THEORETICAL AND PRACTICAL TRAINING IN

HAEMATOLOGICAL RARE DISEASES:
from genetic counselling - through bench - to bed

CAMPUS OF HEMATOLOGY
Franco e Piera Cutino
A.O.R. "Villa Sofia - V. Cervello"
Palermo (Italy)

15-23 September, 2017
Dear Colleague,

it is our pleasure to invite you to join us for the theoretical and practical training in

**HAEMATOLOGICAL RARE DISEASES: from genetic counselling - through bench - to bed**

which will take place in Palermo at the CAMPUS OF HAEMATOLOGY "Franco e Piera Cutino" A.O.R. "Villa Sofia - V. Cervello" from 15th to 23rd September, 2017.

The main purpose of the Course is to gather experienced scientists in the field of Haematological Rare Diseases at the same table with young Physicians (Haematologists, Gynecologists) and Biologists, Radiologists, Medical Technicians already committed to become experts in this field. The discussion will be on relevant topics related to Prenatal Diagnosis, Thalassemia Syndromes and Haemoglobinopathies, from basic research to clinical and therapeutic dilemmas.

The course will take place at the Campus of Haematology "Franco e Piera Cutino", a pioneering center for prevention, care and research on Thalassemia and Haemoglobinopathies, and for the reception of patients’ families and their guests.

The structure was inaugurated on May 8, 2013 at the Hospital "V. Cervello", A.O.O.R. "Villa Sofia - V. Cervello" of Palermo, under the agreement between the Foundation "Franco e Piera Cutino", the Association for Research "Piera Cutino" Onlus, the U.O.C. Haematology for Rare Diseases of Blood and Haematopoietic Organs and the Health Department of the Regione Sicilia.

The Campus was entirely built thanks to private fundraising by the Association for Research "Piera Cutino", a non-profit organization established in 1998 by the Cutino family.

The Campus of Haematology "Franco e Piera Cutino" stands out as center of excellence across the country, not only for clinical routine tasks but also for the several and exclusive current activities in different clinical and research settings with impact at national and international level.

This is possible thanks to the presence of highly skilled professionals involved in innovative research projects in collaboration with the Foundation "Cutino".

Among the numerous activities carried out at The Campus we are proud to mention

- **Coelocentesis:** exclusive technique of prenatal diagnosis on fetal cells from Coelomic fluid (the only Center in the world).
- **Prenatal diagnosis by Chorionic Villus Sampling and by Amniocentesis.**
- **Tests to detect Thalassemia healthy carrier.**
- **First level screening for β-Thalassemia and Haemoglobinopathies.**
- **Genetic studies for the identification of the CysticFibrosis healthy carrier.**
- **Biobanking for Research "Anna Maria Ferrara Cutino"**
- **Outreach and training activities**
- **Erythroid Cell Culture for the study of HbF drug activation.**
The course includes 15 attendees, Physicians and Researchers selected from different countries highly motivated in becoming experts in the field of Haematological Rare Diseases. Participants will be directly involved in performing the specific practical part of the course in the laboratory or in the clinic unit.

An international stuff of experts will facilitate exchange of opinions and expertise among participants and between participants and tutors.

The duration of the course will be of nine days (15-23 September).

Applicants may wish to attend the entire program divided in three modules or otherwise select one module out of three.

We would like to thank you in advance for your willingness to share your expertise and experiences with us at this important educational activity.

Looking forward to welcoming you to Palermo

Yours sincerely

Prof. Aurelio Maggio - Dr. Santina Acuto
Directors
the course foresees attendance in three modules

**Module 1**

**International Meeting**
1st International Working Group on Thalassemia: is it time to revisit classification of Thalassemia Syndromes?

