



European School of Genetic Medicine

5th Course in Eye Genetics Bertinoro, Italy, September 24-26, 2017

Bertinoro University Residential Centre
Via Frangipane, 6 – Bertinoro
www.ceub.it

Target Audience: The Eye Genetics Course is a 3-day long postgraduate level course aimed at both clinicians and researchers seeking an up-to-date introduction to, as well as an overview of ophthalmic genetics today. The course will cater for an audience including ophthalmologists, clinical and molecular geneticists, and genetic counselors. Topics covered include genetics of inherited retinal diseases, age-related macular degeneration, myopia, glaucoma, corneal disease, optic neuropathies, ocular developmental disease, and gene therapy. The faculty combines experts from all fields of ophthalmic genetics known for their didactic skills. Participants are encouraged to present a clinical or genetic case of eye disease and will be coached before presenting. Prizes will be awarded for three best presentations.

Fees: The Registration fee for the course (€ 610,00) includes tuition, course material, meals, coffee breaks, social dinner, and transportation from Bologna airport to the course venue on Sept. 23rd and back on September 27th.

Accommodation: You'll be lodged at the Course Venue (or hotels nearby the course venue if necessary). The rate for a double room occupancy (to be shared with another participant) is €35,00/night and includes bed and breakfast.

A limited number of fellowships covering both registration fees and accommodation will be sponsored by the European Society of Human Genetics (ESHG). Following the guidelines established by ESHG, students from economically less favoured countries are strongly encouraged to apply for ESHG fellowships: https://www.eshg.org/fileadmin/eshg/countries/Collective_Members_Country_List_from_01_01_2017.pdf However fellowships are not limited to these countries.

Applicants for fellowships should submit their request together with CV and a reference letter to rpartisan@ceub.it. Closing date for fellowship applications: July 12th, 2017. A communication of acceptance (or non acceptance) will be sent by mail within July 21st

Applicants without fellowships can register at the following link: www.ceub.it
Closing date for registration will be when all places are taken.

Directors:

Rando Allikmets (Columbia University, New York, USA) Antonio Ciardella (U.O. Oftalmologia, Policlinico Sant'Orsola, Bologna, Italy) Bart P. Leroy (Ghent University, Ghent, Belgium) Marco Seri (U.O Genetica Medica, Bologna, Italy).

Faculty:

Alberto Auricchio (Telethon Institute of Genetics and Medicine, Naples, Italy), Sandro Banfi (Telethon Institute of Genetics and Medicine, Naples, Italy), Piero Barboni (Studio D'Azeglio, Bologna, Italy), Wolfgang Berger (Institute of Medical Genetics, Univ. of Zurich, Zurich, Switzerland), Graeme Black (Manchester Academic Health Science Centre, Manchester, UK), Valerio Carelli (Lab. Of Neurogenetics, Univ. of Bologna, Bologna, Italy), Frans P.M. Cremers (Radboud Univ. Medical Centre, Nijmegen, The Netherlands), Claudio Graziano (Dept. of Medical Genetics, Sant'Orsola Malpighi Hospital, Bologna, Italy), Susanne Roosing (Dept. of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands), Georgina Hall (Manchester Children's Foundation Trust, Manchester, UK), Nicholas Katsanis (Dept. of Cell Biology & Centre of Human Disease Modeling, Duke Univ. School of Medicine, Durham, USA), Andrea Sodi (Dept. of Ophthalmology, Univ. of Florence, Italy), Jane C Sowden. (UCL Inst. of Child Health, Univ. College London, UK)

