

**COLUMN NAME:** X-tra Special Advice

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**HEADLINE:** Familiar Faces... but... Fragile X?

**COPY:** Each one of us has the Fragile X (FMR1) gene on our X chromosome.

We all have a number of CGG repeats on that gene.

Some of us have a typical range of repeats on our X.

Some of us have 50 to one-thousand-plus repeats. Those repeats can affect us all in many different ways that may seem unrelated at first glance.

There are those with numbers that fall within a range that deem them a premutation carrier of Fragile X and they may have certain symptoms affecting them on a daily basis throughout their lives or as they age. There are those with the full mutation of Fragile X, who are said to be affected by Fragile X Syndrome.

But what does someone affected by the Fragile X gene look like?

Consider these possible faces of Fragile X:

- a 5-year-old boy with autism and social anxiety
- a 44-year-old woman with an autoimmune disorder such as Lupus, Multiple Sclerosis or Rheumatoid Arthritis
- a 7-year-old girl with selective mutism and a math learning disability
- a 50-year-old woman with near-constant muscle pain
- a 4-year-old boy who is beginning to speak and has severe sensory issues
- an 8-year-old boy with PDD-NOS and a very selective diet
- a 76-year-old man with Alzheimer's Disease and weakness in his legs
- a 12-year-old boy with autism and a seizure disorder
- a 4-year-old girl with strabismus (an eye muscle issue)
- a 37-year-old woman who is post-menopausal and struggling with anxiety
- an 80-year-old man with Parkinson's Disease
- a 3-year-old girl who can only verbalize a few words
- a 40-year-old woman who has struggled with infertility for ten years
- an 8-year-old boy who has detailed obsessions, attention issues, is not reading at grade level and wasn't toilet trained until age 4

Mary Lee Shelton of Mesilla, New Mexico says "to this day we are questioned by professionals outside the Fragile X world if we are sure our son has Fragile X. I just know there are more boys like our son! I think it was a lucky break that he was diagnosed correctly. His neurologist was stunned. Fragile X professionals aren't surprised by his diagnosis because they see so many persons who are

affected all over the spectrum. But I think local doctors only see the severely affected and miss many others that should have the blood test.”

Many professionals still use the chromosome analysis to rule out Fragile X. Our Andrew is a perfect example of why that is not the most accurate test — he tested negative with it, yet positive with the less expensive Fragile X test. We’re happy we saw a geneticist who orders the Fragile X test when a diagnosis of autism is being considered. At that point, we were saying “Fragile What?” as we knew nothing of the genetic disorder.

Shelton also shared that “the doctor who diagnosed our son told me he had a professor in medical school who told him when anyone had learning difficulties to always test them for Fragile X. He cited a case of a man who was in the army who was having some kind of problem and this professor had done the Fragile X test on him and it came back positive. Even he was shocked at this, but used it as an example to encourage more Fragile X testing.”

Accurate diagnoses can help with accurate treatment and support. For example, when better treatment for Fragile X Syndrome is soon determined — an actual cure may even be in reach — your child will need an accurate diagnosis for you to have access to such information. If a cure for FXTAS is found, your father or mother or you will need to know they have FXTAS and not a misdiagnosis of Parkinson’s or Alzheimer’s Disease.

We are one year away from 800 family members and professionals coming together in Dearborn, Michigan, for the 12<sup>th</sup> International Fragile X Conference from July 21-25, 2010.

For that reason — among others — now is a great time for those in Michigan to be tested. The world’s Fragile X gurus will be in your backyard next year. They will be presenting scientific and family-friendly sessions to help those affected by all of the Fragile X-associated Disorders. As you can see, Fragile X has quite a few possible physical and behavioral characteristics and we haven’t even mentioned them all.

Do you see the faces of some of your family in the above list?

Take a good look, in honor of National Fragile X Awareness Day, on July 22.

*For more on Fragile X-associated Disorders, including other possible characteristics due to the Fragile X gene, plus accurate testing information to share with your medical professional, visit [fragilex.org](http://fragilex.org).*

*Grosse Pointe residents Theodore G. Coutilish and Mary Beth Langan created this column to share experiences from their journey as parents of a child with Fragile X Syndrome. Send your questions or comments to [mblangan@hotmail.com](mailto:mblangan@hotmail.com).*

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