6 Months in Retrospect

Milestones, Memories and Moments to remember in FraXI's journey

www.fraxi.org

#Resolution4Rare

At the 78th World Health Assembly in Geneva, countries adopted the world's first ever first-ever WHO resolution recognizing rare diseases as global equity issues. This is a HUGE development in promoting health and social equity for those living with rare conditions worldwide!

Running for Fragile X!

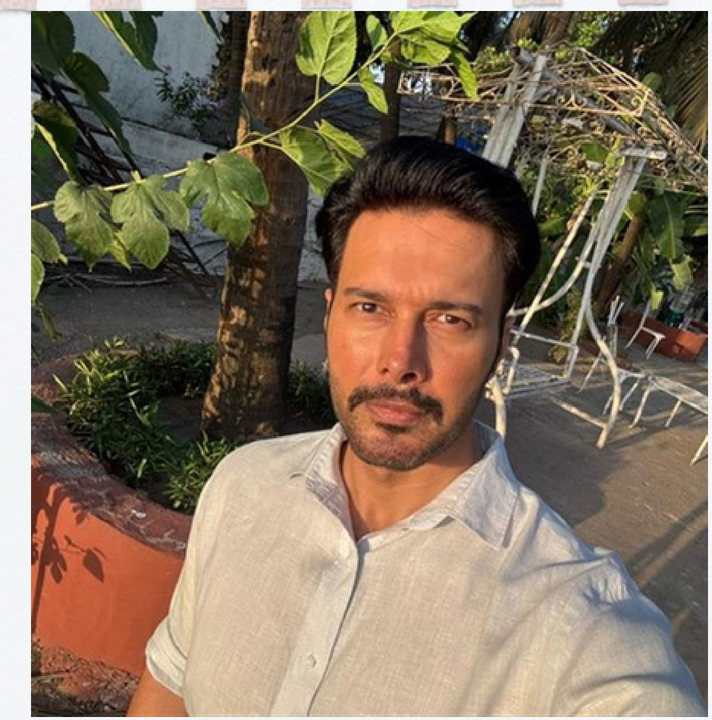
Bríd Quinn, who is a proud mum of two daughters living with Fragile X Syndrome, is taking on five half marathons across five European cities, all in the name of raising awareness and donations for FraXI. You can follow her journey on Instagram @runningthexforfragilex

A Star is Born!

Rishabh Jain is making history as the first actor living with FXS to star in a feature film! “Sitaare Zameen Par” is a Bollywood Production featuring an ensemble of extremely talented neurodivergent actors. It was released in theatres on the 20th of June 2025.

International Workshop on FXS

Experts, advocates and supporters working in the field of FXS awareness gathered at the International Workshop on FXS held in Padua, Italy! This workshop was organised in memory of Prof. Alessandra Murgia, a FraXI advisor and founder of the Padua Fragile X Centre of Expertise.Excellent insights on what we expect from research, increasing social inclusion and supporting families living with FXS were discussed.



FXS Short film premieres in Italy!

FraXI Full Member Fragile X Society India has collaborated with actor and director Rajniesh Duggal to produce the short film “Fragile” which raises awareness about FXS. The film had its premiere at the Ami Corti International Film Festival in Italy. We congratulate Shalini and her team for this amazing achievement!

FXS represented at TCD Summer School

Two young people from the Irish FX Society attended the summer school run by Trinity College Dublin's Trinity Centre for People with Intellectual Disabilities (TCPID). The programme consisted of a three-day workshop for people with intellectual disabilities who want to experience college life at Trinity College Dublin.

