
Fragile Syndrome

Survey of Developmental Disabilities

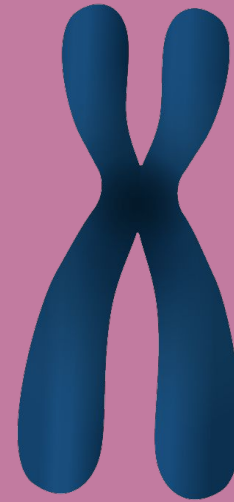
HSDD 5000

By: Carina Bergouignan

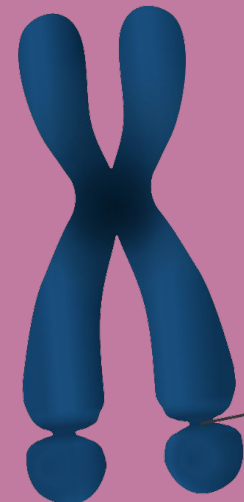


- Fragile X Syndrome is also known as FXS and Martin-Bell Syndrome
- Fragile X Syndrome is an intellectual disability defined as “an inherited condition characterized by an X chromosome that is abnormally susceptible to damage, especially by folic acid deficiency. Affected individuals tend to be mentally handicapped.”
- Fragile X Syndrome is caused by a genetic disorder. “FXS is caused by changes in the fragile X mental retardation 1 (FMR1) gene. The FMR1 gene usually makes a protein called fragile X mental retardation protein (FMRP) FMRP is needed for normal brain development. People who have FXS do not make this protein. People who have other fragile X- associated disorder have changes in their FMR1 gene but usually make some of the protein.” - Center of Disease Control and Prevention (CDC)

Fragile X Syndrome



Normal X chromosome

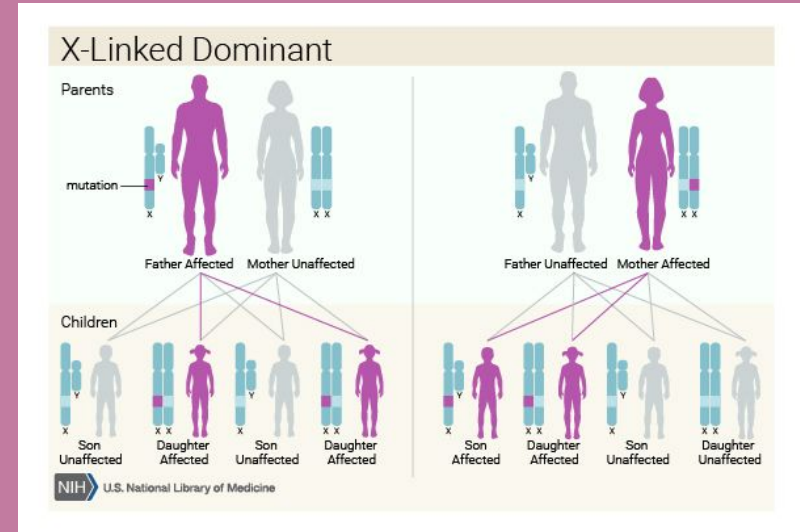


fragile site

Fragile-X chromosome

Fragile X Syndrome continued...

