

INSIEME PER LA RICERCA PCDH19 - ETS



SEVENTH WORLD CONFERENCE ON PCDH19

Scientific Research Status ad Therapeutic Perspectives

28 March 2025 at Fondazione Telethon, via Varese, 16/B - ROMA

Session 1: Clinical updates

Moderators:

Nicola Specchio – Ospedale Pediatrico Bambino Gesù - Roma

Carla Marini - Presidio Ospedaliero G. Salesi; Ospedali Riuniti - Ancona

SPEAKERS	TOPICS	Time 9.15 – 11.00
Francesca Squillante Insieme per la Ricerca PCDH19 ETS	Welcome greetings	9.15 – 9.30
Federico Vigevano IRCCS San Raffaele - Roma	State of knowledge about PCDH19	9.30 – 10:00
Alice Dainelli Ospedale pediatrico Meyer - Firenze	Initial findings from the Italian RESIDRAS: advantages and complexities in registries implementation	10.00 – 10.20
Francesca Ragona Istituto Neurologico Carlo Besta – Milano	Cognitive and behavioural features in PCDH19	10.20 - 10.40
Marina Trivisano Ospedale Pediatrico Bambino Gesù - Roma	Study Proposal on Therapy Approaches, Effective Treatments, and Therapy Discontinuation	10.40 – 11.00
COFFEE BREAK		11.00 - 11.30

Session 2: Molecular mechanisms

Moderators:

Patrizia D'Adamo - Ospedale San Raffaele - Milano

Gaia Colasante – Ospedale San Raffaele - Milano

SPEAKERS	TOPICS	Time 11.30 – 13.00
Joseph Gecz University of Adelaide	Genotype-Phenotype complexities of PCDH19 variation	11.30 – 12.00
Isabel Martinez Garay University of Valencia	Roles of the epilepsy-related protein PCDH19 during cortical development	12.00 -12.30
Rossella Borghi Ospedale Pediatrico Bambino Gesù - Roma	Altered cytoskeleton dynamics in patient-derived iPSC-based model of PCDH19 clustering epilepsy	12.30 – 13.00
LUNCH		13.00-14.00

Session 3: Therapeutic approaches

Moderators:

Gaia Colasante – Ospedale San Raffaele - Milano

Gabriele Lignani - UCL Queen Square Institute of Neurology - Londra

SPEAKERS	TOPICS	Time 14.00 – 15.30
Jack Parent University of Michigan	Studying interneuron development in Protocadherin-19 Clustering Epilepsy using mouse and brain organoid models	14.00- 14.30
Paul Thomas/Stefka Tasheva University of Adelaide	Assessing precision medicine strategies for PCDH19-Clustering Epilepsy using preclinical mouse models	14.30 – 15.00
Steven Petrou Praxis Precision Medicines Boston	Targeting Mosaicism in PCDH19 Epilepsy: Development of an ASO Therapy to Silence X-Linked Expression	15.00- 15.30

Session 4: ROUND TABLE GENETIC & CLINIC

16.00 – 17.00

Moderatori:

Raffaella Cusmai – Lega Italiana Contro Epilessia (LICE)

Federico Vigevano - IRCCS San Raffaele - Roma

SESSIONI IN ITALIANO DEDICATE ALLE FAMIGLIE DEI PAZIENTI

Sessione 1 BIS:	09.30 – 10.30
IL COMITATO SCIENTIFICO INCONTRA LE FAMIGLIE	
Moderatori:	
Gaia Colasante – Ospedale San Raffaele - Milano	
Patrizia D'Adamo - Ospedale San Raffaele - Milano	
Sessione 2 BIS:	10.30 – 13.00
I CLINICI INCONTRANO LE FAMIGLIE	
Moderatori:	
Raffaella Cusmai - Lega Italiana Contro Epilessia (LICE)	
Federico Vigevano - IRCCS San Raffaele - Roma	



IRCCS San Raffaele

