



European School of Genetic Medicine
20th Course in Medical Genetics
 May 5th – 11th, 2007 Bertinoro, Italy

دانشگاه علوم پزشکی تهران
 گروه ژنتیک پزشکی



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دارای ۱۰ امتیاز بازآموزی

شبکه پزشکی مولکولی کشور

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All times are in Italian local time (Last update: January 16th, 2007)

DIRECTORS: McKusick (Baltimore, USA), Brunner (Nijmegen, Netherlands), Antonarakis (Geneva, Switzerland), Romeo (Bologna, Italy)

FACULTY: Capecchi (USA), Cucca (Sassari, Italy), Dragani (Milano, Italy), di Bernardo (Naples, Italy), Emanuel (Philadelphia, USA), Katsanis (Baltimore, USA), Lichter (Heidelberg, Germany), Mandel (Strasbourg, France), Matthijs (Leuven, Belgium), Meitinger (Munich, Germany), Mendell (Baltimore, USA), Metspalu (Tartu, Estonia), Monckton (Glasgow, Scotland), Speicher (Gratz, Austria), Stevanin (Paris, France), Todd (Cambridge, UK), Zeviani (Milano, Italy)

Saturday May 5 – Introduction to Genetic Analysis

Morning Session: 8.30 – 13.00

V.A. Mc Kusick Some new perspectives on the history of medical genetics & genomics
H. Brunner Genotypes and phenotypes
G. Stevanin Linkage analysis
B. Emanuel FISHing for Breaks on Ch. 22: A Paradigm for the Analysis of Genomic Disorders

Sunday May 6 – Clinical Genetics and Cytogenetics

H. Brunner Chromosomal syndromes
M. Speicher Development of technologies for the new cytogenetics
B. Emanuel Studies to find Modifier Genes for the 22q11.2 Deletion
S. Antonarakis Aneuploidy

Monday May 7 – From clinical observations to the lab

S. Antonarakis Nature & consequences of human gene mutations
H. Brunner Understanding genetic heterogeneity
G. Matthijs Disorders of glycosylation
J. Mendell MicroRNAs

Tuesday May 8 – Translational research in genetics

N. Katsanis Dissecting oligogenic traits
G. Romeo Mitochondrial mutation and cancer
G. Stevanin Genetic & physiopathological aspects of spastic paraplegias
N. Katsanis t.b.d

Wednesday May 9 – System Biology and Animal Models

D. di Bernardo System Biology approaches to drug discovery and gene function
P. Lichter Challenges in Tumor Genetics Approached by Molecular Profiling
T. Dragani t.b.d
M. Capecchi Modeling Human Disease in the Mouse

Thursday May 10 – Neurogenetics

D. Monckton Myotonic dystrophy: simple repeats in a complex disorder
J. L. Mandel Fragile X syndrome
D. Monckton Molecular mechanisms of genetic instability in the triplet repeat disorders
M. Zeviani Mitochondrial neuropathies

Friday May 11 – Complex genetic disorders

J. Todd Genetic analysis of common disease using genome-wide association
F. Cucca Genetic analysis of complex traits in isolated populations: facts and myths
T. Meitinger Genome-wide association studies for quantitative traits in man
A. Metspalu The Estonian Genome project