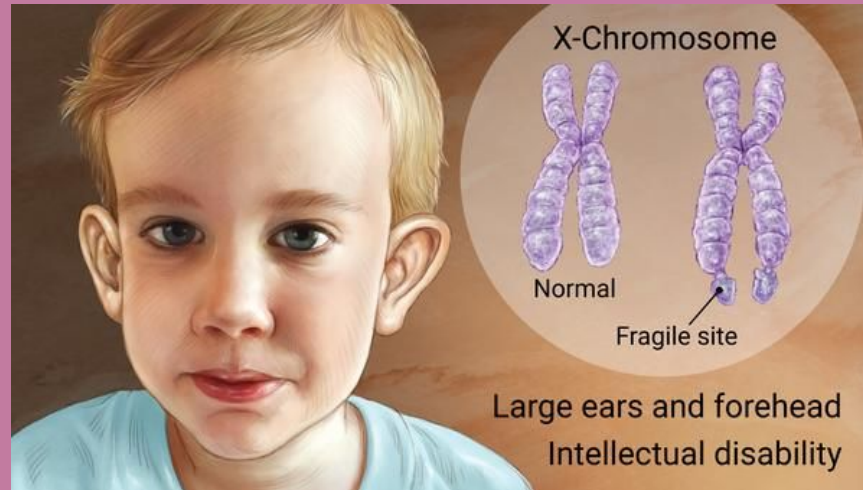
- 1 in 4,000 males and 1 in 8,000 females have Fragile X Syndrome
- There are fewer than 200,000 fragile X syndrome cases a year
- From the time Fragile X Syndrome begins life expectancy ranges 5 to 25 years
- Any individual with fragile X syndrome who has a FMR1 gene with 5 to 44 CGG repeat is very unlikely to pass the gene of fragile X syndrome on to his or her children.
- Explanation of Fragile X Syndrome:
https://www.youtube.com/watch?v=9_PEW6ocSNs



<https://my.clevelandclinic.org/health/articles/fragile-x-syndrome>
<https://www.counsyl.com/services/foresight/diseases/fragile-x-syndrome/>

Possible Fragile X Syndrome Physical Characteristics

- Normal Structure
- Long face
- Large forehead
- Large ears
- Tendency to squint
- Cross eyed
- Flat feet
- Hollow chest
- Scoliosis
- Prominent jaw
- Teeth crowding
- Murmurs
- Mitral valve prolapse
- Soft skin



<https://my.clevelandclinic.org/health/articles/fragile-x-syndrome>
<https://sunbio30b.wikispaces.com/Fragile+X+Syndrome+-+Keely+%26+Bishop>

Fragile X Syndrome Signs and Symptoms

- Hand flapping
- Difficulty learning any skills
- Anxiety
- Social issues
- Hyperactivity
- Speaking without thinking
- Autistic like qualities
- Gross motor delays
- Speech Delays
- Seizures (in 10%)
- Fragile X Syndrome causes intellectual disabilities. Both males and females can be affected. Females tend to have less aggressive symptoms.



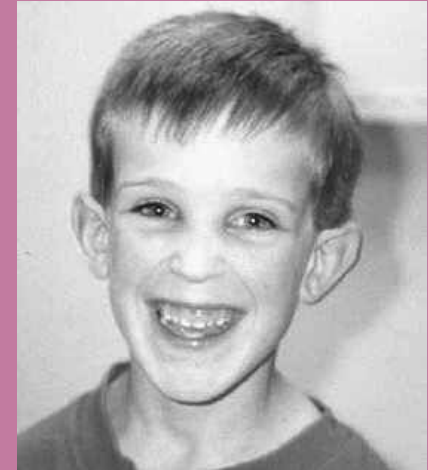
Fragile X Syndrome Diagnosis

- Newborn screening
- CBC
- Urine test
- MRI
- Fragile X can be determined before birth if your partner is a carrier through prenatal test like amniocentesis and chorionic villus sampling (CVS)



Fragile X Syndrome Treatments

- Therapy such as Behavior Therapy, speech therapy, occupational therapy, and physical therapy
- Medications such as sedatives, muscle relaxants, vitamins and Cognition enhancers
- Seek additional treatments from medical professionals such as pediatricians, medical geneticist, clinical psychologist and neurologist



