



# European School of Genetic Medicine

## 4<sup>th</sup> Course in Eye Genetics

**Bertinoro, Italy, September 27-29, 2015**

Bertinoro University Residential Centre  
Via Frangipane, 6 – Bertinoro  
[www.ceub.it](http://www.ceub.it)

### Course Directors:

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**4<sup>TH</sup> COURSE IN EYE GENETICS**  
**Bertinoro University Residential Centre**  
**Bertinoro, Italy, September 27-29, 2015**

**Arrival day: Saturday, September 26<sup>th</sup>**

**September 27**

8:30 - 8:40     **Welcome**

8:40 - 9:10     History of the Medical Genetics and Eye Genetics courses in Bologna and Bertinoro  
**Giovanni Romeo**

9:15 - 10:00    **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**

10:05 - 11:35   **2 talks (40 min + 5 min discussion)**

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

4. Genetics of corneal diseases  
**Graeme Black**

11:35 - 12:00   **Break**

12:00 - 13:30   **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  
**Lonneke Haer-Wigman**

13:30 - 14:30   **Lunch**

14:30 - 16:15   **3 parallel workshops**

**Garrison room**

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

**Jacopo da Bertinoro room**

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

**Computer room**

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

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

16:45 - 18:30 **3 parallel workshops**

**Garrison Room**

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

**Jacopo da Bertinoro room**

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

**Computer room**

**WS3** Disease-causing mutations: finding, interpretation, nomenclature (**Berger & Allikmets**)

**September 28**

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 AMD  
**Rando Allikmets**

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

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

4. Gene therapy for recessive and dominant eye diseases  
**Alberto Auricchio**
5. Retinal ciliopathies: diverse phenotypes with overlapping genetic structure  
**Nicholas Katsanis**

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

14:15 - 16:00 **3 parallel workshops**

**Jacopo da Bertinoro Room**

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

**Garrison Room**

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

**Computer room**

**WS3** Disease-causing mutations: finding, interpretation, nomenclature (**Berger & Allikmets**)

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

16:30 - 18:15 **2 parallel workshops**

**Jacopo da Bertinoro Room**

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

**Computer room**

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

**September 29**

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. Overview of developmental eye anomalies  
**Graeme Black**

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**