

Fast Facts

Fragile X Syndrome

- Genetic condition
- Affected individual makes too little or none of the FMR protein
- Protein helps nerve cells communicate and is used by the brain to develop normally
- Gene responsible for this protein is the FMR1 gene and is on the X chromosome
- Boys are more likely to have Fragile X than girls since they have only one X chromosome because girls can have one X chromosome with the defect and the other be fine
- Boys are also more likely to have more severe symptoms
- Individuals with the condition also have more copies than usual of a specific DNA segment (CGG). For most people, this segment repeats 5 – 40 times. In individuals with Fragile X, it repeats over 200 times
- Research has shown that the more times the DNA segment repeats, the worse the symptoms
- Some people inherit the disorder but don't have symptoms; they're known as carriers

<i>Symptoms</i>	<i>Treatment</i>	<i>Prevention</i>
<p><i>Physical:</i></p> <ul style="list-style-type: none"> -A large head -A long, narrow face -Large ears -A large forehead and chin -Loose joints -Flat feet -Enlarged testicles (after puberty) <p><i>Psychological:</i></p> <ul style="list-style-type: none"> -Trouble learning skills (sitting, crawling, or walking) -Problems with language and speech -Hand-flapping -Not making eye contact -Temper tantrums -Poor impulse control -Anxiety -Extreme sensitivity to light or sound -Hyperactivity -Trouble paying attention -Aggressive and self-destructive behavior (boys) 	<p><i>Medicines:</i></p> <ul style="list-style-type: none"> -Prevent seizures -Manage ADD symptoms -Treat other behavior problems <p><i>Therapy:</i></p> <ul style="list-style-type: none"> -Behavior -Occupational -Speech/Language 	<ul style="list-style-type: none"> -Get diagnosis early to prevent long-term negative impacts

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