15-16 September 2017

**Module 2**

**Lectures - Meet the expert**

- Counselling and prenatal diagnosis
- Genotype-phenotype correlations
- Current management of B-thalassemias
- Sickle Cell Disease: past, present and future
- Gene Therapy for Hemoglobinopathies
- Hypercoagulable State on Thalassemia
- Tissue Iron Measurement by MRI: Advances, Challenges, and Pitfalls
- C-licnet Network: potential worldwide MRI network for the management of thalassemia syndromes

17 September 2017

**Module 3**

**TRAINING COURSE in**

- Clinics (Campus of Hematology, F. & P. Cutino)
- Celocentesis, Villocentesis and Amniocentesis (Prenatal Diagnosis Unit)
- Molecular Diagnosys (Labs for diagnosis of rare disease)
- Research (Labs for molecular and cellular biology)

18-23 September 2017
International Meeting
1st International Working Group on Thalassemia: is it time to revisit classification of Thalassemia Syndromes?

Main Topics

- Thalassemia Major: how had conventional treatment been changing survival?
- Current survival of Thalassemia in eastern countries: the Iranian experience
- Thalassemia Intermedia: is disease morbidity observed today less severe compared with Thalassemia Major?
- The role of current chelation treatment in improving prognosis of Thalassemia Major and Intermedia
- The impact of new direct anti-viral treatment in improving prognosis of Thalassemia Syndromes
- What is the impact of novel chelators or new formulations of available chelators treatments on outcomes in Thalassemia Syndromes?
- Current state of total body iron burden in Thalassemia Major and Thalassemia Intermedia determined by MRI: which is the difference in terms of Liver Iron Concentration?
- Practical training for the use of C-licnet as tool for the management of thalassemia syndromes
- Transfusion therapy: what it has been changing with respect to safety and control of body iron loading?
- How much are novel innovative therapies expected to influence the prognosis of Thalassemia Syndromes?
- Gene therapy for Thalassemia and Sickle-Cell-Disease: what it will be the future impact of procedure on this disease?
Module 2

17 September 2017

Introduction to the course: Santina Acuto

Prenatal Diagnosis and Genotype-Phenotype Correlation

Chairperson: Paolo Moi, Italy

Proposed Questions to be addressed:

- How to counsel the couple for beta thal/HbE disease with low severity or predictive factors to decrease the severity of disease?
- How different factors in severity of HbH disease (non-dependent vs dependent blood transfusion) ?
- What is the "flowchart" for prenatal diagnosis of thalassemia?
- Which mistakes may I make in taking clinical decision on the basis of genotype ?
- Which is the target population?
- Which are the main reasons for failure on the thalassemia prevention programs?
- How to do best for the counselling?
- According to the high complication of PND test for pregnant mothers and false negative results, development of another test such as Pre Implantation Genetic Diagnosis (PGD) is necessary, so what are the methods for simplifying the performance of these?
- According of Iran Health Ministry data, the new case of B-thalassemia are 250 newborn patients per year. This happens performing two screening tests in the last years. Therefore, which are the useful methods in order to decrease the birth-rate newborn with B-thalassemia?
- Which is the cut off value of HB A2 to screen for silent beta thalassemia trait?
- Which is the different clinical phenotype of silent beta thalassemia?
- Which is the significance of coinheretence rare hemoglobin band e.g HB Hope
Which is the most recent and easy molecular methods for detection of thalassemia mutation to be used in hematology Lab?

08:30 | 08:50 Counselling and prenatal diagnosis  
Antonis Kattamis, Greece

08:50 | 09:10 Genotype-phenotype correlations  
Antonio Piga, Italy

09:10 | 09:30 Q: Najat Roohaldeen, Mahmoud Hajipour, Tachjaree Panchalee, Mara Memoli, Hala Gabr  
A: Antonis Kattamis and Antonio Piga

Current management of β-thalassemias and hypercoagulable state  
Chairperson: Antonis Kattamis, Greece

Proposed Questions to be addressed:

- What is the pathophysiology of the multiple silent cerebral lesions in patients with Thal major and intermedia?
- Identify the characteristics of an optimal unit of donor red cells for transfusion in thalassemia major.
- What are the most important indications for splenectomy in thalassemia major.
- Identify at least one appropriate use of each of the three available iron chelators.
- What are the therapeutic options for Hypercoagulable state in Thalassemia?
- Which are the main indicators to change chelation treatment?
- Which are the main predictors of disease prognosis?
- Which is the limit of serum ferritin levels in the management of thalassemia iron overloading?
- Which are the main parameters for deciding to start chelation treatment?
- How to manage iron overloading in anemic women during pregnancy?

