



World Fragile X Day

[Source: IE](#)

Why in News?

The World Fragile X Awareness Day is marked on 22nd July every year, started in 2021 to raise awareness about the rare genetic illness, **Fragile X or Martin-Bell syndrome**.

What is Fragile X or Martin-Bell Syndrome?

▪ About:

- Fragile X syndrome (FXS) is an **inherited genetic disease passed down from parents to children** that causes intellectual and developmental disabilities.
- FXS is the most common **hereditary cause of mental disability** in boys. It affects 1 in 4,000 boys.
 - It's less common in girls, affecting about 1 in every 8,000. Boys usually have more severe symptoms than girls.
- People with FXS usually experience a range of developmental and learning problems.
- The disease is a **chronic or lifelong condition**. Only some people with FXS are able to **live independently**.

▪ Causes:

- FXS is caused by a defect in the FMR1 gene located on the **X chromosome**.
- The FMR1 (Fragile X Mental Retardation 1 gene) gene is located on the X chromosome in humans. It is responsible for **producing a protein called FMRP (Fragile X Mental Retardation Protein)**, which plays a crucial role in normal brain development and function.

▪ Risks:

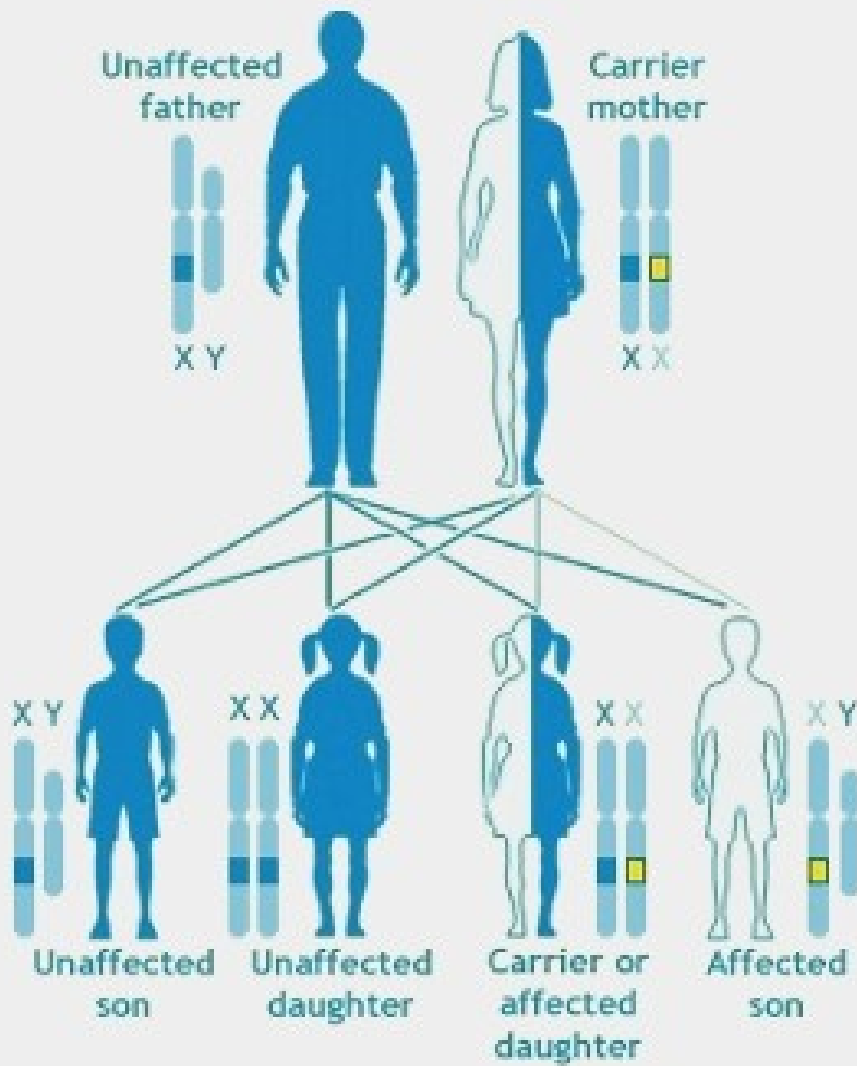
- Women who are carriers are at an increased risk for premature menopause, or menopause that starts before the age of 40.
- Men who are carriers are at increased risk for a condition known as [Fragile X Tremor Ataxia Syndrome \(FXTAS\)](#).
 - The disorder results in **progressive cerebellar ataxia**, action tremor, Parkinsonism and cognitive decline.
- It also can lead to **difficulty with balance and walking**. Male carriers may also be at an **increased risk for [Dementia](#)**.

▪ Linked with Inheritance:

- Women who carry **Fragile X have a 50% chance** of passing the mutated gene to **each of her children**. If she passes the affected gene, her children **will either be carriers, or they will have Fragile X syndrome**.
- Men who carry Fragile X will pass the **premutation to all their daughters** but none of their sons. These daughters are **carriers, but they do not have Fragile X syndrome**.

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X-Linked Inheritance in Fragile X Syndrome



adapted from US National Library of Medicine