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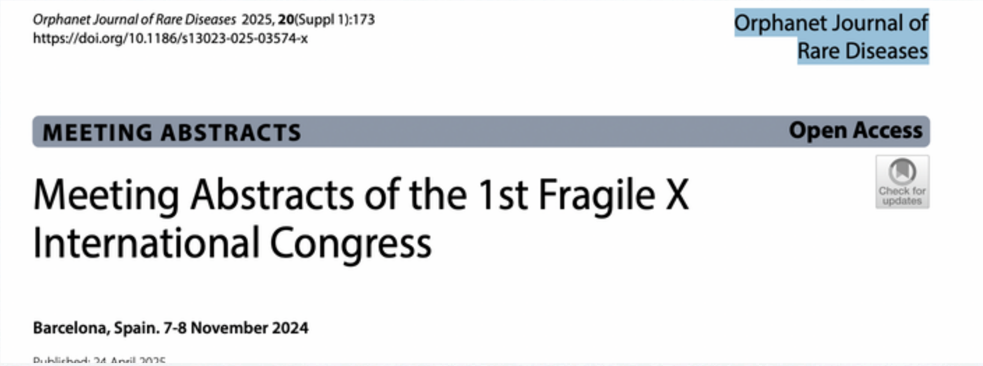
FraXI
Fathers' Group

DAD

We created a special group for dads of children who live with FXS to connect with and support each other. With the help of online meetings, this international group of fathers is learning every day that they are not alone, and that they are stronger together!



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Orphanet Journal of
Rare Diseases

MEETING ABSTRACTS **Open Access**

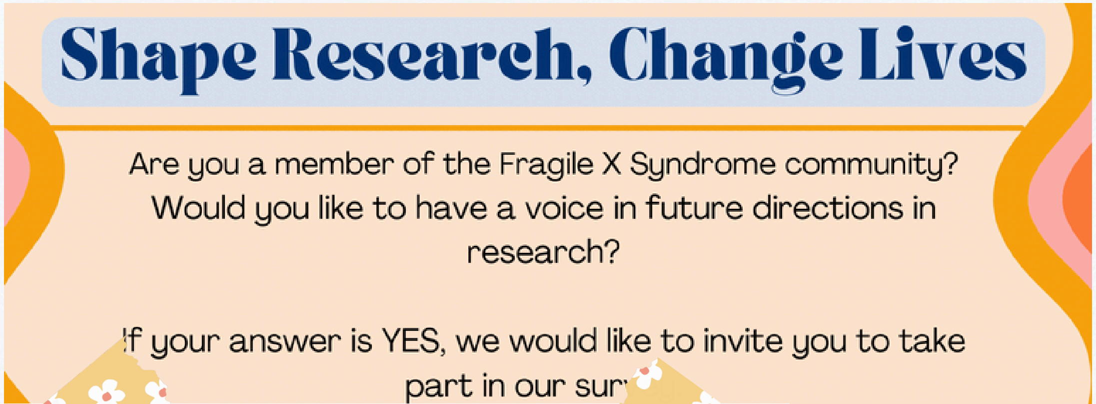
Meeting Abstracts of the 1st Fragile X International Congress

Barcelona, Spain. 7-8 November 2024

Published: 24 April 2025

Abstracts of our 1st Congress now published!

The abstracts of authors who presented their stellar work at FraXI's first congress in Barcelona last year have published in the Orphanet Journal of Rare Diseases. We are incredibly grateful to Oxford University's Prof. Gaia Scerif who made this possible.



Shape Research, Change Lives

Are you a member of the Fragile X Syndrome community? Would you like to have a voice in future directions in research?

