



7th EYE GENETICS COURSE
Bertinoro di Romagna, Italy
25-27 September 2022

Course Director

Rando ALLIKMETS, PhD, Columbia University, New York, NY, USA

Course Co-directors

Graeme CM BLACK, OBE, DPhil, FRCOphth, FMedSci, Manchester University, Manchester, UK
Antonio CIARDELLA, MD, University of Bologna, Bologna, Italy
Bart P LEROY, MD, PhD, Ghent University, Ghent, Belgium
Marco SERI, MD, University of Bologna, Bologna, Italy

Local Event Organiser

Roberta PARTISANI, CeUB

Course Location & Contact Information

Centro Residenziale Universitario di Bertinoro (CeUB)
Via Frangipane 6, 47032 Bertinoro (FC), Italy
tel. +39 0543-446500 – fax +39 0543-446599
rpartisani@ceub.it

Additional Information

The Eye Genetics Course consists of teaching sessions and workshops. Teaching sessions are held in the main room, which is the Garrison Room. Workshops are also held in several other rooms. Please check where you need to be according to the program. All workshops are organized in parallel as indicated in the program. Please enlist your name on boards provided near the secretariat for those workshops you would like to participate in.

All participants are encouraged to participate in the Case Presentation Concours. For those interested, Workshop 1 provides coaching. The Concours is held on the last day of the Course.

On Saturday 24 September 2022, an evening welcome dinner will be served.



Course Program

Sunday 25 September 2022

MORNING SESSIONS

8:30 - 8:45hrs **Welcome & Overview of the Course**
Rando ALLIKMETS

TEACHING SESSION I: INTRODUCTION TO OPHTHALMIC GENETICS

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

Garrison Room

1/ Overview of Clinical Ophthalmology for Basic Scientists
Antonio CIARDELLA, Bologna, Italy

Jacopo da Bertinoro Room

2/ Overview of Basic Medical Genetics for Ophthalmologists
Bart P LEROY, Ghent, Belgium

TEACHING SESSION II: MOLECULAR OPHTHALMIC GENETICS

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

3/ Molecular Screening Technologies
Frans PM CREMERS, Nijmegen, The Netherlands

4/ Variant Classification & Molecular Basis of Disease
Frans PM CREMERS, Nijmegen, The Netherlands

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

TEACHING SESSION III: OCULAR DEVELOPMENTAL ANOMALIES

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

5/ Anterior Segment Dysgenesis & Cataract
Graeme CM BLACK, Manchester, UK

6/ Microphthalmia, Anophthalmia & Coloboma (MAC) Spectrum of Diseases
Graeme CM BLACK, Manchester, UK

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

AFTERNOON SESSIONS

WORKSHOP SESSION I

14:00 - 16:00hrs 3 parallel workshops

Garrison Room

WS1 Preparation: Student Presentation & Discussion on Interesting Cases (clinical, molecular, families, etc.) They Have Encountered (*Graeme CM BLACK & Bart P LEROY*)

Jacopo da Bertinoro Room

WS2 Genetic Counseling (*Georgina HALL & Marco SERI*)

Computer Room

WS3 Genomics: Technological Developments & Interpretation of Results: Impact of High-throughput Technology on Retinal Disease Gene Identification (*Frans PM CREMERS, Daan PANNEMAN & Zelia CORRADI*)

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

WORKSHOP SESSION II

16:30 - 18:30hrs 3 parallel workshops

Garrison Room

WS1 Preparation: Student Presentation & Discussion on Interesting Cases (clinical, molecular, families, etc.) They Have Encountered (*Graeme CM BLACK & Bart P LEROY*)

Jacopo da Bertinoro Room

WS4 Clinical Approach to Inherited Retinal Diseases (*Antonio CIARDELLA, Claudio GRAZIANO, Francesco TESTA*)

Monday 26 September 2022

MORNING SESSIONS

TEACHING SESSION IV: INHERITED RETINAL DISEASES

8:15 - 10:30hrs 3 talks (40 min + 5 min discussion)

7/ Generalised Inherited Retinal Diseases

Bart P LEROY, Ghent, Belgium

8/ Vitreoretinopathies

Graeme CM BLACK, Manchester, UK

9/ Inherited Maculopathies

Bart P LEROY, Ghent, Belgium

10:30 - 11:00hrs **Break**

TEACHING SESSION V: GENOTYPE & PHENOTYPE INTEGRATION

11:00 - 13:15hrs 3 talks (40 min + 5 min discussion)

10/ Stargardt Disease, the Complex Simple Retinal Disorder

Rando ALLIKMETS, New York, NY, USA

11/ Role of Phenotyping in Variant Interpretation

Winston LEE, New York, NY, USA

12/ Beyond Classic Mechanisms of Disease (virtual presentation)

Nicholas KATSANIS, Miami, FL, USA

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

AFTERNOON SESSIONS

WORKSHOP SESSION III

14:15 - 16:15hrs 3 parallel workshops

Garrison Room

WS2 Genetic Counseling (*Georgina HALL & Marco SERI*)

Jacopo da Bertinoro Room

WS4 Clinical Approach to Inherited Retinal Diseases (*Antonio CIARDELLA, Claudio GRAZIANO, Francesco TESTA*)

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

WORKSHOP SESSION IV

16:45 - 18:45hrs 2 parallel workshops

Jacopo da Bertinoro Room

WS1 Final Preparation for Student Presentations & Selection of 10-12 Presentations for Concours (*Graeme CM BLACK & Bart P LEROY*)

Computer Room

WS3 Genomics: Technological Developments & Interpretation of Results: Impact of High-throughput Technology on Retinal Disease Gene Identification (*Frans PM CREMERS, Daan PANNEMAN & Zelia CORRADI*)

Tuesday 27 September 2022

MORNING SESSIONS

TEACHING SESSION VI: THERAPEUTIC STRATEGIES FOR INHERITED RETINOPATHIES

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

13/ Cell Therapy

Michael CHEETHAM, London, UK

14/ Gene Augmentation Therapy for Autosomal Recessive & Dominant Retinopathies

Ivana TRAPPANI, Napoli, Italy

15/ CRISPR/Cas9 & Antisense Oligonucleotides

Michael CHEETHAM, London, UK

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

TEACHING SESSION VII: COMPLEX EYE GENETICS

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

16/ Genetics of Age-related Macular Degeneration

Rando ALLIKMETS, New York, NY, USA

17/ “Meet the faculty”

Faculty discusses with students all question they may have and provides career guidance

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

AFTERNOON SESSIONS

TEACHING SESSION VII: STUDENT PRESENTATIONS

14:15 - 15:45hrs **10-12 student presentations**

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

TEACHING SESSION VIII: MITOCHONDRIAL EYE GENETICS

16:15 - 17:45hrs 2 talks (40 min +5 min discussion)

18/ Mitochondrial Genetics & Retinopathies

Bart P LEROY, Ghent, Belgium

19/ Inherited Optic Neuropathies & Treatment Strategies

Valerio CARELLI, Bologna, Italy

FINAL SESSION IX: FEEDBACK & AWARDS

18:00 - 19:00hrs

20/ Feedback on Student Presentations, Awards Presentation, Summary of the Course & Farewell

Rando ALLIKMETS, New York, NY, USA

With the contribution of

