Programme

Thursday, 20 March

08:00	**Registration Opening**
08:50	**Symposium Opening - Welcome Address on behalf of the SPDM** Isabel Tavares de Almeida, SPDM President (Lisboa, PT)

Session I – Mitochondrial Energy Metabolism

Chairpersons:
Isabel Rivera | *iMed.UL, FFUL,PT*
Manuela Grazina | *Lab. Bioch. Genetics, FMUC, PT*

09:00	**Defects of lipoic acid biosynthesis and incorporation** *Garry Brown*	*Oxford Univ., United Kingdom*
09:30	**Pyruvate oxidation deficiency: new insights and relevance to human disease** *Wolfgang Sperl*	*Paracelsus Medical Univ., Austria*
10:00	**Lypoylation defect of the 2-ketoacid dehydrogenase complexes: a new diagnostic challenge** *Antonia Ribes*	*Hospital Clinic, Barcelona, Spain*
10:30	Coffee Break
11:00	**Mitochondrial fatty acid oxidation: enzymology, newborn screening, new therapeutic options and novel defects** *Ron Wanders*	*Amsterdam Medical Center - Amsterdam Univ., The Netherlands*
11:30	**Carnitine shuttle deficiency in fatty acid oxidation: the contribution of the peroxisomes to the acylcarnitine homeostasis** *Sara Violante*	*Mount Sinai Hospital, NY-USA & MetGen.iMed. Lisboa Univ., Portugal*
11:50	**The muscle image in the suspicion of fatty acid oxidation defect** *Teresinha Evangelista*	*Newcastle Univ., United Kingdom*
12:10	**Update on clinical aspects, molecular mechanisms and treatment of selected mitochondrial disorders** *Rita Horvath*	*Newcastle Univ., United Kingdom*
12:40	Poster View / Lunch

Session II – Intermediary Metabolism Defects

Chairpersons:
Margarida Baptista e Silva | *iMed.UL,FFUL,PT*
Claúdia Costa | *Hosp. Sta Maria,CHLN, PT*

13:50	**The relevance of 3-methylglutaconic aciduria in inborn errors of metabolism** *Ron Wevers*	*Radboud Univ.- Medical Center, The Netherlands*
14:20	**Inherited disorders of glucose transport** *René Santer*	*Hamburg Univ., Germany*

Session III – Methionine Metabolism Defects

Chairpersons:

Rita Castro | *iMed.UL,FFUL,PT*
Esmeralda Martins | *C. Hospitalar Porto, PT*

14:50	**Inherited methylation disorders: clinical presentation, biomarkers and therapeutic possibilities** *Ivo Baric* | *Zagreb Univ., Croatia*
15:15	**New insights in cobalamine defects: an update** *Manuel Schiff* | *Robert Debré Univ. Hosp., France*
15:40	**Folate and methionine metabolism: a crossroad with several tales** *Henk Blom* | *Freiburg Univ., Germany*
16:05	Coffee Break

Session IV – Oral Communications

Chairpersons:
Sílvia Sequeira | *Hosp. D. Estefânia, CHLC, PT*
Paula Leandro | *iMed.UL, FFUL, PT*

16:30	**Identification of a novel SURF1 gene mutation in Leigh syndrome** *Carolina Ribeiro* | *CNC, Biochemical Genetics Lab., U. Coimbra, PT*
16:45	**Phenotypic spectrum of mitochondrial disorders in a pediatric neurology clinic** *Sandra Jacinto* | *Hosp. D. Estefânia, CHLC Lisboa, PT*
17:00	**Modulation of the mitochondrial acetylome by a fatty acid β-oxidation inhibitor: a novel mechanism with implications on metabolic homeostasis** *Marco F. Moedas* | *Met&Gen, iMed.UL, Lisboa, PT*
17:15	**Lenz-Majewski syndrome: disturbed phosphatidylserine metabolism causes intellectual disability and a sclerosing bone dysplasia** *Sérgio B. Sousa* | *Medical Genetics Unit, Hosp. Pediátrico Coimbra, U. Coimbra, PT*
17:30	**PMM2-CDG (CDG IA): phenotype/genotype characterization of seven patients** *Anabela Bandeira* | *Unid. de D. Metabólicas, CHPorto, PT*
17:45	**S-Adenosylhomocysteine alters the selenoproteome through RNA hypomethylation** *Madalena Barroso* | *Met&Gen, iMed.UL, Lisboa, PT*
18:00	End of the Session

18:30	Departure from the hotel to Cascais Historical Centre
20:00	Symposium Dinner at the Cascais Cidadela (Fortress)
22:30	Return to the Hotel Quinta da Marinha

Friday, 21 March

Session V – Oral Communications

Chairpersons:
Laura Vilarinho | *Inst. Nac. Saúde (INSA), PT*
Luisa Diogo | *Hosp. Pediátrico Coimbra, PT*