09:30 | 09:50 Current management of β-thalassemias  
Alan Cohen, USA

09:50 | 10:10 Hypercoagulable State on Thalassemia  
Eliezer Rachmilewitz, Israel

10:10 | 10:30 Q: Najat Roohaldeen, Mahmoud Hajipour, Tachjaree Panchalee, Mara Memoli, Hala Gabr  
A: Alan Cohen and Eliezer Rachmilewitz
Module 2

10:30 | 10:45  Coffee Break

Sickle-Cell Disease and Gene Therapy

Chairperson: Alan Cohen, USA

Proposed Questions to be addressed:

▶ Please describe the background for design of CRISPR for gene correction of thalassemia and sickle cell disease
▶ May you give me an overview of genome editing approaches for gamma-globin gene reactivation?
▶ Which is the role of bone marrow transplantation in the care of people with SCD?
▶ Which are the prevalent phenotype of Sickle Cell carriers?
▶ Which are the main prognostic factors of SCD?
▶ Intrauterine gene therapy for hematological diseases, can it be possible?
▶ What are the current indications for treatment of sickle cell disease with (1) hydroxyurea and (2) regular blood transfusion?

10:45 | 11:05  Sickle Cell Disease: past, present and future Paul Telfer, UK

11:05 | 11:25  Gene Therapy for Hemoglobinopathies Paolo Moi, Italy

11:25 | 11:45  Q: Najat Roohaldeen, Mahmoud Hajipour, Tachjaree Panchalee, Mara Memoli, Hala Gabr

A: Paul Telfer and Paolo Moi

Tissue Iron Measurement by MRI

Chairperson: Paul Telfer, UK

Proposed Questions to be addressed:

▶ Which are the organs where the determination of iron by MRI is crucial?
Module 2

17 September 2017

FICI MEETING ROOM

- Which are the limits of MRI measurements?
- Which are the cut-off of heart and liver iron overloading that I have to monitor for changing chelation treatment?
- Which are the options for managing iron overloading if I do not have access to MRI or this access is limited?
- Which is the timing to repeat LIC and Heart MRI determinations in the patients?

11:45 | 12:05 Tissue Iron Measurement by MRI: Advances, Challenges, and Pitfalls
Tim St Pierre, Australia

12:05 | 12:25 C-licnet Network: potential worldwide MRI network for the management of thalassemia syndromes
Aurelio Maggio, Italy

12:25-12:45 Q: Najat Roohaldeen, Mahmoud Hajipour, Tachjaree Panchalee, Mara Memoli, Hala Gabr
A: Tim St. Pierre and Aurelio Maggio

sponsored by Agilent:

Chairperson: Santina Acuto, Italy

12:45 | 13:10 Measuring Mitochondrial function and Glycolysis for the Metabolic Phenotype detection in Hematological Diseases
E. Di Capua

13:10 | 13:30 Genomic strategies for target capture in onco-hematology
A. D'Agostino

Closing remarks: Aurelio Maggio

13:30 | 14:30 Light Lunch
Module 3

TRAINING COURSE

CLINIC TRAINING in cooperation with the Clinical Nursing Coordinator G. Di Liberto and the Vice Clinical Nursing Coordinator A. Travia

