EUROPEAN SCHOOL OF GENETIC MEDICINE-ESHG



2nd Course in Eye Genetics September 28th - October 1st, 2011

Location

EuroMediterranean University Centre of Ronzano, Bologna (Italy)

Directors

R. Allikmets (Columbia University, New York), A. Ciardella (U.O. Oftalmologia, Bologna), M. Seri (U.O. Genetica Medica, Bologna).

Course Description

Eye Genetics is a 4-day long postgraduate level course addressed to both researchers and clinicians seeking an up-to-date introduction to the field of ophthalmogenetics today. It provides an overview of the clinical developments of modern genetics in different fields of ophthalmology. The topics covered in the course are: hereditary retinal diseases, genetics of retinitis pigmentosa, genetics of age related macular degeneration, genetics of myopia, genetics of glaucoma, genetics of corneal pathology, genetics of optic nerve diseases, gene therapy.

Speakers

R. Allikmets (New York, Usa), A. Auricchio (Naples, Italy), S. Banfi (Naples, Italy), P. Barboni (Bologna, Italy) W. Berger (Zürich, Switzerland), P. Bonneau (Paris, France), A. Ciardella (Bologna, Italy), F. Cordeiro (London, Uk), F. Cremers (Nijmegen, The Netherlands), A. Gal (Hamburg, Germany), C. Inglehearn (Leeds, UK), N. Katsanis (Baltimore, USA), P. Yu Wai Man (Newcastle upon Tyne, UK), B. Leroy (Ghent, Belgium), T. Moore (London, Uk), K. Neveling (Nijmegen, the Netherlands), M. Seri (Bologna, Italy), A. Sodi (Florence, Italy), B. Veronica Van Heyningen (Edinburgh, UK), J. Sowden (London, UK)

For updates, programme and registration please visit the EGF website <u>www.eurogene.org</u> CONTACTS: Tel. +39.051. 2088414 Fax +39.051.2088424 <u>serena.paterlini@eurogene.org</u>









Registration Fee:

650 €*(tuition, course material, lunches, coffee breaks and shuttle bus service).

* VAT is excluded (Value Added Tax=20%)



