The Top Ten Things You Should Know About Fragile X Syndrome

By Mary Beth Langan and Sally Nantais

1. It's **genetic**.
2. If a woman is a carrier (55-200 CGG repeats) she has a 50/50 chance of passing it on to her son(s) or daughter(s). **1 in 151** women are carriers. In the gray zone, defined as 45-54 CGG repeats, prevalence is 1 in 35 for females. **
3. If a man is a carrier he will pass it only to his daughter(s), and they will only be carriers. **1 in 468** men are carriers. In the gray zone prevalence is 1 in 42 for males. **
4. Fragile X Syndrome (FXS) does **NOT** discriminate; it doesn't care which ethnic group you belong to.
5. Fragile X Syndrome is a **spectrum disorder**. Symptoms may vary from mild learning disabilities (including shyness and social anxiety) to severe cognitive impairment (mental retardation).
6. **Fragile X-Associated Primary Ovarian Insufficiency (FXPOI)**, more commonly known as early menopause, is a condition that affects 20-28% of the female FXS carrier population.
7. **Fragile X-associated Tremor/Ataxia Syndrome (FXTAS**), discovered in 2001, is a neurological disorder that can involve tremors, balance irregularities, difficulty walking and dementia which sadly is often misdiagnosed as Parkinson's and/or Alzheimer's. This condition is present in some older FXS carriers (typically after the age of fifty); usually in males but FXTAS can also affect female carriers.
8. There are minor physical traits noted in many persons with Fragile X Syndrome, but not in all. These are traits which may also be present within the typical population, nothing unique which would necessarily indicate FXS testing is necessary for your child.
9. When testing for Fragile X Syndrome, it is critical that the correct tests are ordered – the Fragile X DNA (Southern Blot) and Polymerase Chain Reaction (PCR) tests, (also known as the FMR1 DNA test) which is 99% accurate.
    - Test typically costs between $200 and $600, and takes about two weeks for results.
    - Inaccurate results occur far too often with the generic chromosomal panel.
    - Test for FXS to obtain a diagnosis or to **rule it OUT**. If you don't have what may be the correct diagnosis of FXS, then you will never be aware of improved treatments or the cure when it's found.
    - ***New*** **-** Quest Diagnostics has created a new test which is as reliable as the FMR1 DNA test and only requires a drop of blood, being used in newborn screening studies at this time.
10. Where to go for more information on Fragile X:
    - FragileX.org - The National Fragile X Foundation
    - FRAXA.org – FRAXA Research Foundation
    - youtube.com/watch?v=BgcQi0bbaJQ - First Down Towards a Cure
    - youtube.com/watch?v=-6-J_YcVRi4 Fragile X – Hitting the Mark
    - content.time.com/time/magazine/article/0,9171,1818268,00.html – Fragile X: Unraveling Autism's Secrets
    - livingwithfragilex.com - Living with Fragile X
    - cdc.gov/ncbddd/fxs/video/FragileX-4.html - What causes Fragile X

** SeltzerMM, Baker MW, Hong J, Maenner M, Greenberg J, Mandel D. 2012. Prevalence of CGG expansions of the FMR1 gene in a US population-based sample. American Journal of Medical Genetics, October 2011,

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