If your answer is YES, we would like to invite you to take part in our sur

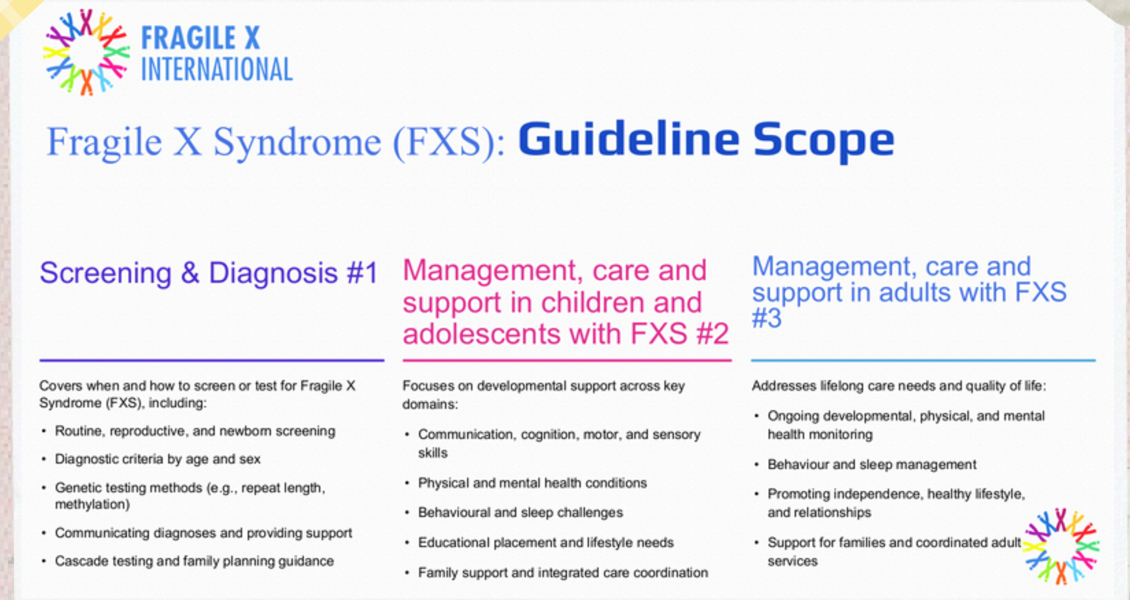
Shape Research, Change Lives

Setting priorities in genetic syndrome research

Shape Research, Change Lives:
Setting priorities in genetic syndrome research
June 2024

Launch of the "Shape Research, Change Lives Survey"

FraXI is proud to support the University of Surrey with its "Shape Research, Change Lives" project, which involves listening to people living with FXS to amplify the voices and hear how they wish to be supported through research.



FRAGILE X INTERNATIONAL

Fragile X Syndrome (FXS): **Guideline Scope**

Screening & Diagnosis #1

Covers when and how to screen or test for Fragile X Syndrome (FXS), including:

- Routine, reproductive, and newborn screening
- Diagnostic criteria by age and sex
- Genetic testing methods (e.g., repeat length, methylation)
- Communicating diagnoses and providing support
- Cascade testing and family planning guidance

Management, care and support in children and adolescents with FXS #2

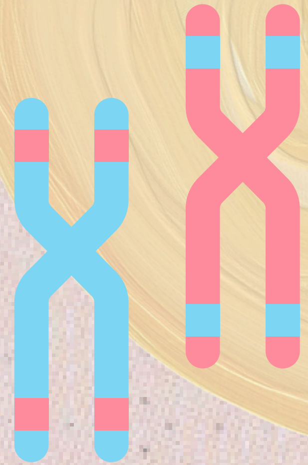
Focuses on developmental support across key domains:

- Communication, cognition, motor, and sensory skills
- Physical and mental health conditions
- Behavioural and sleep challenges
- Educational placement and lifestyle needs
- Family support and integrated care coordination

Management, care and support in adults with FXS #3

Addresses lifelong care needs and quality of life:

- Ongoing developmental, physical, and mental health monitoring
- Behaviour and sleep management
- Promoting independence, healthy lifestyle, and relationships
- Support for families and coordinated adult services



Development of FXS Guidelines

A writing group co-chaired by Dr Kirsten Johnson and Prof.Andrew Stanfield is developing important guidelines in the following areas: Screening and Diagnosis,Care and support in Children and adolescents with FXS, and FXS Guideline on Management, care and support in adults with FXS

Look at what we achieved together in half a year!

There is a lot of work to be done, and we are only just getting started. Thank you for being part of this very important journey.

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