

1st Fragile X International Congress

Palau Macaya, Barcelona, Spain

Thursday, 7 November, 2024

08:30 *Sign in and networking*

09:00 Opening Welcome

Kirsten Johnson, [FraXI](#)
Pilar Elias, L'Associació Catalana
de la Síndrome X Fràgil

Topic 1 *FXPAC: FXTAS & FXPOI*

09:10 Development of molecular biomarkers as
susceptibility/risk of development, progression
and severity of FXTAS

Flora Tassone, [MIND
Institute, UC Davis](#) (US)

09:30 Neuroimaging evidence of glymphatic system
dysfunction in FXTAS

Andrea Elias (ES)

09:50 Care for patients with FXTAS

Sundus Alusi, [The Walton
Centre, Liverpool](#) (UK)

10:10 FMR4 as a potential blood biomarker for fragile X-
associated primary ovarian insufficiency

Laia Rodríguez-Revena,
[Hospital Clinic Barcelona](#)
(ES)

10:30 *Networking coffee and tea break*

Topic 2 *FXPAC: Other premutation issues*

11:00 Social and emotional wellbeing in females with the
fragile X premutation

Jo Moss, [University of
Surrey](#) (UK)

11:20 Other FXPAC-Related Symptoms and Conditions

Dragana Protic, [University
of Belgrade](#) (RS)

11:40 Age and CGG-dependent changes in depression
symptom severity occurring across 20-70 years in
a longitudinal cohort of 87 women with the FMR1
premutation

Jessica Klusek, [University
of South Carolina](#) (US)

Topic 3 FXS: *Diagnosis Delivery and Genetic Counseling*

- 12:00 The impact of the focus on negative aspects of FXS: delivering a diagnosis in a positive, helpful way. Jonathan Herring, [University of Oxford](#) (UK)
- 12:20 Specificities and difficulty of Fragile X genetic counselling in Arab Societies: Example of Morocco Siham Elalaoui, [Faculty of Medicine and Pharmacy of Rabat](#) (MA)
- 12:40 *Posters and lunch*

Topic 4 FXS: *Screening + Testing*

- 14:00 Preconceptional fragile X screening Matthias Nybro Smith (DK)
- 14:20 Newborn Screening for Fragile X (U.S.) Don Bailey, [RTI International](#) (US)
- 14:40 Newborn Screening for Fragile X (Australia) David Godler, [University of Melbourne](#) (AU)
- 15:00 Genetic testing for Fragile X — Challenges, experiences and results from University Clinical Centre Hospital in Gdańsk Karolina Śledzińska, [Medical University of Gdansk](#) (PL)
- 15:20 *Networking coffee and tea break*

Topic 5 FXS: *Biomarkers, Pathways, Outcome Measures, Medical Therapies/Trials*

- 15:50 Key scientific findings, therapeutic avenues to explore, some failures in past FX research Claudia Bagni, [University of Lausanne](#) (CH)
- 16:10 Ageing trajectories and dementia biomarkers in adults with FXS Hadassa Kwetsie, [Amsterdam UMC](#) (NL)
- 16:30 Nonsense mediated mRNA decay pathway and Fragile X Jozef Gecz, [University of Adelaide](#) (AU)
- 16:50 Getting personal in a CBD-trial: Individualized trial design and outcome measures Agnies van Eeghen, [Amsterdam UMC](#) (NL)

- 17:10 Current, approved, medical intervention, and what is emerging on horizon/clinical trials Andrew Stanfield, [University of Edinburgh](#) (UK)
- 17:30 *Break*
- 19:30 Conference Dinner

Friday 8 November, 2024

- 08:55 Conference welcome Fragile X International

Topic 6 FXS: Mosaicism

- 09:00 The role of allelic instability and mosaicism in premutation Flora Tassone, [MIND Institute, UC Davis](#) (US)
- 09:20 The importance and clinical role of somatic mosaicism in Fragile X Syndrome FT for Alessandra Murgia, [Pediatric Research Institute, Padova](#) (IT)
- 09:30 Impact of mosaicism on diagnostic, prognostic and screening applications David Godler, [University of Melbourne](#) (AU)

Topic 7 FXS: Development + Intervention

- 09:50 Early attention development as it relates to numeracy Gaia Scerif, [University of Oxford](#) (UK)
- 10:10 AI and gait analysis in the assessment of motor skills in FXS Zimi Sawacha, [University of Padova](#) (IT)
- 10:30 *Networking coffee and tea break*
- 11:00 Cognitive and environmental factors contributing to social strengths and challenges in people with FXS Katherine Ellis, [University College London](#) (UK)
- 11:20 Cognitive Behavioural Treatments in Adolescents and Young Adults with FXS Alice Montanaro, Bambino Gesù Children's Hospital; [University of Bari](#) (IT)
- 11:40 Social inhibition across early childhood in fragile X syndrome Abigail Hogan, [Arnold School of Public Health](#) (US)

12:00 Sleep problems in Fragile X Syndrome

Alexander Dück, [University Medical Centre Rostock](#) (DE)

Topic 8.1 FXS: Care – General

12:20 Fragile X Syndrome – What can we learn about health and healthcare from large-scale, routinely-acquired data in the United Kingdom?

Andrew McKechnie, [University of Edinburgh](#) (UK)

12:40 Holistic, person-centred care

Andrew Stanfield, [University of Edinburgh](#); Kirsten Johnson, [FraXI](#)

13:00 *Lunch*

Friday 8 November, 2024, afternoon

Common session with professionals and representatives from Fragile X International member associations joined together

14:00 Official opening: Welcome from Catalunya and FraXI Self-Advocate

FraXI and L'Associació Catalana de la Síndrome X Fràgil

Topic 8.2 FXS: Care – FX Centres

14:10 Multidisciplinary care at the FX Centre Barcelona

Ana Roche, [SJD Barcelona Children's Hospital](#) (ES)

14:25 The management of children with FXS: experience from a neuropsychiatric center in Italy

Paolo Alfieri, [Bambino Gesù Children's Hospital](#) (IT)

14:40 FXS and Adolescents/Young Adults: therapeutic education program for adolescents implemented in Lyon centre.

Vincent des Portes, [Mother and Child Hospital](#), Bron Cedex (FR)

14:55 FXS – Care at the Kinderzentrum Maulbronn

Ulrike Gaiser, [Children's Center Maulbronn](#) (DE)

15:10 Care at the FX centre in Padua

Zimi Sawacha, [University of Padova](#) (IT)

15:25 Questions

15:35 *Networking coffee and tea break*

Topic 9 FXS: *Living with Fragile X Syndrome*

- 16:00 The research called for by people living with FXS Gaia Scerif, [University of Oxford](#) (UK)
- 16:15 The Joys of Fragile X: Understanding the strengths of people with FXS Katherine Ellis, [University College London](#) (UK)
Jörg Richstein (DE)
- 16:30 *Change rooms*
- Parallel sessions**
- 16:35 Workshop on diagnosis (**Room: Sala Macaya**) Andre Rietman, [Sophia Children's Hospital](#); Bram Dierckx, [Erasmus University, Rotterdam](#) (NL)
- 16:35 FXTAS longitudinal registry to form a platform for research and increase our understanding of the natural history of FXTAS (**Room: Aula 1**) Sundus Alusi, [The Walton Centre, Liverpool](#) (UK)
- 17:15 Conclusions
- Time to network between professionals and family association delegates*
- 18:00 End