1ST COL4A1-A2 EUROPEAN CONFERENCE

ROME 19.02.2024

Sala Zuccari, Palazzo Giustiniani
Senato della Repubblica,
Via della Dogana Vecchia 29, Roma
10AM-6PM CET

ASSOCIAZIONE FAMIGLIE COL4A1-A2

AGENDA

09:30 Registration of participants

10:00 Welcome greetings -Institutions
Sen. Marco Lombardo

Welcome greetings - Associazione Famiglie COL4A1-A2 APS
Simona Manodoro, Francesca Manodoro

10:15 The phenotypic spectrum of COL4A1/A2-related disorders: a focus on paediatric age
Simona Orcesi
Q&A

10:45 COL4A1 and COL4A2: what the literature tells us
Eleonora Bonaventura
Q&A

11:15 Coffee break

11:30 Molecular genetics of all known COL4A1/COL4A2 diseases
Elisabeth Tournier Lasserve
Q&A

12:00 Molecular genetics of fetal intracranial hemorrhage
Thibault Coste
Q&A

12:30 Research session:
Establishing foundations for translational interventions
Douglas Gould

Gene therapy for rare genetic vascular disorders: a patient-driven journey in drug development
Patricia Musolino

Q&A

Moderator: Simona Balestrini

13:30 Lunch buffet

14:30 Neuroimaging aspects: from prenatal to adulthood
Anna Pichiecchio, Cecilia Parazzini
Q&A

15:00 Adult neurology session:
Handling genetic cerebral small vessel diseases: care organization in France and collaboration at the European level
Dominique Hervé

Overview of clinical and neuroradiological features of COL4A1/A2 adult patients
Anna Bersano

The French COL4A1/COL4A2 cohort: phenotypic spectrum
Stephanie Guey

Q&A

16:10 Coffee break

16:30 Kidney manifestations in COL4A1/COL4A2 related diseases: how to detect and how to treat
Emmanuelle Plaisier
Q&A

17:00 The COL4A1/COL4A2 ocular phenotypic spectrum: the pediatric ophthalmology perspective
Giacomo Bacci
Q&A

17:30 Cardiological aspects highlighted in patients with COL4A1-A2
Giulio Porcedda
Q&A

18:00 End

With the non-conditioning contribution of
Valentina Nardi
CONSULENTE DEL LAVORO - PSICOLOGA

Con il patrocinio di
ÜNIAMO
Federazione Italiana Malattie Rare

Under the Patronage of
EURORDIS
RARE DISEASES EUROPE
OMAR
OSSERVATORIO MALATTIE RARE
ALLEANZA MALATTIE RARE

Associazione Famiglie COL4A1-A2 APS
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FACULTY

Bacci Giacomo Oftalmologia – A.O.U. Meyer, Firenze, Italia

Balestrini Simona Neurologia pediatrica – A.O.U. Meyer, Firenze, Italia

Bersano Anna Unità Cerebrovascolare, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano, Italia

Bonaventura Eleonora Neurologia Pediatrica – Ospedale dei Bambini V. Buzzi, Milano, Italia

Coste Thibault Service de génétique neurovasculaire Hôpital Saint Louis, Paris, France

Gould Douglas Department of Ophtalmology and Anatomy - Institute for Human Genetics, School of Medicine, University of California, San Francisco, USA

Guey Stephanie Service de Neurologie, Centre de Référence des Maladies Vasculaires Rares du Cerveau et de l'Oeil, Hôpital Lariboisière, Paris, France

Hervé Dominique Service de Neurologie, Centre de Référence des Maladies Vasculaires Rares du Cerveau et de l'Oeil, Hôpital Lariboisière, Paris, France

Musolino Patricia Pediatric Stroke and Vascular Gene Therapy Programs - Center for Genomic Medicine - MGH-Harvard Medical School, USA

Orcesi Simona U.O. Neuropsichiatria infantile – IRCCS Fondazione Istituto Neurologico C. Mondino, Pavia e Università di Pavia, Italia

Parazzini Cecilia Radiologia e Neuroradiologia Pediatrica – Ospedale dei Bambini V. Buzzi, Milano, Italia

Pichiecchio Anna Neuroradiologia – IRCCS Fondazione Istituto Neurologico C. Mondino, Pavia e Università di Pavia, Italia

Plaisier Emmanuelle Department of Nephrology and Dialysis INSERM Hôpital Tenon Université Pierre et Marie Curie Paris, France

Porcedda Giulio Cardiologia pediatrica e della transizione – A.O.U. Meyer, Firenze, Italia

Tournier Lasserve Elisabeth Service de génétique moléculaire Neurovasculaire, Hôpital Saint-Louis, Paris, France - Université de Paris, INSERM Paris, France

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