GENE Seeker

You know the risk of your child being a carrier of a genetic disease.

Fragile X syndrome

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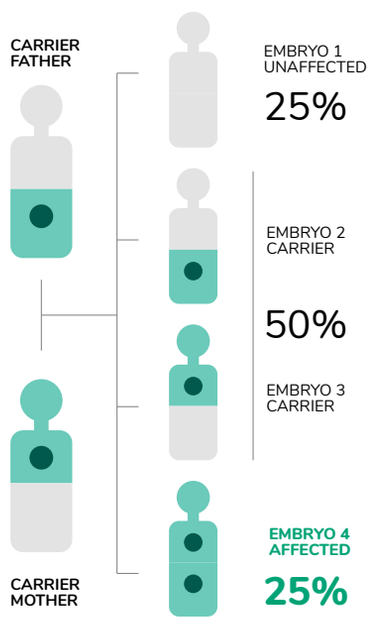
What is Fragile X syndrome?

A rare genetic disease associated with mild to severe intellectual deficit that may be associated with behavioral disorders and characteristic physical features including a high forehead, prominent and large ears, hyperextensible finger joints, flat feet with pronation and, in adolescent and adult males, macroorchidism.

Fragile X syndrome (FXS) presents with a variable clinical phenotype. In males, the disease presents during childhood with delayed developmental milestones. Intellectual deficit can be of variable severity and may include problems with working and short-term memory, executive function, language, mathematics and visuospatial abilities. Behavioral anomalies can be mild (e.g. anxiety, mood instability) to severe (e.g. aggressive behavior, autism). Autistic-like behavior can include hand flapping, poor eye contact, hand biting, gaze avoidance, social phobia, social and communication deficits and tactile defensiveness. In females, intellectual and behavioral disorders are typically mild and usually consist of shyness, social anxiety, and mild learning problems with a normal IQ, although 25% of girls have an IQ less than 70. Attention deficit hyperactivity disorder (ADHD) is present in over 89% of males and 30% of females and behavioral disinhibition is very common. Recurrent otitis (60%) and seizures (16 to 20%) can also be observed.

Management is symptom-based and requires a multidisciplinary approach. Speech, physical and sensory integration therapy as well as individualized educational plans and behavioral interventions may be combined with medication, such as stimulants for attention deficit-hyperactivity disorder; selective serotonin reuptake inhibitors (SSRIs) for anxiety, depression, obsessive-compulsive disorder; and atypical antipsychotic agents for self-injury and aggressive behaviors. New targeted treatments for FXS are being studied.



What is the next step if I'm a carrier of Fragile X syndrome?

If you are found to be a carrier of Fragile X syndrome, it is important that your partner be tested for the same genetic disorder.

What if my partner is not a carrier?

If your partner's test for Fragile X syndromenegative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Fragile X syndrome?

When both parents are carriers of Fragile X syndrome, the probability of having a child with Fragile X syndrome is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.

If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

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