

# COURSE PROGRAM

## September 22

8:30 - 8:45     **Welcome**  
                  **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 (**Susanne Roosing & assistant**)

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 23**

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  
**Mike Cheetham**
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 (**Susanne Roosing & assistant**)

**September 24**

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  
**C. Gustavo De Moraes**
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. CRISPR/Cas9 technology and applications in vision research  
**Mike Cheetham**

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**