

Course on Collagenopathies

Bologna, 17–19 March 2015

(Preliminary Programme)

Tuesday, 17th March 2015

ETHICAL AND PRACTICAL CHALLENGES OF DATA SHARING

9.00 - 9.30 REGISTRATION

9.30 - 10.00 INTRODUCTION AND WELCOME
Dr. Luca Sangiorgi (*IRCCS - ISTITUTO ORTOPEDICO RIZZOLI, Bologna, IT*)

10.00 - 14.00 **ROUND TABLE**

Dr. Marina Mordenti (*IRCCS - ISTITUTO ORTOPEDICO RIZZOLI, Bologna, IT*)
“OI and EDS Italian Registry and biobanking”

Dr. Sara Casati (*Bicocca University, Milan, IT*)
“Social, ethical and legal aspects in rare diseases”

Dr. Alessandra Bassotti (*AISED President - Associazione Italiana Sindrome di Ehlers Danlos – Italian Non-profit association of Ehlers-Danlos syndrome*) and **Dr. Barbara Marinelli** (*University of Milan and AISED Scientific Committee*)

OIFE - As.It.O.I. (*Associazione Italiana e Europea di pazienti affetti da OI - EU and Italian Association for patients with OI*)

14.00 - 14.45 LUNCH

STICKLER SYNDROME AND CHONDRODYSPLASIAS

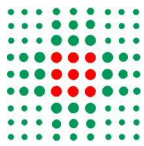
14.45 - 16.30 **LECTURES AND DISCUSSIONS**

Dr. Geert Mortier (*Department Medical Genetics, Antwerp University Hospital, BE*)
“Stickler syndrome: a clinical and molecular overview”

Dr. Geert Mortier (*Department Medical Genetics, Antwerp University Hospital, BE*)
“Collagenopathies causing chondrodysplasias”

16.30 - 16.45 COFFEE BREAK

16.45 - 18.00 **CLINICAL CASE SESSION**



**SERVIZIO SANITARIO REGIONALE
EMILIA - ROMAGNA**

Istituto Ortopedico Rizzoli di Bologna
Istituto di Ricovero e Cura a Carattere Scientifico



Wednesday, 18th March 2015

OSTEOGENESIS IMPERFECTA AND BONE FRAGILITY

9.00 - 13.00 LECTURES AND DISCUSSIONS

Dr. Fleur S. Van Dijk (*Department of clinical genetics, VU medical center, Amsterdam, NL*)

“A clinical and genetic OI overview: re-examined classification with clinical and radiological features” + “Overlap OI/bone fragility: PLS3 mutations in osteoporotic patients with fractures”

Dr. Margherita Maioli (*IRCCS - ISTITUTO ORTOPEDICO RIZZOLI, Bologna, IT*)

“Genotype-phenotype correlation in autosomal dominant OI”

Dr. Fransiska Malfait (*Center for Medical Genetics, Ghent University Hospital, BE*)

“Overlap syndrome OI/EDS”

Dr. Maria Luisa Bianchi (*Istituto Auxologico, Milan, Italy*)

“Hypophosphatasia and idiopathic juvenile osteoporosis”

13.00 - 14.00 LUNCH

14.00 - 16.00 LECTURES AND DISCUSSIONS

Dr. Maria Gnoli (*IRCCS - ISTITUTO ORTOPEDICO RIZZOLI, Bologna, IT*)

“Clinical evaluation and follow-up strategies for OI patients”

Dr. Paolo Fraschini (*IRCCS “E. Medea”, Lecco, IT*)

“Management, rehabilitation and therapy path for OI patients”

Dr. Paul Coucke (*Department of Medical Genetics, Ghent University Hospital, BE*)

“Mouse and zebrafish models and new research updates in collagenopathies”

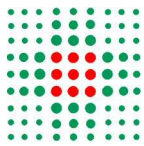
“Laboratory Testing: Whole Exome Sequencing in diagnostics”

16.00 - 16.15 COFFEE BREAK

16.15 - 18.00 CLINICAL CASE SESSION

Dr. Mauro Celli (*Department of Pediatrics, La Sapienza University of Rome, IT*)

Clinical cases of patients with Osteogenesis Imperfecta



Thursday, 19th March 2015

EHLERS-DANLOS SYNDROME

9.00 – 13.00 LECTURES AND DISCUSSIONS

Dr. Fransiska Malfait (*Center for Medical Genetics, Ghent University Hospital, BE*)
“Overview of connective tissue biology and EDS as of recent discoveries”

Dr. Marco Castori (*Department Medical Genetics, San Camillo Forlanini, Rome, IT*)
“Clinical assessment of joint hypermobility and management of related symptoms”

Dr. Cecilia Giunta (*Division of Metabolism, Connective Tissue Unit, University Children's Hospital, Zürich, CH*)
“Clinical and molecular insights into the kyphoscoliotic type of Ehlers-Danlos syndrome and its differential diagnoses”

Dr. Francesca Ponti (*IRCCS - ISTITUTO ORTOPEDICO RIZZOLI, Bologna, IT*)
“Contribution of COL5A1 G530S polymorphism as a disease modifying factor in Ehlers-Danlos Syndrome”

13.00 - 14.00 LUNCH

14.00 - 16.00 LECTURES AND DISCUSSIONS

Dr. Fransiska Malfait and Dr. Paul Coucke (*Center for Medical Genetics, Ghent University Hospital, BE*)
“Functional and biochemical analysis of proteins involved in collagenopathies”

16.00 - 16.15 COFFEE BREAK

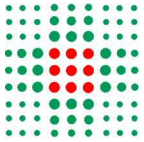
16.15 - 18.00 CLINICAL CASE SESSION

Dr. Filippo Boriani (*IRCCS - ISTITUTO ORTOPEDICO RIZZOLI, Bologna, IT*)
Clinical cases of patients with the hypermobility type of Ehlers-Danlos syndrome

Dr. Marco Castori (*Department Medical Genetics, San Camillo Forlanini, Rome, IT*)
Clinical cases of patients with the Ehlers-Danlos syndrome

Dr. Alessandra Bassotti (*Ehlers-Danlos Disease Regional Service, Dept. of Preventive Medicine, Fondazione Cà Granda, IRCCS Osp. Maggiore Policlinico, Milan, Italy*) and **Dr. Francesca Cortini** (*Center of Molecular and Genetic Epidemiology, Dept. of Environmental and Occupational Health, University of Milan, Fondazione Cà Granda, IRCCS Osp. Maggiore Policlinico, Milan, Italy*)

The Regional Service of "Milan Policlinico Hospital" for Ehlers-Danlos syndrome: interdisciplinary cooperation for treatment and care with a highlight on next generation sequencing approach for improving research and diagnosis



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18.00 – 18.15 WRAP UP TALK

Dr. Luca Sangiorgi (*IRCCS - ISTITUTO ORTOPEDICO RIZZOLI, Bologna, IT*)

*A maximum of **30 people** can take this course, which confers Continuing Medical Education credits (CME). The course is organized by the Department of Medical Genetics and Rare Orthopaedic Diseases and Clibi Laboratory.*

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