

European School of Genetic Medicine
16th COURSE IN MEDICAL GENETICS
March 23-29, 2002
Bertinoro, Forlì, Italy

University Residential Centre
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Directors:

V.A. McKusick (Baltimore, USA)

S.E. Antonarakis (Geneva, CH)

G. Romeo (Bologna, I)

Faculty:

Adriano Aguzzi (Zurich, CH), Sandro Banfi (Naples, I), Martin Bobrow (Cambridge, UK), Peter Byers (Seattle, USA), Chris Cannings (Sheffield, UK), Aravinda Chakravarti (Baltimore, USA), Maria Pia Cosma (Naples, I), Francesco Cucca (Cagliari, I), Maria Benedetta Donati (Campobasso, I), Luis J.V. Galietta (Genova, I), Peter Harper (Cardiff, UK), Julie Hoover-Fong (Baltimore, USA), Nico Katsanis (Baltimore, USA), Elio Lugaresi (Bologna, I), Elena Maestrini (Bologna, I), Stavros Malas (Nicosia, CY), Jean-Louis Mandel (Strasbourg, F), Thomas Meitinger (Munich, D), Akhilesh Pandey (Baltimore, USA), Kerry Rhoden (New Haven, USA), Albert Schinzel (Zurich, CH), Giulio Superti-Furga (Heidelberg, D), Stefano Volinia (Ferrara, I), Robin Winter (London, UK), Joel Zlotogora (Jerusalem, IL).

Sunday, March 23, 2003
INTRODUCTION TO GENETIC ANALYSIS

Morning Session

8.30	V.A. McKusick (Baltimore, USA) March of Dimes key-note lecture
9.20	P. Harper (Cardiff, UK) Myotonic dystrophy - lessons for basic genetics and biology from a clinical disease
10.10	Coffee break

- 10.30 **J. Zlotogora** (Jerusalem, IL)
Why are genetic diseases frequent?
- 11.20 **M. Bobrow** (Cambridge, UK)
Advances in Genetics - Medical social and ethical issues
- 12.30 Questions from students on morning lectures
- 13.00 Lunch

Afternoon Session

CONCURRENT WORKSHOPS

- 14.30 **E. Maestrini** (Bologna, I)
Linkage analysis with pencil and paper
- F. Vogl** (Lyon, F)
Linkage analysis with computer
- 16.00 Coffee break
- 16.30 **C. Cannings** (Sheffield, UK)
General aspects of IBD
- J. Zlotogora** (Jerusalem, IL)
Medical consequences of consanguinity
- J. Hoover-Fong** (Baltimore, USA)
Pedigree construction and analysis
- A. Schinzel** (Zurich, CH)
Uniparental disomy in human and its consequences
- S. Malas** (Nicosia, CY)
How to design a mouse mutant using transgenic technology
- S. Volinia** (Ferrara, I)
Analysis of Microarray Data: Gene Ontology and Genomics
- P. Harper** (Cardiff, UK)
Genetic counselling

Monday, March 24, 2003

GENETICS OF COMPLEX TRAITS

Morning Session

- 8.30 **C. Cannings** (Sheffield, UK)
Identity by descent (IBD) across the genome
- 9.20 **E. Maestrini** (Bologna, I)
Genetic analysis of complex disorders: the example of autism
- 10.10 Coffee break

10.30	F. Cucca (Cagliari, I) Dissection of the genetic bases of common multifactorial traits in the founder isolated population from Sardinia
11.20	M.B. Donati, L. Iacoviello (Campobasso, I) Genetic modulation of inflammatory and thrombosis in cardiovascular risk
12.30	Questions from students on morning lectures
13.00	Lunch
Afternoon Session	
14.30	Concurrent workshops
16.00	Coffee break
16.30	Concurrent workshops

