Oporto, Portugal
- Ulrich Stölzel, Chemnitz, Germany

The Organizing committee is proud to enlist here above the colleagues that will serve as faculty members of the 1st RARO Symposium 2021, and warmly thank all of them for the valuable contribution they will give for a successful meeting.

1st RARO Symposium

Program **FINAL**

09H00 - WELCOME MESSAGES

Luísa Pereira (NEDR), Lèlita Santos (SPMI)

09H15 - RARE OVERVIEW: RARE DISEASES & COVID PANDEMIC

Session: The effects of the COVID Pandemic in the Rare Disease World

Chair: Luísa Pereira

Speakers: Luís Brito Avô, NEDR; Valter Fonseca, DGS; Paulo Gonçalves, RD Portugal

10H00- RECENT ADVANCES: Transtirretin Amyloid Cardiomyopathy

Session: TAFAMIDIS - New Data on Transtirretin Amyloid Cardiomyopathy (ATTR-CM)

Chair: Luís Brito Avô

Speakers: Carlos Aguiar, CHLO - Santa Cruz Hospital, Lisbon, Portugal

Sponsor: Pfizer

10H45 - RECENT ADVANCES AND RARE OVERVIEW: Gaucher Disease

Session: Gaucher Disease - Unravelling the Pathophysiology in the present, for a better future to our Patients

Chair: Patrício Aguiar

Speakers: Patricio Aguiar, CHLN - Santa Maria Hospital, Lisbon, Portugal; Derralynn Hughes, The Royal Free London NHS Foundation Trust, London, United Kingdom

Lecture 1: Late breaking news in Gaucher's disease pathophysiology

Lecture 2: Bone unmet needs: how can the research and the available treatments address them?

Sponsor: Takeda

11H30 - BREAK

11H45 - RECENT ADVANCES: SRT in Gaucher Disease

Session: Eliglustat: The importance of a new oral treatment option confirmed by Real World Evidence

Chair: Patrício Aguiar

Speakers: Marta Leal Bento, CHLN - Santa Maria Hospital, Lisbon, Portugal

Sponsor: Sanofi - Genzyme

12H30 - RECENT ADVANCES: Chronic Immune Thrombocytopenia (CIT)

Session: Recent Advances in CIT

Chair: Diogo Cruz

Speakers: Lígia Peixoto, CHLN - Santa Maria Hospital, Lisbon, Portugal; Maria Luisa Lozano, Hospital General Universitario Morales Meseguer, Murcia, Spain

Lecture 1: Diagnostic red flags

Lecture 2: Challenges and innovation

Sponsor: Sobi

13H15- LUNCH BREAK

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14H30 - RECENT ADVANCES: atypical Hemolytic Uremic Syndrome (aHUS)

Session: Malignant Hypertension and aHUS

Chair: Diogo Cruz

Speaker: Manuel Praga, 12 de Octubre Hospital, Madrid, Spain

Sponsor: Alexion

15H15 - RECENT ADVANCES: Fabry Disease

Session: Galafold: New data, Real-world experience

Chair: Luísa Pereira

Speaker: Teresa Cardoso, CHUSJ - São João Hospital, Oporto, Portugal

Sponsor: Amicus

16H00 - BREAK

16h15 - RECENT ADVANCES: RNA interference - From lab bench to bedside

Session: How is RNAi leading the transformation in therapeutical approach?

Chair: Luís Brito Avô

Speakers: Salil Patel, SVP, Medical Affairs Alnylam, Cambridge, Massachusetts, USA; Ulrich Stölzel, Klinikum Chemnitz, Chemnitz, Germany

Lecture 1: RNA interference mechanism of action and therapeutical platforms

Lecture 2: Acute Hepatic Porphyria pathophysiology , chronic impact and treatment

Sponsor: Alnylam

17H00 - RECENT ADVANCES AND RARE OVERVIEW: Gene Therapy

Session: Ultragenyx - an overview on gene therapy programs

Chair: Luís Brito Avô

Speakers: Eric Crombez, Ultragenyx Chief Medical Officer, USA

Sponsor: Ultragenyx

17H45 - RECENT ADVANCES: Treatment Strategies

Session: GUIMARÃES PILOT PROJECT - Home-Based Care for Patients with Lysosomal Storage Disease

Chair: Luísa Pereira

Speaker: Olga Azevedo, Nossa Sra da Oliveira Hospital, Guimarães, Portugal

18H15 - CLOSING REMARKS: Symposium Highlights

Luís Brito Avô, Luísa Pereira

1st RARO Symposium

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