COURSE PROGRAM

Arrival day: Saturday, September 23rd

September 24

8:30 - 8:45 **Welcome**
Giovanni Romeo and Rando Allikmets

8:45 - 9:30 **2 parallel talks: (40 min + 5 min discussion)**

Garrison Room

1. Overview of clinical ophthalmology for basic scientists
Antonio Ciardella

Jacopo da Bertinoro Room

2. Overview of basic medical genetics for ophthalmologists
Bart Leroy

9:35 - 11:05 **2 talks (40 min + 5 min discussion)**

3. Stargardt disease, the complex simple retinal disorder
Rando Allikmets

4. Genetics of corneal diseases
Graeme Black

11:05 - 11:30 **Break**

11:30 - 13:00 **2 talks (40 min + 5 min discussion)**

1. Molecular basis of non-syndromic and syndromic retinal and vitreoretinal diseases
Wolfgang Berger

2. Introduction to next-generation sequencing for eye diseases
Susanne Roosing

13:00 - 14:00 **Lunch**

14:00 - 16:00 **3 parallel workshops**

Garrison room

WS1 Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (**Graeme Black & Bart Leroy**)

Jacopo da Bertinoro room

WS4 Genetic counseling (**Georgina Hall & Marco Seri**)

Computer room

WS5 Genomics: technological developments and interpretation of results; the impact of next generation sequencing on retinal disease gene identification (**Frans Cremers & Susanne Roosing**)

16:00 - 16:30 **Break**

16:30 - 18:30 **3 parallel workshops**

Garrison Room

WS1 Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (**Graeme Black & Bart Leroy**)

Jacopo da Bertinoro room

WS2 Clinical approach to hereditary retinal diseases (**Antonio Ciardella, Claudio Graziano, Andrea Sodi**)

Computer room

WS3 Disease-causing mutations: finding and interpretation (**Wolfgang Berger & Rando Allikmets**)

September 25

9:00 - 11:15 **3 talks (40 min + 5 min discussion)**

1. Genetics of RP/LCA/CSNB
Bart Leroy
2. Stem cells in eye diseases
Jane Sowden
3. Genetics of age-related macular degeneration
Rando Allikmets

11:15 - 11:45 **Break**

11:45 - 13:15 **2 talks (40 min + 5 min discussion)**

4. Overview of developmental eye anomalies
Graeme Black
5. Retinal ciliopathies: diverse phenotypes with overlapping genetic structure
Nicholas Katsanis

13:15 - 14:15 **Lunch**

14:15 - 16:15 **3 parallel workshops**

Jacopo da Bertinoro Room

WS2 Clinical approach to hereditary retinal diseases (**Antonio Ciardella, Claudio Graziano, Andrea Sodi**)

Garrison Room

WS4 Genetic counseling (**Georgina Hall & Marco Seri**)

Computer room

WS3 Disease-causing mutations: finding and interpretation (**Wolfgang Berger & Rando Allikmets**)

16:15 - 16:45 **Break**

16:45 - 18:45 **2 parallel workshops**

Jacopo da Bertinoro Room

WS1 Final preparation for student presentations and selection of 10-12 cases for presentation (Graeme **Black & Bart Leroy**)

Computer room

WS5 Genomics: technological developments and interpretation of results; the impact of next generation sequencing on retinal disease gene identification (**Frans Cremers & Susanne Roosing**)

September 26

9:00 - 11:15 **3 talks (40 min + 5 min discussion)**

1. Architecture of genetic disease: causes, modifiers and the concept of genetic load
Nicholas Katsanis
2. Genetics of glaucoma
Jane Sowden
3. Gene therapy for recessive and dominant eye disorders
Alberto Auricchio

11:15 - 11:45 **Break**

11:45 - 13:15 **2 talks (40 min + 5 min discussion)**

4. The role for non-coding RNAs in eye development, function and diseases
Sandro Banfi
5. Modifier genes in retinal diseases
Frans Cremers

13:15 - 14:15 **Lunch**

14:15 - 15:45 **Student presentations**

15:45 - 16:15 **Break**

16:15 - 17:45 **3 shorter talks (25 min +5 min discussion)**

6. Genetics of mitochondrial diseases and retinopathies
Bart Leroy
7. Mitochondrial optic neuropathies
Piero Barboni
8. The paradigm of mitochondrial optic neuropathies: naturally occurring compensatory strategies and treatment options
Valerio Carelli

18:00 - 19:00 **Feedback on student presentations, awards presentation, summary of the course**