

# 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 biomarkers in FXTAS

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

09:30 Neuroimaging evidence of glymphatic system  
dysfunction in FXTAS

Andrea Elias, [Hospital  
Universitari Mútua de  
Terrassa](#) (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-Revenga,  
[Hospital Clinic Barcelona](#)  
(ES)

10:30 *Networking coffee and tea break*

## **Topic 2 *FXPAC: Other premutation issues***

11:00 Other FXPAC-Related Symptoms and Conditions

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

11:20 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***

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|-------|--|--|
| 11:40 | The impact of the focus on negative aspects of FXS: delivering a diagnosis in a positive, helpful way. | Jonathan Herring, <a href="#">University of Oxford</a> (UK)                    |
| 12:00 | Specificities and difficulty of Fragile X genetic counselling in Arab Societies: Example of Morocco    | Siham Elalaoui, <a href="#">Faculty of Medicine and Pharmacy of Rabat</a> (MA) |
| 12:20 | Discussion to end the morning sessions, areas for research   |  |
| 12:40 | <i>Posters and lunch</i>   |  |

### **Topic 4 FXS: *Screening + Testing***

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

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

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|-------|--|--|
| 15:50 | Key scientific findings, therapeutic avenues to explore, some failures in past FX research | Claudia Bagni, <a href="#">University of Lausanne</a> (CH) |
| 16:10 | Ageing trajectories and dementia biomarkers in adults with FXS                             | Hadassa Kwetsie,<br><a href="#">Amsterdam UMC</a> (NL)     |
| 16:30 | Nonsense mediated mRNA decay pathway and Fragile X   | Jozef Gecz, <a href="#">University of Adelaide</a> (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, [Hospital Parc Tauli](#) (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

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