- How and when to change chelation in TM and TI L. Pitrolo, A. Giangreco
- C-licnet training G. Padovano, L. Tesè
- Heart Functional Studies L. Mancuso, M. G. Carmina
- Approach to the patient with SCD: what is changing? R. Di Maggio, D. Renda
- Management of direct anti-HCV drugs in Hemoglobinopathies S. Madonia, R. Barone
- Management of HCC in Thalassemia Syndromes R. Virdone, R. Volpes (ISMETT)

HPLC TECHNOLOGY TRAINING AND DATA INTERPRETATION S. Frediani (sponsored by Tosoh)

- Training (20’)
- Q&A (10’)

PRENATAL SAMPLING

- Practical training on celocentesis, villocentesis and amniocentesis G. Damiani, V. Cigna, G. Schillaci, E. Orlandi

MOLECULAR DIAGNOSIS (route laboratory from the identification of carrier to prenatal diagnosis of thalassemia and other rare Haematological disorders)

- First-level diagnosis of β-thalassemia carrier A. Giambona, M. Cannata
- Interpretation of phenotype and correlation with genotype A. Giambona, M. Vinciguerra
- Second-level diagnosis for genetic disorders: sequencing, array-CGH and Next-Generation F. Leto, C. Passarello, A. Crivello
- Diagnostic and Molecular approach of Congenital dyserythropoietic anemias F. Listì, V. Agrigento, S. Sclafani
- Prenatal genetic counselling A. Giambona, A. Maggio
- Prenatal diagnosis, molecular analysis F. Cassarà, M. Vinciguerra

RESEARCH

- Fertility: gametes cryopreservation M. C. Renda, R. Schillaci, N. Minniti, M. Fecarotta
- Erythroid Cell Culture as model for induction of Fetal Hemoglobin A. Pecoraro, A. Troia
- Citofluorimetry as tool in Haematological rare diseases studies A. Pecoraro, A. Troia
- Cell Sorting for isolation of single cell J. A. Formaro (sponsored by Beckman Coulter)
- Hematopoietic Stem Cell transduction by Recombinant - Lentiviral Vectors S. Acuto, E. Baiamonte, M. Lo Iacono, R. Di Stefano

Dates:
- Module 1 • September 15-16, 2017
- Module 2 • September 17, 2017
- Module 3 • September 18-23, 2017

Course Objective:
The main aim of the course is to bring together experts and young researchers, clinicians and biologists, in order to address key issues in the field of Rare Haematological Diseases. The course is organized into individual sessions to provide a versatile and unique opportunity to get an overview of Rare Haematological Diseases, with particular reference to hemoglobinopathies. For the theoretical part, participants may attend the international meeting and the keynote lectures given by world leaders in the field. For the practical part, attendees will be trained through specific routes ranging from prenatal sampling techniques to laboratory and clinical paths, and to innovative research.

Audience:
- Haematologists, Gynecologists (with particular regards to Prenatal Diagnosis Specialists)
- Biologists, Biotecnologists
- Radiologists
- Medical Technicians

Course Directors:
- Aurelio Maggio - Italy
- Santina Acuto - Italy

International Scientific Board:
- Alan Cohen  - USA
- Ali Taher - Lebanon
- Suthat Fucharoen  - Thailand
- John Porter - UK

Application Deadlines:
- Module 2 & 3: July 20, 2017
- Module 1 only: July 20, 2017

All applications will be submitted to our International Scientific Board. We anticipate being able to notify applicants regarding their applications during the second half of July 2017.

Required Documents for participation:
The following supporting documents should be submitted:
- Curriculum Vitae (CV) - short version
- Official Medical School Transcript
- Personal Statement
- Two (2) letters of reference, including one from your program director.

