

Getting the Most Out of the 13th International Fragile X Conference

A get-together for those who want to
maximize their learning
and minimize their exhaustion

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Welcome & Introductions

- NFXF Staff
- Board of Directors
- Florida Association

Layout of the Hotel

- Conference Rooms
- Childcare
- Food
- Pool

The Binder

- General Information
- At-A-Glance
- Day By Day
- Faculty Bios

Session Information

- General Sessions
- Family-Friendly vs Clinical Practice and/or Research vs Basic Science
- Choosing a Session
- What if there are two at the same time that you want to go to
- Tracks
- Medical Terms You My Hear
- Recording a session

Extra Activities

- Wednesday Meet - n - Greet
- Thursday Lunch Sessions
- Thursday Poster Session
- Friday Luncheon
- Friday Trip to the Marlins Baseball Game
- Saturday Banquet
- Sunday Panel Sessions
- Exhibit Hall

The People You Will Meet

- Doctors – M.D. and Ph.D.
- Other Professionals
- Parents & Other Family Members

Chance to Make New Friends and Re-connect

- People from your state, with the same age children
- People you have met on facebook
- Have fun and laugh

Other

- Coffee – each morning Thursday through Saturday
- Families For Fragile X Booth

Medical Terms You May Hear

- FXS - Intellectual Disability, MR, etc
- Other Medical Terms (see end of document)

The Emotional Aspect

- Can feel like a whirl wind
- There are a lot of people
- There is a lot of information
- Don't feel like you have to go to every session
- You will learn from others that you meet - in the hallways
- Much of the information will be available on-line
- There is hope everywhere!

Take Care of Yourself

- Eat well
- Get plenty of sleep
- Take time to relax

Questions or Concerns

- See Robby or Linda or Jayne
- Any of the NFXF Staff

- Enjoy!!

Medical Terms You May Hear

- **Fragile X-associated disorders (FXD):** The overall term describing the following 3 conditions: fragile X syndrome, fragile X-associated primary ovarian insufficiency, and fragile X-associated tremor/ataxia syndrome.
- **Fragile X Syndrome (FXS):** Individuals with over 200 CCG repeats; also considered to have a "full mutation."
- **Fragile X-associated Primary Ovarian Insufficiency (FXPOI):** Carriers of the FMR1 premutation (55-200 CCG repeats) are at risk for FXPOI, early menopause and ovarian dysfunction (decreased fertility) in general. Carriers of the full mutation do not appear to be at risk for these conditions.
- **Fragile X-associated Tremor/Ataxia Syndrome (FXTRAS):** A neurological disorder, involving progressively swaying and difficulty with walking and balance, and appears to specifically affect some older premutation carriers, generally grandfathers of children with fragile X syndrome. Although this neurological disorder occurs by a completely separate mechanism from fragile X syndrome, it is caused by the same gene.

Medical Terms You May Hear

- **Chromosomes:** Chromosomes are packages of genes that are passed from generation to generation. Most individuals have 46 chromosomes, two of which are sex chromosomes. In females, these are two X's; in males they are and X and Y. The fragile X gene is on the X chromosome, and is therefore said to be "X-linked."
- **Genes:** There are thousands of genes along the chromosomes and are composed of DNA; the fragile X gene is called FMR1.
- **DNA:** The chemical that makes up the genes. The FMR1 gene is defined by the number of repeats of a pattern of DNA called CCG repeats.

Medical Terms You May Hear

- **CGG Repeats:** The number of repetitions of CCG determines whether a person has a typical range, a grey zone, a premutation or carrier range, or is a full mutation.
- **Typical Range of CGG Repeats:** CGG repeats from 0 to 44
- **Grey Zone:** CGG repeats from 45-54
- **Premutation or Carrier:** Described in terms of the DNA code/CGG Repeat: 55-200 CCG repeats.
- **Full Mutation:** Individuals with over 200 CCG repeats and which causes fragile X syndrome

Medical Terms You May Hear

- **Fragile X Mental Retardation Protein (FMRP):** The protein synthesized by the FMR1 gene. The full mutation causes the gene to shut down and when this gene is turned off, the individual does not make FMRP. The lack of this specific protein causes fragile X syndrome.
- **Methylation:** The term used to describe the shutting down of the region of the FMR1 gene, thereby stopping the production of the FMRP.
- **mRNA:** The biochemical derived from DNA that takes the genetic message to where proteins are made; too much mRNA may cause FXTRAS.
- **Mosaicism:** When there are different sizes of the repeat expansion (CGG Repeats) in different cells. In other words, some of the cells in mosaicism carry a fully expanded repeat with more than 200 copies, while other cells carry a partially expanded repeat with 55-200 copies.
- **Knock-out mouse:** A strain of mice that has been bred to have the fragile X gene change; used in research to model what happens in the human body.
- **mGluR:** Metabotropic Glutamate Receptors (mGluRs) which function in excitatory neurotransmission. Interestingly, mGluR activation also leads to protein synthesis, and the "mGluR Theory" states that symptoms of FXS are due to an exaggeration of this protein synthesis, i.e., FMRP acts as a "brake" on mGluR-mediated protein synthesis.

Medical Terms You May Hear

- **Fragile X Clinical & Research Consortium (FXCRC):** was initiated by the National Fragile X Foundation in 2006 and is supported in part by a grant from the Centers for Disease Control.
- **Fragile X Clinic:** A fragile X clinic provides individuals and families affected by one or more of the Fragile X-associated Disorders with a comprehensive evaluation and treatment recommendations.
<http://www.fragilex.org/treatment-intervention/fragile-x-clinic/>
- **LINKS:** LINKing Individuals in Knowledge and Support - NFXF Parent Support Groups -
<http://www.fragilex.org/community/links-support-network/>

Medical Terms You May Hear

- ▣ **Clinical Trials:** See this link on the NFXF website—
<https://www.fragilex.org/2012/multimedia/podcast/questions-answers-about-clinical-trials/>
- ▣ **Minocycline:** An antibiotic medication. The Bilousova study demonstrated that minocycline lowered matrix metalloproteinase 9 (MMP9) levels in the KO mouse which are elevated in the absence of FMRP.
<http://www.fragilex.org/2009/research/previoursly-funded/a-2009-blind-randomized-controlled-cross-over-trial-of-minocycline-in-children-with-fragile-x-syndrome/>
- ▣ **STX209 (arbaclofen):** An investigational medicine for people with fragile X syndrome to improve social function.
<http://www.fragilex.org/2012/research/opportunities-for-families/researchers-need-your-help-for-new-targeted-treatment-for-fragile-x-syndrome/>