

MASTER

CLINICAL GENETICS

Multiple Congenital Anomalies (MCA)

Year 2007-2008

Promoted by Società Italiana di Genetica Umana - SIGU

Endorsed by European Society of Human Genetics - ESHG

This is the second edition of an inter-University Master aimed at training MD graduate students in Clinical Genetics.

It is recommended to those who already have a Specialty or a PhD degree in Medical Genetics, or those who have been trained in Medical Genetics for at least two years.

This year, the focus of the Master is on Multiple Congenital Anomalies (MCA).

It has been realized on the basis of a special agreement between the following 5 Universities:

- University of Bologna – Medical Genetics – Prof. Giovanni Romeo
- University of Genova – Medical Genetics – Prof. Roberto Ravazzolo
- University of Padova – Medical Genetics – Prof. Romano Tenconi
- University of Rome – La Sapienza – Medical Genetics – Prof. Bruno Dallapiccola
- University of Siena – Medical Genetics – Prof. Alessandra Renieri

In addition, a special agreement has been signed with the following institutes that will host students for clinical practice:

- Azienda Ospedaliera Galliera, Genova – Medical Genetics – Dott. Franca Dagna Bricarelli
- Fondazione Ospedale Maggiore Policlinico Mangiagalli e Regina Elena, Milano – Medical Genetics – Dott. Faustina Lalatta

The following Italian institutes provide additional teachers:

- Azienda Ospedaliera Bianchi-Melacrino-Morelli, Reggio Calabria – Medical Genetics – Dott. Manuela Priolo
- Azienda Ospedaliera Clinica Pediatrica, Milano – ambulatorio di Genetica Clinica – Dott. Angelo Selicorni
- OASI Troina – Medical Genetics – Dott. Corrado Romano
- Azienda Ospedaliera G Rummo Benevento – Medical Genetics – Dott. Gioacchino Scarano
- Ospedale G. Gaslini, Genova- Pediatrics – Dott. Maja di Rocco
- Istituto Scientifico Stella Maris, Calmbrone –Pisa –Dott. Agatino Battaglia
- Università Degli Studi di Torino – Struttura Semplice di Genetica, Dip Scienze Pediatriche- Prof.ssa Margherita Silengo
- Azienda Ospedaliera di Reggio Emilia – Struttura Semplice di Genetica Clinica – Dott. Livia Garavelli

The following foreign Universities provide addition teachers:

- Freiburg (D), Centre for Pediatrics and Adolescent Medicine, Dept. of Pediatrics, Freiburg University Hospital, Prof. Andrea Superti-Furga
- Manchester (UK), Medical Genetics and Regional Genetic Service, St Mary's Hospital, University of Manchester, Prof Dian Donnai
- Nijmegen (NL), Dept. Human Genetics, Nijmegen Medical Centre, Radboud University, Prof. Han Brunner
- Taunton (UK), Health Genetics, Faculty of Health and Social Work, University of Plymouth, Prof. Heather Skirton

The overall objective of the program is to educate and train master level candidates to main principles on multiple congenital anomalies and congenital syndromes, their identification, diagnosis and counselling in medical genetics, genotype/phenotype correlations, introduction to techniques of communication in genetic counselling, and functional genomics.

The training program lasts about 1 year from November 2007 to November 2008. Students will move monthly in 6 different training places. In each place, students will receive one main lesson per day (usually in the morning), the remaining time of the morning and the afternoon being spent in clinics (depending on the total number, students may be divided in groups). With this program students will become aware of clinical genetics services present in Italy, useful for future training programs and positions.

Lessons are organized in 6 main areas: Propedeutic, Functional Genomics, Heart and CNS disorders, Skin and Eyes Disorders, Skeletal Disorders, Kidney and Bowel Disorders, Genetic Counselling. One or two teaching unit (week) are assigned to each area. At the end of each teaching unit a common syndrome will be discussed in a manner to recapitulate a basic ABC of the clinical geneticist.

Announcement is published at the University of Siena. Available subscriptions are restricted to a maximum of 5 students. Dead line for application for this second edition of the Master is scheduled the 9th of October 2006.

