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2026
PISA



IRCCS FONDAZIONE
STELLA MARIS



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ÜNIAMO
Federazione Italiana Malattie Rare



International Symposium on KIF1A Disorders

International Scientific Symposium in Pisa dedicated to the
current state and future directions on KIF1A disorders

02.10.2026
IRCCS STELLA MARIS FOUNDATION

DAY 1
FRIDAY

14:00 OPENING CEREMONY

Director of IRCCS Stella Maris: Prof. Giovanni Coni
KIFIA Italia APS ETS President: Dr Alessandra Bison
UNIAMO President: Dr Annalisa Scopinaro

SESSION 1

KIFIA: INTRACELLULAR ROLE, MOLECULAR BIOLOGY AND GENETIC

Moderators: Dr Alessandro De Falco
and Prof. Nicola Brunetti-Pierri (TIGEM)

14:30 PROF. ANTONELLA DE MATTEIS (TIGEM, NAPLES)

KIFIA and its intracellular role

14:50 PROF. GENNARO GAMBARDILLA (TIGEM, NAPLES)

The KIFIA gene and long-read NGS sequencing

15:10 DR CLAUDIA CIACCIO (IRCCS BESTA, MILAN)

Genetic basis and recurrence risk

SESSION 2 - PART I

KIFIA-RELATED DISORDERS: FROM NEUROLOGY TO NEUROPSYCHIATRY

Moderators: Dr Stefania Della Vecchia and
Prof. Roberta Battini (IRCCS Stella Maris, PISA)

15:30 DR CHIARA PAOLELLA (ORN SANTOBONO, NAPLES)

Neuroimaging in KIFIA-associated disorders

15:50 DR STEFANO D'ARRIGO (IRCCS BESTA, MILAN)

Neurodevelopmental disorders in KIFIA syndrome

16:10 DR WENDY CHUNG

Neurodegeneration



16:30 COFFEE BREAK



SESSION 2 - PART II

Moderators: Prof. Filippo Maria Santorelli (IRCCS Stella Maris, PISA), Dr Stefano D'Arrigo (IRCCS Besta, MILAN)

16:40 DR GABRIELA SILVESTRI (FONDAZIONE POLICLINICO A. GEMELLI, IRCCS BESTA, UOC NEUROLOGIA)

Phenotypic spectrum of adult-onset KIFIA syndrome

17:00 DR FRANCESCO NICITA (HOSP. BAMBINO GESÙ, ROMA)

Pediatric Neurology

17:20 DR STEFANIA DELLA VECCHIA (IRCCS STELLA MARIS, PISA)

Retrospective Italian KIFIA patient cohort study

PROGRAMMA

SESSION 3

THERAPEUTIC APPROACHES FOR KIFIA-RELATED DISORDERS

Moderators: Prof. Nicola Brunetti-Pierri (TIGEM),
Prof. Roberta Battini (IRCCS Stella Maris, PISA)

17:40 DR ALESSANDRO DE FALCO (TIGEM)

Clinical Development Roadmap and Trial Readiness for ASO Therapy

18:00 DR LUIS ALBERTO BAENA LOPEZ (CENTRO DE BIOLOGÍA MOLECULAR SEVERO OCHOA, CSIC, MADRID, SPAIN)

Caspases as guardians of KIF1A homeostasis and function



18:20 DOTT. SSA WENDY CHUNG (BOSTON CHILDREN'S HOSPITAL, BOSTON, MA, USA)

Clinical experience with ASO therapy



03.10.2026

IRCCS FONDAZIONE STELLA MARIS

DAY 2
SATURDAY

SESSION 4

VISUAL SYSTEM INVOLVEMENT IN KIFIA-RELATED DISORDERS

Moderators: Dr Alessandro De Falco (TIGEM),
Dr Stefania Della Vecchia (IRCCS Stella Maris, Pisa)

09:00 DR SABRINA SIGNORINI (FONDAZIONE MONDINO, PAVIA)

Visual system involvement in KIF1A-related neurological disorders: from assessment to intervention

09:20 DR GIACOMO MARIA BACCI (AOU MEYER, FLORENCE)

Diagnostic clues and management of ocular manifestations in KIF1A-related phenotypes

SESSION 5

BEYOND DIAGNOSIS: ASSESSMENT PATHWAYS, REHABILITATION AND FAMILY SUPPORT

Moderators: Dr Alessandro De Falco (TIGEM),
Dr Stefania Della Vecchia (IRCCS Stella Maris, PISA)

09:40 DR ASSUNTA GUILLARI (AOU FEDERICO II, NAPLES)

Beyond diagnosis: Living with a rare disease as a family what we know, what is missing and why it matters

10:00 DR ELENA MORETTI (UNIVERSITÀ DI PISA)

Communication and language in KIF1A: understanding together to provide better support

PROGRAMMA

- 10:20 DR FEDERICA D'ACUNTO (IRCCS STELLA MARIS, PISA)**
From assessment to rehabilitation in individuals with KIFIA-related disorders: adaptation of motor scales and recommendations for orthoses and assistive devices
- 10:40 DR PAOLA CRISTOFANI (IRCCS STELLA MARIS, PISA)**
KIFIA: understanding cognitive profiles and emotional experiences - current knowledge and open questions
- 11:00 DR MICHELINA ARMANDO (HOSP. BAMBINO GESÙ, ROME)**
Individual Rehabilitation Plan (IRP): integrating motor and visual-perceptual rehabilitation

11:20 COFFEE BREAK



SESSION 6

12:00 ROUND TABLE - MULTIDISCIPLINARY DISCUSSION