

1st Fragile X International Congress

Thursday 7 November

| | |
|-------|-------------------------------|
| 08:15 | <i>Sign in and networking</i> |
|-------|-------------------------------|

| | | |
|-------|-----------------|--|
| 09:00 | Opening Welcome | Kirsten Johnson (FraXI), Pilar Elias (L'Associació Catalana de la Síndrome X Fràgil) |
|-------|-----------------|--|

Topic 1 FXPAC: FXTAS

| | | |
|-------|--|--------------------|
| 09:10 | Developing biomarkers in FXTAS (but also touching FXS) | Flora Tassone (US) |
| 09:30 | FXTAS in males: when the numbers don't add up | Mike Field (AU) |
| 09:50 | FXTAS and imaging in carriers | Andrea Elias (ES) |
| 10:10 | Care for patients with FXTAS | Sundus Alusi (UK) |

| | |
|-------|--|
| 10:30 | <i>Networking coffee and tea break</i> |
|-------|--|

Topic 2 FXPAC: FXPOI

| | | |
|-------|---|-----------------------------|
| 11:00 | The molecular basis of FXPOI | Laia Rodríguez-Reventa (ES) |
| 11:20 | The clinical aspects of women with FMR1 premutation | Shai Elizur (IL) |

Topic 3 FXPAC: Other premutation issues

| | | |
|-------|---|--------------|
| 11:40 | Autistic traits and mental health in females with the fragile X premutation | Jo Moss (UK) |
|-------|---|--------------|

| | | |
|-------|--|---------------------|
| 12:00 | Other FXPAC-Related Symptoms and Conditions | Dragana Protic (RS) |
| 12:20 | 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 (US) |
| 12:40 | <i>Posters, lunch & conference photo</i> | |

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 (US) |
| 14:40 | Newborn Screening for Fragile X (Australia) | David Godler (AU) |
| 15:00 | Genetic testing for Fragile X — Challenges, experiences and results from University Clinical Centre Hospital in Gdańsk | Karolina Śledzińska (PL) |
| 15:20 | <i>Networking coffee and tea break</i> | |

Topic 5 FXS: Diagnosis Delivery and Genetic Counseling

| | | |
|-------|--|-----------------------|
| 15:50 | The impact of the focus on negative aspects of FXS: delivering a diagnosis in a positive, helpful way. | Jonathan Herring (UK) |
| 16:10 | Specificity and Complexities of Genetic Counseling in a Conservative Society, example of Morocco | Siham Elalaoui (MA) |

Topic 6 FXS: Biomarkers, Pathways, Outcome Measures, Medical Therapies/Trials

| | | |
|-------|--|------------------------|
| 16:30 | Key scientific findings, therapeutic avenues to explore, some failures in past FX research (title TBC) | Claudia Bagni (CH) |
| 16:50 | Current, approved, medical intervention, and what is emerging on horizon/clinical trials | Andrew Stanfield (UK) |
| 17:10 | <i>Short comfort break</i> | |
| 17:20 | Getting personal in a CBD-trial: Individualized trial design and outcome measures | Agnies van Eeghen (NL) |
| 17:40 | Cognitive ageing and blood biomarkers for neurodegeneration in adults with FXS | Hadassa Kwetsie (NL) |
| 18:00 | Nonsense mediated mRNA decay pathway and Fragile X | Jozef Gecz (AU) |
| 18:20 | <i>Break</i> | |
| 20:00 | Catalan Dinner | |

Friday 8 November

Topic 7 FXS: Mosaicism

| | | |
|-------|--|--------------------------------------|
| 08:55 | Conference welcome | FraXI |
| 09:00 | The role of allelic instability and mosaicism in premutation | Flora Tassone (US) |
| 09:20 | The importance/clinical role of somatic mosaicism in Fragile X Syndrome | Alessandra Murgia (IT) |
| 09:40 | Impact of mosaicism on diagnostic, prognostic and screening applications | David Godler (AU) Mike Field (AU) |

Topic 8 FXS: Development + Intervention

| | | |
|-------|---|-------------------|
| 10:00 | Early attention development as it relates to numeracy | Gaia Scerif (UK) |
| 10:20 | AI and gait analysis in the assessment of motor skills in FXS | Zimi Sawacha (IT) |

| | | |
|-------|--|--|
| 10:40 | <i>Networking coffee and tea break</i> | |
|-------|--|--|

| | | |
|-------|--|----------------------|
| 11:10 | Cognitive and environmental factors contributing to social strengths and challenges in people with FXS | Katherine Ellis (UK) |
| 11:30 | Cognitive Behavioural Treatments in Adolescents with FXS | Alice Montanaro (IT) |
| 11:50 | Social fear and anxiety across early childhood in fragile X syndrome | Abigail Hogan (US) |
| 12:10 | Sleep problems in Fragile X Syndrome | Alexander Dück (DE) |

Topic 9.1 FXS: Care – General

| | | |
|-------|---|---|
| 12:30 | Fragile X Syndrome – What can we learn about health and healthcare from large-scale, routinely-acquired data in the United Kingdom? | Andrew McKechnie (UK) |
| 12:50 | Holistic, person-centred care | Andrew Stanfield (UK) Kirsten Johnson (UK) |

| | | |
|-------|--------------|--|
| 13:10 | <i>Lunch</i> | |
|-------|--------------|--|

Friday 8 November 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 9.2 FXS: Care – FX Centres

| | | |
|-------|---|-------------------------|
| 14:10 | Multidisciplinary care at the FX Centre Barcelona | Ana Roche (ES) |
| 14:25 | The management of children with FXS: experience from a neuropsychiatric center in Italy | Paolo Alfieri (IT) |
| 14:40 | FXS and Adolescents/Young Adults: therapeutic education program for adolescents implemented in Lyon centre. | Vincent des Portes (FR) |
| 14:55 | FXS – Care at the Kinderzentrum Maulbronn | Ulrike Gaiser (DE) |
| 15:10 | Care at the FX centre in Padua | Alessandra Murgia (IT) |
| 15:25 | Questions | |
| 15:35 | <i>Networking coffee and tea break</i> | |

Topic 10 Living with Fragile X Syndrome

| | | |
|-------|---|---|
| 16:00 | The research called for by people living with FXS | Gaia Scerif (UK) |
| 16:15 | The Joys of Fragile X: Understanding the strengths of people with FXS | Katherine Ellis (UK) Jörg Richstein (DE) |

| | | |
|-------|--|----------------------------------|
| 16:30 | <i>Change rooms</i> | |
| | Parallel sessions | |
| 16:35 | Workshop on diagnosis (Room: Sala Macaya) | Andre Rietman, Bram Dierckx (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 (UK) |
| 17:15 | Conclusions, thank yous | |
| | <i>Time to network between professionals and family association delegates</i> | |
| 18:00 | End | |