Every year the focus of the Master will change. Scheduled topic for the next years are:

2008-09 CLINICAL GENETICS: hereditary cancer and tumoral syndromes

2009-10 CLINICAL GENETICS: neurological diseases

Program/schedule of the Master (2007-2008):

Unit 1: Padova December 10-14, 2007

PROPEDEUTIC AREA

Lessons

December 10	09-11 AM	Epidemiology Congenital Anomalies (Clementi M) Definitions and terms of congenital anomalies (Tenconi R)
December 11	09-11 AM	Case History (Scarano G); Databases in Clinical Genetics (Clementi M)
December 12	09-11 AM	Clinical examination (Romano C) Bayes in the risk evaluation (Clementi M)
December 13	09-11 AM	Bayes in differential diagnosis (Tenconi R)
December 14	09-11 AM	Diagnostic flow-chart for Mental Retardation (Romano C)

Clinics

Daily 11.00 AM -06.30 PM.

Student will assist at consults in neonatal intensive care, and in pediatrics department. Scheduled clinics will start at 02.30 PM: mainly MCA-MR patients and 1 or 2 daily genetic counselling for therapeutic abortion. Depending on the total number, students may be divided in groups (3-4 students for each group).

Supervisor: Tenconi R

Credits: 6

Accommodation: ESU 4 Accommodation
Ivan Campagnola
Via Tiepolo 46
Tel. +39 0498941160 fax +39 0497800187
accommodation@esu.pd.it

Unit 2: Siena January 7- 11, 2008

FUNCTIONAL GENOMICS

Lessons

January 7	09-11 AM	Syndrome families: genotypes and phenotypes (Brunner H)
January 8	09-11 AM	Making a clinical diagnosis (Brunner H)
January 9	09-11 AM	Old and new microdeletion/duplication syndromes detected by array-CGH (Renieri A) The 1p36 microdeletion syndrome (Battaglia A)
January 10	09-11 AM	Cleft Lip Palate: clinical and molecular correlation (Priolo M)
January 11	09-11 AM	The syndromes of the month: Mowat-Wilson syndrome (Garavelli L); Cohen syndrome (Renieri A).

Clinics

Daily 11.00 AM -07.00 PM.

Student will assist at consults in internal medicine, ophthalmology and neurosurgery departments.

Scheduled clinics will start at 03.30 PM: mainly MCA-MR patients and 1 or 2 daily cases of patients with Rett syndrome.

Supervisor: Renieri A

Credits: 6

Accommodation: Collegio universitario S. Chiara, Siena tel. 0577 235940
<http://www.unisi.it/santachiara/>

Unit 3: Rome March 5- 9, 2008

HEART AND CNS DISORDERS

Lessons

March 5	09-11 AM	Managing genetic data using SHIRE software (Dallapiccola B)
March 6	09-11 AM	Joubert syndrome and related disorders (Valente M)
March 7	09-11 AM	Congenital heart diseases:clinical features (Marino B)
March 8	09-11 AM	Syndromic congenital heart diseases (Digiglio MC)
March 9	09-11 AM	The syndromes of the month: Williams syndrome (Merla G); Noonan syndrome (Dallapiccola B)

Clinics

Daily 11.00 AM -01.00 PM.

Student will assist at consults in neurology and pediatrics departments.

Scheduled clinics will start at 03.00 PM: mainly patients with cardiomyopathy and counselling for prenatal diagnosis

Supervisor: Dallapiccola B

Credits: 6

Accommodation: Suore Medee Via Eustachio 18 (dietro all'Istituto Mendel)
06/4402716

Albergo S. Giuliana (c/o Senato) 06/68803344
Casa Accoglienza in Piazza di Spagna 06/4745324

Unit 4: Genova

May 5-9, 2008

FUNCTIONAL GENOMICS

Lessons

May 5	9-11 AM	Williams syndrome and the reciprocal duplication syndrome (Donnai D)
May 6	9-11 AM	Manifestation of mosaicism (Donnai D)
May 7	9-11 AM	Clinical cases: goal and pitfall in diagnosis (Lerone M, Divizia MT, Di Rocco M) Learning to integrate clinical data with molecular diagnosis (Ravazzolo R)
May 8	9-11 AM	FMR-1 related disorders (Faravelli F.)
May 9	9-11 AM	The syndrome of the month: Sotos syndrome (Faravelli F.)

