

# INTERNATIONAL WORKSHOP ON FRAGILE X SYNDROME



**PADUA, ITALY**

Palazzo del Bo - Aula Nievo



**JUNE 20<sup>th</sup>  
2025**



UNIVERSITÀ  
DEGLI STUDI  
DI PADOVA



PIÙ INSIEME  
Azienda  
Ospedale  
Università  
Padova



SOCIETÀ ITALIANA  
DI DIAGNOSTICA PRENATALE



UNITI PER  
CRESCERE



FONDAZIONE  
ISTITUTO DI RICERCA  
PEDIATRICA



DIPARTIMENTO  
DI INGEGNERIA  
DELL'INFORMAZIONE



SOCIETÀ ITALIANA  
DI PEDIAGOGIA



Associazione Italiana  
Sindrome 'X-Fragile'

**Partecipazione gratuita, previa registrazione | Free participation, upon registration**

[www.motoresanita.it](http://www.motoresanita.it)

**MOTORE  
SANITÀ**  
Cure the Future

**In memory of Alessandra Murgia, to celebrate her outstanding contributions to improving the knowledge and the care of children and families affected by Fragile X Syndrome.**



INTERNATIONAL WORKSHOP ON

# FRAGILE X SYNDROME

PADUA, ITALY - JUNE 20<sup>th</sup> 2025

## INTRODUCTORY NOTES

Since the description of a syndrome of X-linked intellectual disability associated with a cytogenetic fragile site on the distal long arm of the X chromosome 56 years ago, Fragile X Syndrome still represents a unique and demanding challenge for basic scientists as well as clinicians. Still a lot needs to be understood regarding the involvement of the responsible gene, the *FMR1* gene, in normal brain development and function and, more importantly, the genetic alterations which can become druggable targets. Much also needs to be done to advance our clinical understanding of the Fragile X Syndrome.

In fact, few doctors have specialized knowledge of this disease and still fewer are located in centers of excellence for the care of these patients within institutions capable of providing effective, efficient and comprehensive care and follow-up for the affected patients and their families. As a consequence, it is still a priority to create opportunities for scientists and clinicians to meet together and to share data and experience and ideas and models on how to best respond to the needs of the affected patients and their families.

Padua, thanks to an intimate relationship between its University and its great Hospital, led by an outstanding clinical scientist, the late prof. Alessandra Murgia, adds its name to the centers in the world that have made a significant contribution to increasing our knowledge and our ability to provide quality care for the patients affected by the Fragile X Syndrome. We all recognize the importance and value of bringing together basic science, clinical research and clinical care within the "Fragile X community" and are taking this opportunity to also celebrate the contributions of Prof. Alessandra Murgia by inviting you all to be with us on June 20th of this year here in the unique setting of this more than 800-hundred-year-old University.



INTERNATIONAL WORKSHOP ON

# FRAGILE X SYNDROME

**PADUA, ITALY - JUNE 20<sup>th</sup> 2025**



## PRELIMINARY PROGRAM

**9.30** [Note di Benvenuto](#) | Welcome address

**10.00** [Memoria di Alessandra Murgia](#) | In memory of Alessandra Murgia  
**Robert Nussbaum**

**10.15** SESSIONE I

[Quadri clinici e Nuove Terapie](#) | Clinical spectrum and New Therapies  
[Modera](#) | Chairperson: **Stefano Sartori**

**10.20** [Disabilità intellettiva legata al cromosoma X e il gene FMR1](#) | X-linked intellectual disability (XLID) and the FMR1 gene  
**Giovanni Neri**

**10.40** [Studi clinici e nuove terapie per la sindrome dell'X Fragile](#) | FXS clinical trials and new therapies  
**Randi Hagerman**

**11.00** [FXTAS clinica / neuropatologia e basi molecolari](#) | FXTAS clinical / neuropathology and molecular underpinnings  
**Paul Hagerman**

**11.20** [Basi molecolari del locus FMR1, biomarcatori molecolari suscettibilità/rischio di sviluppo della malattia nella condizione associata alla premutazione dell'X Fragile](#) | Molecular basis of the FMR1 locus, molecular biomarkers susceptibility/risk of disease development in Fragile X-premutation associated condition (FXPAC)  
**Flora Tassone**

**11.40** [Discussione](#) | Discussion

**12.00** [Colazione di lavoro](#) | Lunch break



INTERNATIONAL WORKSHOP ON

# FRAGILE X SYNDROME

**PADUA, ITALY - JUNE 20<sup>th</sup> 2025**



**13.00 Ricerca di Base e Translazionale nella FXS - Meccanismi Molecolari e Biomarcatori** | Basic and Translational research in FXS - Molecular Mechanisms and Biomarkers

