

## **September 28<sup>th</sup>**

### **Morning Session:**

9:00 - 9:15 **Welcome**

9:15 - 10:00 Introduction to EGF and Bologna (**G. Romeo**)

10:00 - 10:50 2 talks in parallel: (45 min + 5min discussion)

### **Lecture Hall:**

1) Overview of clinical ophthalmology for basic scientists (**A. Ciardella and A. Sodi**)

### **Workshop Room:**

2) Overview of basic medical genetics for ophthalmologists (**B. Leroy**)

10:50-11.35 (40 min + 5 discussion) Genetics of cone dystrophies/dysfunction syndromes (**T. Moore**)

11:35-12:00 **Coffee Break**

(40 min talk + 5 discussion)

12:00-12:40 Molecular basis of non-syndromic and syndromic retinal and vitreoretinal diseases (**W. Berger**)

12:40-13.30 Introduction to next-generation sequencing for eye diseases (**K. Neveling**)

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

### **Afternoon Session : Concurrent Workshops 14:30-16:00**

### **Lecture Hall:**

1) Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (**T. Moore and B. Leroy**)

### **PC Lab:**

2) Disease-causing mutations: finding, interpretation, nomenclature (**W. Berger, R. Allikmets**)

### **Workshop Room:**

3) Model organisms to study eye biology and disease (**V. van Heyningen, N. Katsanis**)

17:00-19:00 **Guided Tour of Bologna**

## September 29<sup>th</sup>

### **Morning Session:**

9:00- 9:40 IBD mapping in consanguineous and non-consanguineous families: finding retinal disease genes (**F. Cremers**)

9:40- 10:20 Genetics of RP/LCA/CSNB (**B. Leroy**)

10:20- 11:15 Gene therapy for Leber Congenital Amaurosis (**A. Auricchio**)

11:15-11.45 **Coffee Break**

11.45-13:15 ( **2 talks 40 min 5 discussion**)

11:45-12:30 The role of non-coding RNAs in eye development and function (**S. Banfi**)

12.30: 13.15 Retinal ciliopathies: diverse phenotypes with overlapping genetic structure (**N. Katsanis**)

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

### **Afternoon Session : 5 Concurrent Workshops 14:30-18:00**

**14:30-16:00**

#### **Lecture Hall:**

1) Preparation: Student discussion group on interesting cases (clinical, molecular, families, etc.) they have encountered (**T. Moore & B. Leroy**).

#### **Workshop Room:**

2) Model organisms to study eye biology and disease (**V. van Heyningen, N. Katsanis**).

#### **Pc Lab:**

3) Genomics: technological developments and interpretation of results; the impact of next generation sequencing on retinal disease gene identification (**F. Cremers** and his team: **Neveling** and **Inglehearn**).

16.00-16.30 **Coffee Break**

#### **Lecture Hall:**

4) Clinical approach to hereditary retinal diseases (**A. Ciardella, M. Seri, C. Graziano, A. Sodi**)

#### **Pc Room:**

5) Disease-causing mutations: finding, interpretation, nomenclature (**W. Berger, R. Allikmets**)

## **September 30<sup>th</sup>**

9.00-11.15 (40 min + 5 min discussion)

9:00-9:40 Architecture of genetic disease: causes, modifiers and the concept of genetic load ( **N. Katsanis**)

9:40- 10:20 Genetics of congenital cataract ( **T. Moore**)

10:20- 11:15 Overview of developmental eye anomalies ( **V. van Heyningen**)

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

11:45-13:15 ( 40 minut talk + 5 min discussion)

11:45-12:35 Genetics of AMD ( **R. Allikmets**)

12:35-13:15 Modifier genes in retinal diseases ( **F. Cremers**)

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

**14. 15- 16.00 Lecture Hall**

**Student presentations**

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

**Lecture Hall:**

16:30-18:30 Mitochondrial eye diseases ( **P. Barboni, V. Carelli, P. Bonneau, F. Cordeiro, P. Yu Wai Man, B. Leroy**)

## **October 1<sup>st</sup>**

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

9:00-9:40 Stem cells in eye diseases ( **J. Sowden**)

9:40-10:25 Genetics of glaucoma and myopia ( **C. Inglehearn**)

10:25-11:15 Norrin and retinal blood vessel development (EVR, ROP, Norrie disease) ( **W. Berger**)

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

11:45-13:30 2 Concurrent workshops:

**Lecture Hall:**

1) Clinical approach to hereditary retinal diseases (**A. Ciardella, M. Seri, C. Graziano, A. Sodi**)

**Pc Room:**

2) Genomics: technological developments and interpretation of results; the impact of next generation sequencing on disease gene identification (**F. Cremers** and his team: **Neveling** and **Inglehearn**).

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

14:30 Infinitum “Meet the faculty” and Summary of the Course

Careers in science (clinical and molecular genetics): one shoe does not fit all. (**N. Katsanis and all faculty**)

**Conclusions**

**Departure**