

**Friday 17.06.2005**

**8h30 CONFERENCE SECRETARY OPENING**

**9h00 A MEDICINA GERAL E FAMILIAR E A GENÉTICA MÉDICA**

*General Practice and Medical Genetics (session in Portuguese)*

**Moderadores:** Eufémia Ribeiro e Beatriz Calado  
Sinais de Alerta Ecográficos na Grávida  
Luís Graça e Nuno Clode, Maternus, Lisboa  
Risco em Rastreamento Pré-Natal  
Susana Pereira, Hospital de São Teotónio, Viseu  
Genética Médica na Criança  
Luís Nunes, Hospital D. Estefânia, Lisboa  
A Importância da Embriofetopatologia na Medicina Materno Fetal  
Ricardo Laurini, UEFP, CGC, Porto; Lund University Hospital, Lund, Sweden

**Pause**

**10h30 QUALIDADE NA PRESTAÇÃO DE CUIDADOS PRIVADOS DE SAÚDE**

*Quality in Private Health Care (session in Portuguese)*

Qualidade na Saúde  
Luís Pisco, IQS, Lisboa  
Liberdade de Escolha  
Germano de Sousa, Centro de Medicina Laboratorial, Lisboa  
O Custo da Saúde  
Isabel Vaz, Espírito Santo Saúde, Lisboa

**12h00 OPENING CEREMONY**

**Chair:** Purificação Tavares and Jan Marshall  
**Keynote Speaker**  
Genomics, Globalization and Health Care  
Victor Penchaszadeh, Columbia University, New York, USA

**13h30 Lunch**

**15h00 ALSTRÖM SYNDROME: (RE)VIEWS**

**Chair:** Richard Paisey and Sebastian Beck  
Historical overview of Alström Syndrome research and emerging new phenotypes  
Jan Marshall, JAX, Bar Harbor, USA  
Is hyperinsulinism the final common pathway in endocrine disorders and tissue fibrosis in Alström Syndrome?  
Richard Paisey, South Devon Health Care NHS Trust, Torquay, UK  
Cardiomyopathy in Alström Syndrome: presentation evolution and management  
Andrew Warren, IWK Health Centre, Halifax, Canada

**15h30 PARALLEL SESSION: SISTEMAS DE CERTIFICAÇÃO/ACREDITAÇÃO DA QUALIDADE NA SAÚDE**

*Quality Certification/Acreditation Systems in Health Care (session in Portuguese)*

**Moderador:** Adalberto Campos Fernandes  
Sistema de Certificação e Acreditação da Qualidade em Serviços de Saúde (NP ISO 9001, 14001, OSHAS 18001, 15189 e 17025)  
Carla Gonçalves Pereira, Sinase, Lisboa  
Acreditação de Saúde pela Join Commission International  
Herminio Henrique, Sinase, Lisboa  
A Implementação da ISO 9001:2000 no CGC  
Paula Rendeiro, CGC, Porto  
A Certificação do CGC  
Purificação Tavares, CGC

**16h30 POSTER DISCUSSION**

**Moderators:** Pietro Maffei and Jorge Sequeiros  
**Pause**

**17h00 SCIENCE UPDATE**

**Chair:** Gayle Collin and Maximina Pinto  
Characterization of Alms1-deficient mice, a model for human Alström Syndrome.  
Gayle Collin, JAX, Bar Harbor, USA  
Further characterization of cytoplasmic glycoproteins in tissues from Alström patients.  
Roderick Bronson, Harvard University, Boston, USA  
Subcellular localization of ALMS1 supports involvement of centrosome and basal body dysfunction in the pathogenesis of Alström syndrome  
David Wilson, University of Southampton, Southampton UK  
A correlation study among alms1 expression, insulin resistance, and in vitro model of myogenesis and adipogenesis  
Caterina Veronese, University of Padua, Padua, Italy

**Saturday 18.06.2005**

**9h00 CLINICS 1: SYMPTOMS AND COMPLICATIONS**

**Chair:** Andrew Warren and Heloisa Santos  
Gastrointestinal and hepatic complications in Alström Syndrome  
Judy Davis, California Digestive Disease Center, Fresno, USA  
Hypogonadism in Alström Syndrome.  
Pietro Maffei, University of Padua, Padua, Italy  
Augmentation index, brain natriuretic peptide, insulin levels and cardiomyopathy in Alström syndrome  
Richard Paisey, South Devon Health Care NHS Trust, Torquay, UK

**Pause**

**11h30 CLINICS 2: CASE REPORTS**

**Chair:** Pietro Maffei and José Carlos Ferreira  
Alström Syndrome (ALMS) case report  
Sonia Mastrapasqua, Hospital Provincial Neuquén, Argentina  
Impaired GH/IGF1 axis in 2 adults with Alström Syndrome  
Cristina M. Mihai, Constanta County Hospital, Constanta, Romania  
Alström Syndrome: clinical and molecular characterization of two portuguese families  
Cristina Dias, Instituto de Genética Médica, Porto, Portugal  
Molecular analysis and long term clinical follow up of three siblings with alström syndrome in a large consanguineous family  
Koksal Ozgul, Hacettepe University, Institute of Child Health, Sıhhiye, Ankara, Turkey

**13h00 Lunch**

**14h30 CLINICS 3: MEDICAL GENETICS OF ALSTRÖM SYNDROME**

**Chair:** Judy Davis and Jorge Saraiva  
Spectrums of mutations within the ALMS1 gene  
Elizabeth G Hinman, JAX, Bar Harbor, USA  
The psychological challenges of Alström Syndrome  
Ian Bennun, Torbay Hospital, Torquay, UK  
The importance of transcultural genetic counselling in consanguineous families with Alström syndrome  
Gulshan Karbani, St. James's University Hospital, Leeds, UK

**Pause**

**17h00 PERSPECTIVES ON ALSTRÖM SYNDROME Round Table**

**Chair:** Jan Marshall and Roderick Bronson

**18h00 CLOSURE**