Course in Eye Genetics

October 13-15, 2013

University Residential Center of Bertinoro (Bertinoro di Romagna, Italy)

October 13

9:00 - 9:10 **Welcome**

9:10 - 9:55 2 parallel talks: (40 min + 5 min discussion)

Lecture Hall

1. Overview of clinical ophthalmology for basic scientists
   Antonio Ciardella

Meeting Room

2. Overview of basic medical genetics for ophthalmologists
   Bart Leroy

10:00 - 11:30 2 talks (40 min + 5 min discussion)

3. Genetics of glaucoma
   Jane Sowden

4. IBD mapping in consanguineous and non-consanguineous families: finding retinal disease genes
   Frans Cremers

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
   Kornelia Neveling

13:30-14:30 **Lunch**

14:30-16:15 3 parallel workshops

Lecture hall

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

Meeting 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 & Neveling**)

16:15-16:45 break

16:45-18:30 3 parallel workshops

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

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

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

October 14

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 corneal diseases
   Graeme Black

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
   Enrico Surace

5. Retinal ciliopathies: diverse phenotypes with overlapping genetic structure
   Nicholas Katsanis

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

14:15-16:00 3 parallel workshops

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

Meeting 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

Lecture hall
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 & Neveling**)

October 15

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 AMD
   Rando Allikmets

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. Treatment options for mitochondrial eye disease
   Valerio Carelli

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