**Moderano** | Chairpersons: **Maria Giuseppina Miano & Robert Nussbaum**

**13.05 Instabilità della ripetizione CGG di FMR1 e mosaicismismo somatico** | FMR1 CGG repeat instability and somatic mosaicism. **Flora Tassone**

**13.25 Cosa ci insegna un modello murino della sindrome dell'X Fragile** | What a mouse Model for Fragile X teaches Us **Claudia Bagni**

**13.45 Organoidi cerebrali umani per lo studio della sindrome X Fragile (FXS)** | Engineering human brain organoids to study Fragile X Syndrome (FXS) **Nicola Elvassore**

**14.05 Biomarcatori quantitativi della sindrome dell'X Fragile: dall'analisi del cammino alla classificazione dei bambini con X Fragile** | Innovative quantitative biomarkers of FXS: from gait analysis to FXS children classification **Zimi Sawacha**

**14.25 Discussione** | Discussion

**15.00 SESSIONE II**

**"Fragile X Syndrome" – La cura, lo sviluppo e le azioni intraprese** | "Fragile X Syndrome - Care, development and intervention

**Moderano** | Chairpersons: **Agnies van Eeghen & Alessia Brunetti**

**15.05 Il panorama europeo emergente sulle Malattie Rare – Gli "European Reference Network per le Malattie Rare [ERN]" - ERN ITHACA – ERN sulle sindrome malformativa rare, disturbi intellettivi e altri disordini del Neurosviluppo** | Emerging European Panorama – The European Reference Networks on Rare Diseases [ERN] – ERN ITHACA, ERN Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders **Alain Verloes**



INTERNATIONAL WORKSHOP ON

# FRAGILE X SYNDROME

**PADUA, ITALY - JUNE 20<sup>th</sup> 2025**



**15.25 Il bisogno di un approccio olistico integrato per la cura dei pazienti affetti da Sindrome dell'X Fragile | The need for a holistic, integrated approach to care for Fragile X Syndrome**

**Andrew Stanfield**

**15.45 FraXI e sviluppo di linee guida per la sindrome X Fragile | FraXI and Developing Fragile X Guidelines**

**Kristen Johnson**

**16.05 Il modello padovano per la presa in carico dei pazienti con sindrome dell'X Fragile | The Padua Model for the care of patients with Fragile X Syndrome**

**Elisa Di Giorgio**

**16.25 Tavola Rotonda | Round Table Discussion**

**Mosaicismo nella sindrome dell'X Fragile e nei disturbi associati: una nuova sfida diagnostica e clinica | Mosaicism in Fragile X Syndrome and Associated disorders: a new diagnostic and clinical challenge**

**Moderano | Chairpersons: Flora Tassone & Andrew Stanfield**

**17.15 Fine dei lavori | End of the Meeting**



INTERNATIONAL WORKSHOP ON

# FRAGILE X SYNDROME

PADUA, ITALY - JUNE 20<sup>th</sup> 2025

## Prof. Alessandra Murgia's obituary

Professor **Alessandra Murgia** [1955-2024] graduated in Medicine at the Medical School of University of Padua, Italy in 1981, where then she completed a residency in Endocrinology, first [1982/1984] and in Pediatrics, thereafter [1992-1995]. Always at the University of Padua she completed a PhD course in "Human Development Science" [1989-1992]. In 1985 she obtained a post-doctoral fellowship at the Department of Internal Medicine and Genetics at the University of Pennsylvania [1985/1987], in Philadelphia, Pennsylvania, and then at the Department of Human Genetics in the Robert L. Nussbaum's laboratory [1989-1991]. It was during the years spent at the University of Pennsylvania that she developed her knowledge and competence in the field of molecular genetics. In 1992 she came back to the University of Padua, where in 2002, she was appointed as an Associate Professor in the Department of Woman's and Child's Health where she remained until her untimely death. Throughout her career, she maintained a rich and fulfilling personal life that she enjoyed with her husband Giorgio, her two married sons, two beautiful daughters-in-law, two beautiful grandchildren and the numerous friendships she made with colleagues from around the world.



