

# 2<sup>ND</sup> COL4A1-A2 INTERNATIONAL CONFERENCE

ROME 10.02.2025

CNR  
Sala Convegni  
via dei Marrucini, snc  
9:30AM-6:00PM



ASSOCIAZIONE  
FAMIGLIE  
COL4A1-A2

## AGENDA

**09:00** Registration of participants

**09:30** Welcome greetings - Associazione Famiglie COL4A1-A2 APS

Simona Manodoro and Francesca Manodoro

**09:45** Welcome greetings - CNR

CNR

### **Clinical section:**

**10:00** Characterization and management of epilepsy in COL4A1/2-related disorders

Simona Balestrini

**10:30** Presentation of RENDER: Rare Neuropediatric Diseases Electronic Registry

Enza Maria Valente - Davide Politano

**11:00** Building Consensus: Expert Recommendations for COL4A1 and COL4A2-Related Disorders

Diana Tambala

**11:30** Coffee break

**11:50** UCSF Gould Syndrome Clinic: Year One

Rachel Vassar

### **Research section:**

**12:20** Identifying mechanism-based and mechanism-agnostic interventions for Gould syndrome

Douglas Gould

**13:00** Phenotypic and genetic overview of mutations in COL4A1 and COL4A2: insights from the French cohort

Stephanie Guey - Thibault Coste

**13:30** Lunch buffet

**14:30** The collaboration between Associazione Famiglie COL4A1-A2 and Fondazione Telethon

Alessandra Camerini

**14:50** FishCOLler: zebrafish COL4A1/A2 syndrome models as a resource to speed up functional validation of emerging variants and search for pathomechanisms and druggable targets"

Marco Tartaglia - Antonella Lauri

**15:20** Developing zebrafish models to study COL4A1-related disease

Paul Kasher

**15:50** Age-dependent cerebral vascular dysfunction in Col4a1 mutant mice

Scott Earley

**16:20** Coffee break

**16:40** Modelling cerebral Small Vessel Disease in a dish - towards new treatments

Alessandra Granata

**17:10** Developing gene targeted therapies for COL4A1 and COL4A1 related disorders: opportunities and challenges

Patricia Musolino

**Conclusions**

**18:00** End

Moderators: Marina Macchiaiolo, Simona Orcesi, Davide Tonduti

**Associazione Famiglie COL4A1-A2 APS**

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## FACULTY

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

**Camerini Alessandra** Relations with Patients and Patient Organisations - Telethon, Milano, Italia

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

**Earley Scott** Pharmacology and Physiology – University of Rochester, NY, USA

**Gould Douglas** Department of Ophthalmology 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

**Granata Alessandra** Neurology Unit - University of Cambridge, Cambridge, UK

**Kasher Paul** Manchester Stroke research group, University of Manchester, Manchester, UK

**Lauri Antonella** Molecular genetics & Functional genomics - Ospedale Pediatrico Bambino Gesù, Rome, Italy

**Musolino Patricia** Center for Genomic Medicine - Massachusetts General Hospital, Boston, USA

**Politano Davide** U.O. Neuropsichiatria infantile – IRCCS Fondazione Istituto Neurologico C. Mondino

**Tambala Diana** Center for Genomic Medicine - Massachusetts General Hospital, Boston, USA

**Tartaglia Marco** Molecular genetics & Functional genomics - Ospedale Pediatrico Bambino Gesù, Rome, Italy

**Valente Enza Maria** Genetica Clinica e Molecolare delle Malattie Rare - IRCCS Fondazione Mondino, Pavia, Italy

**Vassar Rachel** Pediatric Clinical Care – Clinical Research – University of California, San Francisco, USA

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