



Sociedade Portuguesa de Genética Humana

18th Annual Reunion | 19 – 21 November 2014

Venue | *Egas Moniz Building, Faculty of Medicine of the University of Lisbon (FMUL) – Portugal*

Preliminary Programme

Day 1 19.Nov.2014 (Wednesday)

Clubs: Parallel Sessions

16:00 – 17:45	<p><i>Cytogenetics and molecular genetics</i> (Title to be confirmed) <i>Chairs João Gonçalves Filomena Brito</i></p>	<p><i>Medical genetics and clinical dysmorphology</i> Rasopathies <i>Chairs Oana Moldovan Patrícia Dias</i></p>
17:45 – 18:15	<p>Target Seq (AmpliSeq™), the example of Colon and Lung Cancer <i>Presenters Juan Barba and João Caldeira, Life Technologies, brand of Thermo Fisher Scientific</i></p>	

End of Day 1

Day 2 20.Nov.2014 (Thursday)

08:00 – 08:45	<i>Registration form</i>
08:45 – 09:00	<p><i>Opening Ceremony SPGH 18th Annual Reunion</i> Hildeberto Correia, Juliette Dupont and José Pereira Miguel</p>
09:00 – 09:45	<p>Da Farmacogenética à Ecogenética: o Passado o Presente e os desafios Futuros <i>Manuel Bicho, Instituto de Medicina Molecular, Faculdade de Medicina da Universidade de Lisboa (FMUL), Portugal</i></p>
09:45 – 10:15	<i>Coffee-break / Poster Session</i>
10:15 – 10:45	<p style="text-align: center;"><i>1st. Scientific session – New challenges in Pre-natal Diagnosis</i> <i>Chairs Paula Caetano and Isabel Marques</i></p> <p>Non-invasive prenatal diagnosis using maternal blood as performed in a clinical genetics department: Over a decade of translational research <i>Ana Bustamante, Servicio de Genética, Hospital Universitario Fundación Jiménez Díaz, Instituto de Investigaciones Sanitarias, FJD, Madrid, Spain</i></p>
10:45 – 11:15	<p>From the conception to Prenatal testing: a technical revolution to change the Reproductive Medicine <i>Marcus Hausch, ILLUMINA EMEA RGH Marketing Manager</i></p>

11:15 – 11:45	Why Genotype Information Matters in Prenatal SNP Array Diagnostics Nicole de Leeuw, Department of Human Genetics, Radboud University Medical Center, Nijmegen, the Netherlands
11:45 – 12:15	Dilemmas in prenatal diagnosis Ana Berta, Serviço de Genética Médica, Hospital de Santa Maria – Centro Hospitalar Lisboa Norte (HSM-CHLN), Portugal
2nd. Scientific session – Paramyloidosis: Advance in Care <i>Chairs Lina Ramos and Jorge Sequeiros</i>	
12:15 – 12:45	TTR - FAP: clinical and genetic aspects in non-endemic areas David Adams, French Reference Center for FAP (NNERF), Paris, France
12:45 – 13:00	TTR - FAP: therapeutic approach Isabel Conceição, Serviço de Neurologia, Hospital de Santa Maria – Centro Hospitalar Lisboa Norte (HSM-CHLN), Portugal
13:00 – 14:00	<i>Lunch / Poster Session</i>
3rd. Scientific session – Cancer breaking news <i>Chairs Manuel Teixeira and Glória Isidro</i>	
14:00 – 14:30	Hemato-oncological diseases – New therapies Cristina João Pires, Instituto Português de Oncologia de Lisboa Francisco Gentil (IPOLFG), Portugal
14:30 – 15:00	Genes and neuroblastoma – hand in hand in therapeutic advances in Europe (developments within SIOPEN) Cláudia Constantino and Ana Lacerda, Instituto Português de Oncologia de Lisboa Francisco Gentil (IPOLFG), Portugal
15:00 – 15:30	Using genomics for drug discovery in medulloblastoma Cláudia Faria, Serviço de Neurocirurgia, Hospital de Santa Maria – Centro Hospitalar Lisboa Norte (HSM-CHLN), Portugal
15:30 – 16:00	<i>Coffee-break / Poster Session</i>
Oral Presentations <i>Chairs Sofia Dória and Astrid Vicente</i>	
16:00 – 17:30	6 selected presentations from submitted abstracts (10 minutes each and 5 minutes for each presentation)
18:00	SPGH General Assembly
20:30	<i>Conference dinner (restaurant)</i>
End of Day 2	

Day 3	21.Nov.2014 (Friday)
4th. Scientific session – Hemoglobinopathies in Portugal <i>Chairs Paula Faustino and Luísa Romão</i>	
09:00 – 09:30	Epidemiology, pathophysiology and genetic modifiers of morbidity João Lavinha, Departamento de Genética Humana, Instituto Nacional de Saúde Dr. Ricardo Jorge (INSA), Lisboa, Portugal
09:30 – 10:00	Prevention strategies Armandina Miranda and João Gonçalves, Instituto Nacional de Saúde Dr. Ricardo Jorge (INSA), Lisboa, Portugal

10:00 – 10:30	Treatment – current clinical management and future prospects Alexandra Dias, Hospital Prof. Doutor Fernando Fonseca (HFF), Amadora, Portugal
10:30 – 11:00	<i>Coffee-break / Poster Session</i>
5th. Scientific session – New approaches in Rare diseases <i>Chairs Ana Fortuna and Jorge Saraiva</i>	
11:00 – 11:30	Gene packages and opening the exome – the Nijmegen experience Ilse Feenstra , Department of Human Genetics, Radboud University Medical Center, Nijmegen, the Netherlands
11:30 – 12:00	Causes of rare and recurrent disorders unravelled by array Nicole de Leeuw, Department of Human Genetics, Radboud University Medical Center, Nijmegen, the Netherlands
12:30 – 14:00	<i>Lunch / Poster Session</i>
<i>Chairs Margarida Reis and Maria Rosário Almeida</i>	
14:00 – 14:45	Genetic complexity of hereditary cardiomyopathies in the high-throughput sequencing era – from research to clinical practice Luís Lopes, Hospital Garcia de Orta (HGO), Almada, Portugal
Oral Presentations <i>Chairs Susana Mendes and Oana Moldovan</i>	
14:45 – 15:45	4 selected presentations from submitted abstracts (10 minutes each and 5 minutes for each presentation)
16:00 – 16:30	<i>Coffee-break / Poster Session</i>
6th. Scientific session – Bioethics SPGH Ethics Commission <i>Chairs Carolino Monteiro and Célia Ventura</i>	
16:30 – 17:30	Reflexões bioéticas e recomendações sobre os Rastreios ao Recém-Nascido em Portugal Luísa Diogo, Hospital Pediátrico de Coimbra / Faculdade de Medicina da Universidade de Coimbra (FMUC), Coimbra, Portugal André Pereira, Faculdade de Direito da Universidade de Coimbra (FDUC), Coimbra, Portugal Heloísa Santos, President of SPGH Bioethics Commission, Lisboa, Portugal
SPGH Awards <i>Chairs Hildeberto Correia and Juliette Dupont and Maria do Céu Machado</i>	
17:30 – 18:30	Award Conference Basic and Clinical Research Awards
18:30	Close of Conference SPGH 18th Annual Reunion Hildeberto Correia, Juliette Dupont and Maria do Céu Machado
End of the Reunion	

A Direção da Sociedade Portuguesa de Genética Humana – SPHG 2014

Presidente 2014

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Secretária 2014

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