21ST INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS

RAISING AWARENESS ON INHERITED METABOLIC DISORDERS

26th March | SPDM Working & Nutrition Groups Meeting
27th - 28th March | 21st SPDM International Symposium
29th March | Raising awareness in innovative therapies in IMD

SANA METROPOLITAN HOTEL, LISBON

E-posters guided walk

(Jury: Dulce Quelhas, Patrícia Lipari Pinto)

Station 1:

1. **PO03** "Non-syndromic retinitis pigmentosa caused by COQ8B bi-allelic variants" – **Cristina Santos**
2. **PO25** "Macular Pattern Dystrophy as a Key Feature in Suspected Maternally Inherited Diabetes and Deafness (MIDD): A Case-Based Approach"- **Sara Jesus**
3. **PO04** "Impact of mitochondrial DNA variants in inherited retinal disorders: a revision utilizing the Portuguese national IRD.pt registry" – **Francisca Amaral**
4. **PO06** "Osteogenesis Imperfecta - Experience from a Paediatric Centre"- **Rui Diogo**
5. **PO35** "REVERSE PHENOTYPING AFTER NGS PANEL OF X-LINKED INTELLECTUAL DISABILITY UNRAVELS CREATINE TRANSPORTER (SLC6A8) DEFICIENCY" – **Gonçalo Padeira**
6. **PO08** "Case study: methylmalonic acidemia and episodes of pancreatitis requiring parenteral nutrition" – **Inês Agostinho**
7. **PO38** "A rare complication of Diabetes Mellitus – The Mauriac Syndrome" – **Inês Marques Ferreira**
8. **PO12** "Rhabdomyolysis in Inborn Errors of Metabolism" – **Rosário Stilwell**
9. **PO33** "Genetic insights into adult cardiomyopathy: unraveling mitochondrial disorders through NGS" – **João Durães**
10. **PO15** "Twenty Years of Newborn Screening for MCADD in Portugal: genetic data" – **Helena Fonseca**
11. **PO11** "Forty-four years of newborn screening in Portugal: new challenges, the same commitment to the community" – **Ana Marcão**
12. **PO09** "After all, Porphyria exists in Portugal! A three-year study" – **Filipa Ferreira**
13. **PO05** "Validation of Portuguese version of SCOFF questionnaire to assess eating disorders in adult IMD patients requiring dietary treatment" – **Inês Mendes**

SAVE THE DATE
26th-28th March

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Station 2:

(Jury: Cristina Florindo, Daniel Costa Gomes)

1. **PO27** "DIAGNOSIS OF NIEMANN PICK DISEASE TYPE C IN A PATIENT WITH CHARCOT-MARIE-TOOTH NEUROPATHY - THE IMPORTANCE OF VALUING ATYPICAL SIGNS" –
   Gonçalo Passos Croca
2. **PO36** "Early diagnosis of acid sphingomyelinase deficiency (ASMD) through biomarkers analysis" –
   Raquel Neiva
3. **PO28** "Niemann-Pick Type B: A lifelong battle – Early diagnosis as the key for better outcome" –
   Ana Gonçalves
4. **PO10** "Review of ASMD adult patients from single reference center" –
   Arlindo Guimas
5. **PO16** "Acetyl-Leucine as an adjunct therapy in Niemann-Pick Disease Type C: A Case Report" –
   Inês Aires Martins
6. **PO24** "Olipudase alfa enzyme replacement therapy. One-year outcomes in an adult patient with acid sphingomyelinase deficiency type B" –
   Maria Teresa Cardoso
7. **PO17** "Early Diagnosis of Mucopolysaccharidoses in Pediatrics" –
   Paulo Gaspar
8. **PO07** " Differential Diagnosis of α-Mannosidosis in MPSs" –
   Paulo Gaspar
9. **PO20** "When Does Klinefelter Syndrome Interfere with the Diagnosis of Fabry Disease?" –
   Ana Mendonça
10. **PO32** "Pompe disease: an Azorean family report" –
    Raquel Parece
11. **PO23** "Missense Mutation in the LYRM7 Gene: A Case of Leukoencephalopathy" –
    Bárbara Gonçalves
12. **PO18** "Gone with the Wind" –
    Anabela Bandeira

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Station 3:

(Jury: Hugo Rocha, Rita Santos Loureiro)

1. **PO14** "Protein intake and prevalence of overweight and obesity in patients with phenylketonuria: a 10 year-longitudinal TNSPKU study" – **Catarina Rodrigues**
2. **PO37** "Neurocognitive outcome and personality profile of 28 adult PKU patients followed-up at CGM/ULS de Santo António: a retrospective study" – **Carla Carmona**
3. **PO01** "The Usefulness of Plasma Methylmalonic Acid and Homocysteine as Biomarkers to Assess Vitamin B12 Restriction" – **Cristina Florindo**
4. **PO34** "WES revisited - solving an unsolved inherited retinal disease case" – **Cristina Santos**
5. **PO13** " Serine metabolism disorder - one more metabolic aetiology of cerebral palsy" – **Helena Santos**
6. **PO29** "CPS1 diagnosed in adolescence" – **Joana Tenente**
7. **PO22** "Mapple Syrup Urine Disease – a diagnosis at an elderly age" – **Inês Marques Ferreira**
8. **PO31** "A challenging case of Classic Galactosemia" – **Sara Ferreira**
9. **PO26** "Dilated Cardiomyopathy as a Presentation of Neutral Lipid Storage Disease with Myopathy: A Diagnostic Challenge" – **Inês Santos**
10. **PO30** "Tangier Disease: a rare disorder of lipid metabolism" – **Mariana Pintalhão**
11. **PO19** "Pediatric Smith-Lemli-Opitz Syndrome follow at a Portuguese Level III Hospital" – **Teresa Cachada Baptista**
12. **PO02** "Alkaptonuria - a call for action" – **Catarina Salvador**
13. **PO21** "Longitudinal assessment of body composition in an infant with phenylketonuria treated at an early age" – **Júlio César Rocha**