For further information, please contact the organizing secretariat
Giusy Ventura | Congress Specialist
email: giusy.ventura@collage-spa.it
Office: +39 091 6867 413  direct
Mobile: +39 335 1276715

Physicians (Hematologists, Gynecologists) and Biologists, Radiologists, Medical Technicians will participate in the training course relevant to their specialization.
General Information

Dates:
Module 1 • September 15-16, 2017
Module 2 • September 17, 2017
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Please submit your Application Form

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Giusy Ventura | Congress Specialist

COLLAGE S.p.A.

e-mail: giusy.ventura@collage-spa.it
Office: +39 091 6867 413 direct
Mobile: +39 335 1276715
Attending fees

PACKAGE A:
15-16 SEPTEMBER, 2017 .......................................................... MODULE 1

Registration fees:
(It includes: attendance at the scientific sessions, coffee breaks and buffet lunches as scheduled in the scientific program, congress badge, congress kit) € 400

Accommodation: 2 nights at the Campus in Double Use Single room
(Euro 60,00 per room per night) ............................................................................................ € 120

Transfers: from / to Airport/Hotel/Congress venue (return fare) ........................................ € 100

Total: VAT 22% not included ............................................................................................ € 620

Travel costs are not included

PACKAGE B:
17-23 SEPTEMBER, 2017 .......................................................... MODULE 2/3

Registration fees:
(It includes: attendance at the scientific sessions, congress badge, congress kit) € 1.200

Accommodation: 7 nights at the Campus in Double Use Single room
(Euro 60,00 per room per night) ............................................................................................ € 420

Transfers: from / to Airport/Hotel/Congress venue (return fare) ........................................ € 100

Total: VAT 22% not included ........................................................................................... € 1.720

Travel costs are not included

PACKAGE C:
15-23 SEPTEMBER, 2017 .......................................................... MODULE 1/2/3

Registration fees:
(It includes: attendance at the scientific sessions, congress badge, congress kit) € 1.300

Accommodation: 9 nights at the Campus in Double Use Single room
(Euro 60,00 per room per night) ............................................................................................ € 540

Transfers: from / to Airport/Hotel/Congress venue (return fare) ........................................ € 100

Total: VAT 22% not included ........................................................................................... € 1.940

Travel costs are not included
Terms and conditions

Booking and Payment

All prices are quoted in Euros and do not include 22% VAT.

Confirmation will be sent together with a tax invoice for the total amount of the selected package. For applications sent before July 20, the total due is payable by 30 days from the date of the tax invoice. Applications sent after 20 July must include full payment by 48 hrs from the date of the tax invoice.

All payments should be made in Euros to the following bank account:

COMITATO INTERNATIONAL GROUP FOR THALASSEMIA

IBAN: IT37 E033 5901 6001 0000 0151 884

BIC: BCITITMX

Please note that your registration will only be valid when full payment is received by the administrative office. Payment done by bank transfer must be certified with the copy of the receipt of the bank to be uploaded in the registration form or sent to e-mail to giusy.ventura@collage-spa.it

Cancellation Policy

If notification of cancellation is received in writing:

• between 1 May 2017 and 20 June 2017, you are liable for 50% of the package selected
• from 20 June you are liable for 100% of the package selected.

The Organizing Committee reserves the right to cancel the Congress not later than 15 August 2017 in case of circumstances beyond its control. In such a case all monies paid to date will be refunded in full less any expenses incurred. The liability of the organisers will be limited to that amount.

Our Faculty members possess a strong academic and professional background and years of relevant work experience.

Thank you for taking the time to consider the possibilities this International Meeting has to offer. If you have made your choices, please complete and return to us the application form.

If we can assist with further information, please contact the Organizing Secretariat.

We hope that you are as enthusiastic about the opportunities that are available as we are and look forward to welcoming you to Palermo in September 2017.
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Acknowledgments

We thank Foundation Franco and Piera Cutino for the support to the organization of the course. Moreover, we thank Dr. Paolo Chiesi for his personal kind support aimed at the organization of this event.

Sponsored by

Agilent  Beckman Coulter  Linde

CME Provider and Organizing Secretariat

COLLAGE S.p.A. Via U. Giordano, 37/A - 90144 Palermo - Italy - Tel. +39 091 6867.401 - Fax +39 091 9889354 e-mail: giusy.ventura@collage-spa.it - web: www.collage-spa.it