Tuesday, March 25, 2003
MOLECULAR GENETICS

Morning Session

8.30	S.E. Antonarakis (Geneva, CH) Trisomy 21: understanding its molecular pathophysiology
9.20	T. Meitinger (Munich, D) The genetics of blindness
10.10	Coffee break
10.30	P. Byers (Seattle, USA) Medical genetics of the human collagen genome
11.20	S. Malas (Nicosia, CY) Stem cells in therapeutic medicine
12.30	Questions from students on morning lectures
13.00	Lunch

Afternoon Session

14.30	Concurrent workshops
16.00	Coffee break
16.30	Concurrent workshops

Wednesday, March 26, 2003
CLINICAL GENETICS

Morning Session

- 8.30 **M.P. Cosma** (Naples, I)
Disorders due to deficiency of sulfatases: one gene for 9 diseases
- 9.20 **R. Winter** (London, UK)
Aids for diagnosis in dysmorphology
- 10.10 Coffee break
- 10.30 **A. Schinzel** (Zurich, CH)
Clinical findings in chromosome aberrations
- 11.20 **J. Hoover-Fong** (Baltimore, USA)
An illustrated Glossary of Clinical Genetics
- 12.15 Lunch

Afternoon

- 13.45 Sightseeing tour (destination proposed: Biblioteca Malatestiana-Forlì)

Thursday, March 27, 2003
GENOME PROJECTS

Morning Session

- 8.30 **A. Chakravarti** (Baltimore, USA)
(to be announced)
- 9.20 **A. Morabia**
Genomics of metabolic pathways
- 10.10 Coffee break
- 10.30 **N. Katsanis** (Baltimore, USA)
(to be announced)
- 11.20 **S. Banfi** (Naples, I)
Eurexpress: A European project for large scale gene expression analysis by RNA in situ hybridization
- 12.30 Questions from students on morning lectures
- 13.00 Lunch

Afternoon Session

- 14.30 Concurrent workshops
- 16.00 Coffee break
- 16.30 Concurrent workshops

Friday, March 28, 2003
NEUROGENETICS

Morning Session

8.30	J.-L. Mandel (Strasbourg, F) Fragile X and other causes of X linked mental retardation: Investigation on pathological mechanisms and diagnostic issues
9.20	S. Malas (Nicosia, CY) The use of transgenic mice in disease modelling: Lessons for epilepsy in humans
10.10	Coffee break
10.30	A. Aguzzi (Zurich, CH) Prion Diseases
11.20	E. Lugaresi, P. Montagna (Bologna, I) Fatal familial insomnia: in-depth study of the phenotype underpins the interpretation of molecular genetic data
12.30	Questions from students on morning lectures
13.00	Lunch

Afternoon Session

14.30	Concurrent workshops
16.00	Coffee break
16.30	Concurrent workshops

Saturday, March 29, 2003

FUNCTIONAL GENOMICS

Morning Session

8.30	K. Rhoden (New Haven, USA) Introduction to pharmacogenomics
9.20	J.L.V. Galletta (Genoa, I) High-throughput screening for the identification of new drugs for cystic fibrosis
10.10	Coffee break
10.30	S. Volinia (Ferrara, I) Analysis of microarray data for expression profiling
11.20	A. Pandey (Baltimore, USA) Use of proteomics for functional annotation of genomes
12.30	Questions from students on morning lectures
13.00	Lunch

Afternoon Session

14.30	Concurrent workshops
16.00	Coffee break
16.30	Concurrent workshops

Registration fee for the entire course (7 days, 8 nights): € 1.300,00

Course fee include tuition, course material, room and board for the whole duration of the course. Arrival in Bertinoro is scheduled on the day before the course. Departure is scheduled on March 30 .

Registrants who are admitted to the course will be required to stay for the whole duration. A limited number of fellowships is available from the Euroconferences program of the European Commission; for details and conditions, visit www.eurogene.org. Register online at www.eurogene.org. Offline registration is not available. All course is held in English, unless otherwise stated. For further information about the University Residential Centre, visit www.centrocongressibertinoro.it.

For further information visit the EGF website: [//www.eurogene.org](http://www.eurogene.org)