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Sentember 28 th Sentember 20 th 900 - 915 Victome 900 - 915 Victome 915 - 10:00 Introduction to EGF and Bologna (G. Romeo) 900 - 9:40 IBD mapping in consanguineous and non-consanguineous families: finding refinal disease genes (F. Granetas and A. Sodi) 1) Overview of clinical ophihalmology for basic scientists (A. Leroy) 9:20 - 10:20 Gene thersp for Leber Congenital Anaurosis (A. Auricchio) 1) Overview of Basic medical genetics for ophihalmologists (A. Leroy) 10:20 - 11:00 Gene thersp for Leber Congenital Anaurosis (A. Auricchio) 1) Status Order Berak 11:00 - 11:30 Discussion 11:30 - 12:00 Coffee Break 11:200 - 12:40 Size Method 11:200 - 12:40 Size Method 1:320 - 13:30 Discussion 11:30 - 11:30 Discussion 11:30 - 11:30 Discussion 1:320 - 13:30 Discussion 11:30 - 13:30 Discussion 11:30 - 11:30 Discussion 1:320 - 13:30 Discussion 11:30 - 13:30 Discussion 11:30 - 13:30 Discussion 1:320 - 13:30 Discussion 11:30 - 16:30 Discussion 11:30 - 16:30 Discussion 1:30 - 14:30 Lunch Afternoon Session : Concurrent Workshops 14:30 - 16:00 1) Preparation: Student discussion group on interesting cases 1:00 - 10:00 Guided Tour of Bologna 1) Deparation: Student discussion group on interesting cases 1) Oreparation: Student discussion group on interesting cases <		
9:00 - 9:15 Welcome 9:00 - 9:40 IBD mapping in consanguineous and non-consanguineous families: finding retinal disease genes (F. Cremers) 9:10 - 10:00 - 10:02 Uakis in parallel: 9:00 - 9:40 IBD mapping in consanguineous and non-consanguineous families: finding retinal disease genes (F. Cremers) 10 overview of basic medical genetics for ophthalmologists (A. Leroy) 10:20 11:200 Gene therapy for Leber Coording RNAs in cyc development and function (S. Banf) 11:30 - 12:30 Coffee Break 11:30 11:30 Discussion 11:30 - 12:30 Discussion 11:30 - 11:30 Discussion 13:30 - 14:30 Lunch Afternoon Session : Concurrent Workshops 14:30-16:00 Afternoon Session : Concurrent Workshops 14:30-16:00 1) Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (T. Moore) 17:00 - 19:00 Guided Tour of Bologna 1) Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (T. Moore) 17:00 - 19:00 Guided Tour of Bologna 1) Preparation: Student discussion group on retinal disease (V, van Heyningen, N. Katsanis) 13:00 - 14:30 Lunch 9:00 -9:40 Stem cells in cyc diseases (J. Sowden) 9:00 -9:40 Orefte Break 9:00 -9:40 Stem cells in cyc diseases (J. Sowden) 9:40 - 12:00 Coffee Break 9:00 -9:40 Stem cells in cyc diseases (J. Sowden) 9:40 - 12:00 Coffee Break 9:00 -9:40 Stem cells in cyc diseases (J. Sowden)<	September 28 th	September 29 th
September 30 th October 1 st 9:00-9:40 Architecture of genetic disease: causes, modifiers and the concept of genetic load (N. Katsanis) 9:40-10:20 Genetics of congenital cataract (T. Moore) 10:20-11:00 Overview of developmental eye anomalies (V. van Heyningen) 11:00-11:30 Discussion 11:30-12:00 Coffee Break 12:00-12:40 Genetics of AMD (R. Allikmets) 12:00-12:40 Genetics of AMD (R. Allikmets) 13:20-13:30 Discussion 13:20-13:30 Discussion 13:20-14:30 Lunch9:00-9:40 Stem cells in eye diseases (J. Sowden) 9:40-10:20 Genetics of glaucoma and myopia (C. Inglehearn) 10:20-11:00 Norrin and retinal blood vessel development (EVR, ROP, Norrie disease) (W. Berger) 11:30-12:00 Coffee Break 12:00-13:30 2 Concurrent workshops: 1) Clinical approach to hereditary retinal diseases (A. Ciardella, M. Seri, C. Graziano, A. Sodi) 2) Genomics: technological developments and interpretation of results; the impact of next generation sequencing on disease gene identification (F. Cremers and his team: Neveling and Inglehearn). 13:30-14:30 Lunch 14:30 Infinitum "Meet the faculty" and Summary of the Course Careers in science (clinical and molecular genetics): one shoe dare are field (N. Katsanis and olf femilter)	 9:00 - 9:15 Welcome 9:15 - 10:00 Introduction to EGF and Bologna (G. Romeo) 10:00 - 10:50 2 talks in parallel: Overview of clinical ophthalmology for basic scientists (A. Ciardella and A. Sodi) Overview of basic medical genetics for ophthalmologists (A. Leroy) 10:50-11.35 Genetics of cone dystrophies/dysfunction syndromes (T. Moore) 11:35-12:00 Coffee Break 12:00-12:40 Molecular basis of non-syndromic and syndromic retinal and vitreoretinal diseases (W. Berger) 12:40-13.20 Introduction to next-generation sequencing for eye diseases (K. Neveling) 13:20-13.30 Discussion 13:30-14:30 Lunch Afternoon Session : Concurrent Workshops 14:30-16:00 Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (T. Moore and B. Leroy) Disease-causing mutations: finding, interpretation, nomenclature (W. Berger, R. Allikmets) Model organisms to study eye biology and disease (V. van Heyningen, N. Katsanis) 17:00-19:00 Guided Tour of Bologna 	 9:00- 9:40 IBD mapping in consanguineous and non-consanguineous families: finding retinal disease genes (F. Cremers) 9:40-10:20 Genetics of RP/LCA/CSNB (B. Leroy) 10:20-11:00 Gene therapy for Leber Congenital Amaurosis (A. Auricchio) 11:00-11:30 Discussion 11:30-12:00 Coffee Break 12:00-12:40 The role of non-coding RNAs in eye development and function (S. Banfi) 12.40: 13.20 Retinal ciliopathies: diverse phenotypes with overlapping genetic structure (N. Katsanis) 13:20-13.30 Discussion 13:30-14:30 Lunch Afternoon Session : Concurrent Workshops 14:30-18:00 1) Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (T. Moore & B. Leroy). 2) Model organisms to study eye biology and disease (V. van Heyningen, N. Katsanis). 3) Genomics: technological developments and interpretation of results; the impact of next generation sequencing on retinal disease gene identification (F. Cremers and his team: Neveling and Inglehearn). 4) Clinical approach to hereditary retinal diseases (A. Ciardella, M. Seri, C. Graziano, A. Sodi) 5) Disease-causing mutations: finding, interpretation, nomenclature (W. Berger, R. Allikmets)
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Carelli, P. Bonneau, F. Cordeiro, P. Yu Wai Man, B. Leroy)

Conclusions Departure