09:00	**Exome sequencing in five families with inherited metabolic diseases** *Frederic Tort* | *Hosp. Clinic, IDIBAPS, CIBERER, Barcelona, SP*
09:15	**Glutaric aciduria type 1, before and after expanded newborn screening: the experience of a portuguese metabolic diseases unit.** *Cláudia Dias da Costa* | *Unid. de D.Metabólicas, Serv. de Pediatria, CHLN, Lisboa, PT*
09:30	**Establishing experimental platforms towards structural and functional studies on ETF: QO** *Tânia G. Lucas* | *ITQB, U. Nova de Lisboa, PT*
09:45	**Protein aggregation dictates the fate of the most prevalent missense mutations in classic galactosemia** *João B. Vicente* | *Met&Gen, iMed.UL, Lisboa, PT*
10:00	**Clinical, biochemical and molecular studies: stepwise to achieve diagnosis of Fabry disease** *Lúcia Lacerda* | *Unid. Bioq. Genética, C.G. Médica, CHPorto, PT*
10:15	**Unverricht-Lundborg disease: development of splicing therapeutic approaches for a patient with an homozygous mutation in the cystatin b gene** *Liliana Matos* | *Human Genetics Dep., INSA/Fac. of Sciences - U.Porto, PT*
10:30	Coffee Break

Session VI – Lysosomal Disorders

Chairpersons:
Elisa Leão Teles | *Hosp. S. João, CHS João, PT*
Teresa Cardoso | *Hosp. S. João, CHS João, PT*

11:00	**Clinical neurogenetics: neuropathic lysosomal storage disorders** *Gregory Pastores	Mater Misericordiae Univ. Hosp., Dublin, Ireland*
11:20	**Induced Pluripotent Stem cells for testing small compounds for treatment of neuronopathic Gaucher's Disease** *Gustavo Tiscornia	Algarve Univ., Faro, Portugal*
11:40	**Morquio Clinical trial: from the design of the study to the results** *Paula Garcia	Ped. Hosp. Coimbra, Coimbra, Portugal*
12:00	**Niemann-Pick type C suspicion index tool** *Mercedes Pineda	Hosp S. Joan Deu, Barcelona, Spain*
12:20	Lunch / Poster View
12:50	**SPDM Assembleia Geral**

Session VII – Clues in the Nutritional Management

Chairpersons:
Paula Garcia | *Hosp. Pediátrico Coimbra, PT*
Manuela Almeida | *C. Genética Médica - C. Hospitalar do Porto, PT*

14:00	**Nutritional management: results of the survey for urea cycle disorders** *Anita McDonald	Birmingham Children's Hospital, United Kingdom*
14:20	**Challenges in nutritional follow up in EM in developing countries. Experience of Chile** *Verónica Cornejo	Univ. of Chile, Chile*
14:40	**Nutritional management in PKU: growth, body composition and obesity** *Júlio César Rocha	C. Genética Médica - C. Hospitalar do Porto, Portugal*

Session VIII – Short Oral E-posters Presentations

Chairpersons:
Hugo Rocha | *Inst. Nac. Saúde (INSA), PT*
Patrícia Janeiro | *Hosp. Sta Maria, CHLN, PT*

15:05	**A C.545+5G>A NFU1 splice site mutation leads to a particular biochemical phenotype** *Xènia Ferrer-Cortès	Hospital Clínic, IDIBAPS, CIBERER, Barcelona, ES*
	Structural and functional regulation by substrate induced protein post-translational modifications: studies on lysine glutarylation of the β-oxidation enzyme glutaryl-CoA dehydrogenase *Sara V. Francisco	Instituto de Biologia Experimental e Tecnológica, Oeiras, PT*
	Structural and dynamic behavior of medium-chain Acyl-CoA dehydrogenase by molecular dynamics simulations *Fátima Ventura	Met&Gen, iMed.UL, Lisboa, PT*
	Mitochondrial dysfunction as a pehnotypic modifier in methylmalonic aciduria type B *Sandra Brasil	Centro de Diagnóstico de Enfermedades Moleculares, Centro de Biología Molecular Severo Ochoa, CIBERER, ES*
	Aminoacylase 1 deficiency: the first Portuguese case of a novel inborn error of metabolism *Carla Valongo	Unid. de Rastreio Neonatal, Metabolismo e Genética, Instituto Nacional de Saúde Doutor Ricardo Jorge, I.P., Porto, PT*
	Clinical course and outcome of classical organic acidurias in a Portuguese metabolic diseases unit *Patrícia Janeiro	Unid. de Doenças Metabólicas, Serv. de Pediatria, CHLN, Lisboa, PT*
	Riboflavine responsive MADD presenting with a VLCADD like profile at newborn screening *Helena Santos	Child and Adolescence Neurosciences Unit, Pediatrics Department, Vila Nova de Gaia/Espinho Hospital Center*
	X-linked adrenoleukodystrophy - 29 years later *Orlando Galego	Hospital Pediátrico de Coimbra, CHUC, PT*

16:00	Coffee Break

Session IX – Rare Diseases: the Institutional Point of View

Chairpersons:
Isabel Tavares de Almeida, *SPDM President (Lisboa, PT)*
Luisa Diogo, *SPDM Vice-President (Coimbra, PT)*

16:25	**Orphan Drugs: the regulatory perspective** *Bruno Sepodes | COMP – EMA. EU / FFUL, Lisboa, Portugal*
16:45	**Rare Diseases: the Portuguese National Health Service perspective** *Alexandre Diniz | DGS, Department of Health, Portugal*
17:00	**The importance of a Metabolic Unit within the Pediatric Department** *Maria Céu Machado | Sta Maria Univ. Hosp., Lisboa, Portugal*

17:15	**Awards / Closing Remarks**
17:40	**END**
17:45	**SPDM Assembleia Eleitoral**