Fragile X Syndrome Impacts

- “Parents were asked whether they felt that their child was bullied more than other children. Though the majority of parents did not feel that their child was bullied more, it was concerning that just under a third of parents of boys (27%) and over a third of parents of girls (38%) felt that their child was a victim of increased bullying. However, out of the four syndrome groups in this study, the children with fragile X had the lowest levels of reported bullying.” - Colin Reily (The Impact of Fragile X Syndrome on the Family)
- Verbal bullying was the most common though physical bullying still occurs
- “These reports are concerning and highlight the need for further education for others to promote understanding and acceptance, as well as support for those children experiencing bullying.” - Colin Reily (The Impact of Fragile X Syndrome on the Family)



<https://nobullying.com/fragile-x-syndrome/>

<http://www.fragilex.org.uk/single-post/2016/03/31/The-Impact-of-Fragile-X-Syndrome-on-the-Family>

Fragile X Syndrome Strengths

- Visual learning
- Imitating behaviors
- Long term memory
- Friendly
- Good sense of humor
- Being part of concrete task

Achievements possible to attain for those with Fragile X Syndrome include:

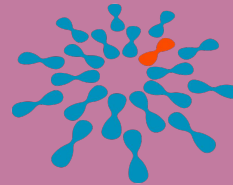
- Driving
- Working independently
- Living alone
- Living in assisted housing
- Education with accommodations or modifications
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Fragile X Syndrome Support Systems and Resources

- The National Fragile X Foundation: has public policy, advocacy, resources and treatment for those with the condition
www.fragilex.org
- FRAXA Research foundation: has given over \$21 million to start various clinical trials, find new treatments, and hopefully a cure
FRAXA.org
- Fragile X Registry: is used as a database for people who want to be kept up to date on new information and studies regarding fragile X syndrome
<https://fragilexregistry.com>

Fragile X Syndrome Support Systems and Resources continued ...

- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/>
- Families and Advocates Partnership for Education (FAPE)
<http://www.fape.org/>
- Parent Advocacy Coalition
<http://www.pacer.org/>
- Parents Helping Parent Resources, Links, Technology and more
<http://php.com/>



Fragile X Syndrome Support Systems and Resources continued ...

- Sensory Resource INC.
<https://www.sensoryresources.com>
- Carolina Fragile X Project
<http://fpg.unc.edu>
- The M.I.N.D. Institute
<http://mindinstitute.ucdmc.ucdavis.edu/>
- Living with Fragile X
<http://www.thinkmediaonline.com/fragilex.html>



Fragile X Syndrome Support Systems and Resources continued ...

- The Development of Fragile X Resource Centre
<http://www.developmentalfx.org/>
- Special Education Law & Advocacy
<http://www.wrightslaw.com>
- AAIDD American Association on Intellectual Disabilities
<http://www.aamr.org/>



Resources

- <https://www.cdc.gov/ncbddd/fxs/facts.html>
 - <https://my.clevelandclinic.org/health/articles/fragile-x-syndrome>
 - <https://www.counsyl.com/services/>
 - <https://my.clevelandclinic.org/health/articles/fragile-x-syndrome>
 - <https://sunbio30b.wikispaces.com/Fragile+X+Syndrome+-+Keely+%26+Bishop>
 - <https://www.nichd.nih.gov/health/topics/fragilex/conditioninfo/Pages/diagnosed.aspx>
 - <http://www.fragilex.org.uk/single-post/2016/03/31/The-Impact-of-Fragile-X-Syndrome-on-the-Family>
 - <https://fragilex.org.au/what-is-fragile-x/future-and-strengths/>
 - FRAXA.org
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Resources continued...

- <https://fragilexregistry.com>
 - <https://www.rarediseases.org/>
 - <http://www.fape.org/>
 - <http://www.pacer.org/>
 - <http://php.com/>
 - <https://www.sensoryresources.com>
 - <http://fpg.unc.edu>
 - <http://mindinstitute.ucdmc.ucdavis.edu/>
 - <http://www.thinkmediaonline.com/fragilex.html>
 - <http://www.developmentalfx.org/>
 - <http://www.wrightslaw.com>
 - <http://www.aamr.org/>
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