INTERNATIONAL WORKSHOP ON

# FRAGILE X SYNDROME

PADUA, ITALY - JUNE 20<sup>th</sup> 2025

Upon coming back to Padua, she was among the first clinical scientists at the Medical School of the University of Padua to start a diagnostic and research lab in molecular genetics. In a very short period of time her lab became a referral diagnostic center for genetic diseases such as Von Hippel-Lindau syndrome, congenital hearing loss and neurodevelopmental disorders, notably Fragile X syndrome. In the course of the years, she focused her clinical and research activity mainly on this latter disorder. This focus required also the creation of a multi-professional clinical and research program on Fragile X syndrome, intimately linked to the lab, thus realizing a unique model to foster innovation and research, from one side, and to provide a comprehensive care to these patients and families, on the other. In brief, thanks to her knowledge, her competence spanning from the bench to the patients' care, her passion, her commitment and her dedication, a unique ecosystem for promoting excellence in research, diagnosis and clinical care dedicated to the Fragile X syndrome was created. Basic scientists, molecular biologists, bioengineers, clinical psychologists started to work with her. Some specific contributions to the field in Padua include an organoid brain model to study the FMR1 gene function and an innovative "clinical biomarker of the disease" based on the results of the gait analysis of the affected children developed through a machine learning approach. But probably, even more importantly, is the fact that due to her attitude and leadership, she brought together a unique group of young doctors and nurses dedicated to the study and care of the Fragile X Syndrome. Prof. Murgia became a nationwide referring person to many children and families affected by this syndrome.

As all who knew her can attest, she was a woman of strong opinions and fierce loyalties who was first and foremost not particularly tolerant of her own frailties. She was a warrior and that personal attitude allowed her to fight her last battle against the inexorable disease she had to face. She confronted the inevitable reality with dignity, courage and determination leaving all the people who knew her with a great sense of appreciation and admiration and served as a role model for how to live a life and how to leave it.



INTERNATIONAL WORKSHOP ON

# FRAGILE X SYNDROME

**PADUA, ITALY - JUNE 20<sup>th</sup> 2025**



## FACULTY

**Claudia Bagni**, Professor of Biology, Department of Biomedicine and Prevention, University "Tor Vergata", Rome

**Alessia Brunetti**, Chair of the Board of the Italian Fragile X Association, Rome

**Elisa Di Giorgio**, Associate Professor of Psychology, Department of Developmental Psychology and Socialisation, University of Padua

**Agnies van Eeghen**, arts VG, Principal Investigator, Amsterdam UMC

**Nicola Elvassore**, Professor of Chemical Engineering, Department of Industrial Engineering, University of Padua

**Kirsten Johnson**, Chair of the Board of Fragile X International (FraXI), UK

**Paul Hagerman**, Distinguished Professor, Department of Biochemistry and Molecular Medicine MIND Institute Sacramento, CA, USA

**Randi J. Hagerman**, Distinguished Professor, Department of Pediatrics, Endowed Chair in Fragile X Research, Medical Director of MIND Institute Sacramento, CA, USA

**Maria Giuseppina Miano**, CNR Senior Scientist, Human Molecular Neurogenetics, Institute of Genetics and Biophysics "ABT" CNR Naples

**Giovanni Neri**, Professor of Medical Genetics, Catholic University "Sacro Cuore", Rome

**Robert Nussbaum**, Professor of Human Genetics, Consulting - Baylor College of Medicine, San Francisco, CA, USA

**Giorgio Perilongo**, Professor of Pediatric, Department on Woman's and Child's Health, University of Padua

**Zimy Sawacha**, Professor of Engineering, Department of Information Engineering, University of Padua

**Stefano Sartori**, Associate Professor of Child Neurology, Department of Woman's' and Child's Health, University of Padua

**Andrew Stanfield**, Senior Clinical Research Fellow, Director of Clinical Research at the Patrick Wild Centre, University of Edinburgh, Scotland

**Flora Tassone**, Professor of Molecular Genetics, Department of Biochemistry and Molecular Medicine MIND Institute Sacramento, CA, USA

**Alain Verloes**, Professor of Genetics, Université Paris Cité (UPC) and R DEBRE University Hospital, Paris – Coordinator ERN ITHACA





**Domestic information**

**Venue of the meeting**

"Bo Palace" [Palazzo del Bo] – via VII Febbraio, 2– 35122 Padova, Italy

**Scientific Committee**

Elisa Bettella, Marilena Cameran, Elisa Di Giorgio, Valentina Liani, Giorgio Perilongo, Roberta Polli, Zimi Sawacha

**Scientific Secretariat**

Giorgia Vigo - [giorgia.vigo@aopd.veneto.it](mailto:giorgia.vigo@aopd.veneto.it) - Tel +39.049.8218070  
Giorgio Perilongo – [giorgio.perilongo@unipd.it](mailto:giorgio.perilongo@unipd.it) – Cell +39.334.6539925

**Organizing Agency**

EXTRA scs

**Partecipazione gratuita, previa registrazione**

**Free participation, upon registration**

[www.motoresanita.it](http://www.motoresanita.it)