Clinics

Daily 11.00 AM -06.00 PM.

Students will be divided in groups of 3 persons. Different groups will be engaged in different weeks of May.

Detailed schedule will be furnished at the time of subscription.

Supervisors: Faravelli F, Bricarelli FD, Ravazzolo R

Credits: 6

Accommodation: Bed & Breakfast, Genova

Unit 5: Bologna

June 9- 13, 2008

KIDNEY AND BOWEL DISORDERS

Lessons

June 9	9-11 AM	Bowel malformations (Seri M)
June 10	9-11 AM	Syndromic and non syndromic Hirschsprung disease (G. Romeo)
June 11	9-11 AM	Sex differentiation and genital anomalies (Silengo M)
June 12	9-11 AM	Renal involvement in syndromic cases (C. Graziano)
June 13	9-11 AM	The syndrome of the month: Turner syndrome (Mazzanti L)

Clinics

Daily 11.00 AM -05.00 PM.

Student will assist at consults in Medical Genetics, Pediatrics and Obstetrics Departments.

Scheduled clinics will start at 02.00 PM: mainly patients with inherited disorders, families with cancer aggregation, MCA patients and counselling for prenatal diagnosis

Supervisor: Seri M

Credits: 6

Accommodation in Bologna:

Bed & Breakfast

Mrs Bergonzoni Marina tel. 051/374349 – 340/2209495

Mrs Meneghetti tel. 051/503636 – 349/4573907

Mrs Foriporta tel. 335/8178175

Mrs Mara tel.348/7471479

ASPPI tel. 051/277148 – 347/8974322

Unit 6: Milano

June 23-27, 2008

GENETIC COUNSELLING

June 23	09-11	The malformed fetus: prenatal genetic counselling (F.Lalatta, G. Zuliani, M. Torricelli,S. Gueneri)
	11-12	Discussion
	14-18	Students will be divided in groups in Obstetrics Department.
June 24	09-11	Genetic counselling in Cistic Fibrosis (L. Spaccini. E.Seia, C. Colombo, Skirton)
	11-12	Discussion
	14-18	Students will be divided in groups “Communication in CF” (F. Lalatta, Skirton) e “Laboratory procedures in CF” (E.Seia, D. Coviello)
June 25	09-11	Genetic counselling in muscle dystrophy (F.Natacci, N. Bresolin, , Skirton)
	11-12	Discussion
	14-18	Students will be divided in groups “Communication in DMD/BMD” (F. Lalatta, Skirton) “Laboratory procedures in DMD/BMD” (S.Tedeschi, D. Coviello)
June 26	09-11	Genetic counselling in hereditary deafness (P.Castorina, U. Ambrosetti , Skirton)
	11-12	Discussion
	14-18	Students will be divided in groups “Communication in hereditary deafness” (F. Lalatta, Skirton) “Laboratory procedures in hereditary deafness” (P. Primignani, D. Coviello)
June 27	09-11	The syndrome of the month: C. De Lange syndrome (Selicorni A)

Supervisor: Lalatta F

Credits: 6

Accommodation: Istituto La Casa, Milano

Unit 7: Siena July 14- 18, 2008

SKELETAL DYSPLASIA

Lessons

July 14	09-11 AM	Skeletal disorders: classification (Superti-Furga A)
July 15	09-11 AM	Letal Skeletal Dysplasia (Superti-Furga A)
July 16	09-11 AM	Skeletal disorders: differential diagnosis by X-Ray (Superti-Furga A)
July 17	09-11 AM	Osteochondrodysplasias (Canepa G)
July 18	09-11 AM	Osteochondrodysplasias (Canepa G)

Clinics

Daily 11.00 AM -07.00 PM.

Student will assist at consults in internal medicine, ophthalmology and neurosurgery departments.

Scheduled clinics will start at 03.30 PM: mainly MCA-MR patients and 1 or 2 daily cases of patients with Rett syndrome.

Supervisor: Renieri A

Credits: 6

Accommodation: Collegio universitario S. Chiara, Siena tel. 0577 235940
<http://www.unisi.it/santachiara/>

Total credits of the Master = 60.

For further information on this Master contact: prioloma@libero.it.