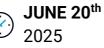
INTERNATIONAL WORKSHOP ON **FRAGILE X SYNDROME**









Partecipazione gratuita, previa registrazione | Free participation, upon registration

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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.



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PADUA, ITALY - JUNE 20th 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.



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



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 mosaicismo 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 guantitativi della sindrome dell'X Fragile: dall'analisi del cammino alla classificazione dei bambini con X Fragile I 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 II 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





15.25 || 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 II 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

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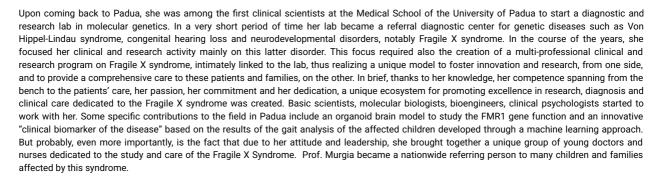
Prof. Alessandra Murgia's obituary

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.

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MOTORE SANITA



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.



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, Unviersity 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

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Organizing Agency

EXTRA scs

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