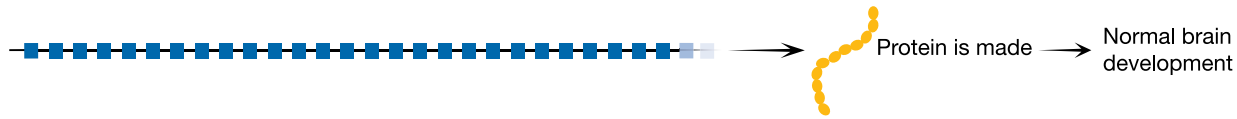


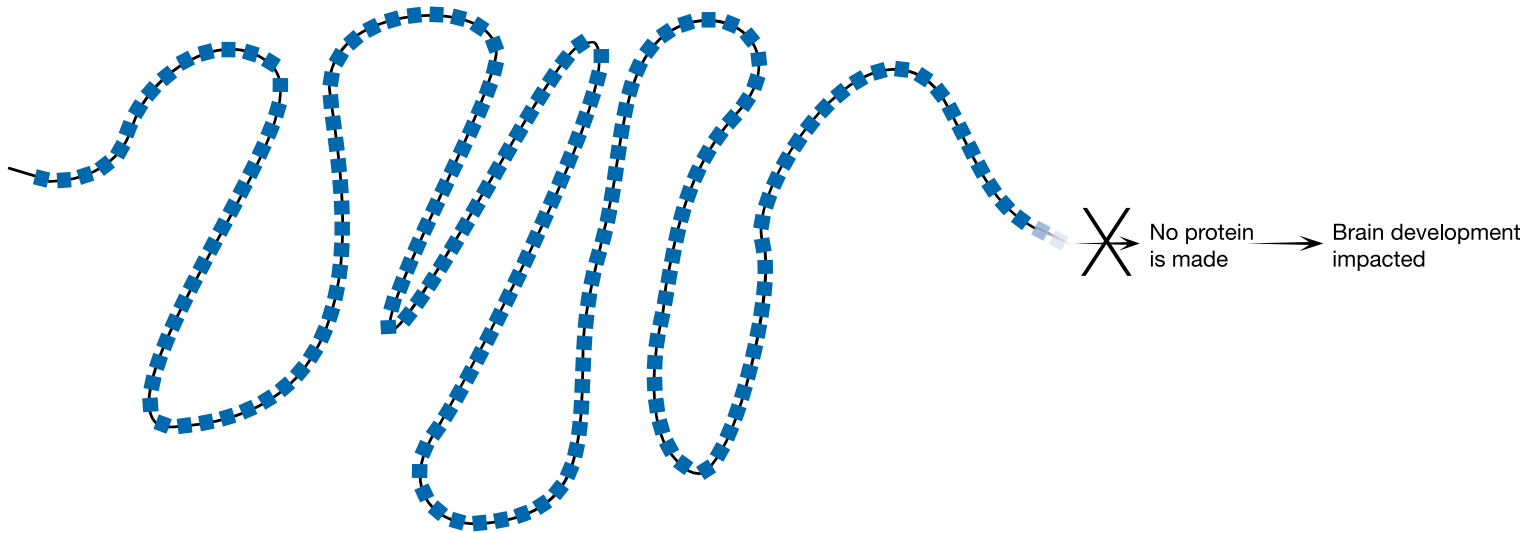
Fragile X Syndrome

CGG Repeat Expansion in the Fragile X Gene

Normal Fragile X Gene (<45 CGG)



Expanded Fragile X Gene (>200 CGG)



■ = 1 CGG repeat

● = Protein

Common Features of Fragile X Syndrome

- Mild to severe cognitive delays
- Seizures
- Behavioral differences such as autism and hand flapping
- Characteristic facial features such as large ears and a long face



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Fragile X Testing

Fragile X testing is appropriate for*:

- individuals (male or female) with mental retardation, developmental delay, or autism.
- individuals (male or female) with late onset intention tremor and cerebellar ataxia of unknown origin, especially if they have a family history of movement disorders, Fragile X syndrome, or undiagnosed mental retardation.
- women with a family history of Fragile X-related disorders, unexplained mental retardation or developmental delay, autism, or premature ovarian insufficiency.
- women with personal history of ovarian insufficiency or failure, or an elevated follicle-stimulating hormone (FSH) level before age 40 years without a known cause.
- women with a request for Fragile X carrier screening, regardless of family history after genetic counseling about the risks, benefits, and limitations of screening.

*ACOG Committee Opinion No. 469: Carrier screening for Fragile X syndrome. *Obstetrics & Gynecology* 2010; Oct: 116(4): 1008-10.

Sherman, et al. Fragile X syndrome: diagnostic and carrier testing. *Genetics in Medicine*; Oct: 7(8): 584-7.

FMR1-Related Disorders†

Fragile X Syndrome: a syndrome characterized by distinctive facial features such as large and/or protruding ears, a long face, prominent forehead, mandibular prognathism, strabismus, and high arched palate with occasional cleft palate; hyperflexible joints; macroorchism; seizures; borderline to severe intellectual disability, autism, or learning disabilities; and behaviors like attention deficits, hyperactivity, anxiety, hand flapping, etc.

Fragile X-associated tremor/ataxia syndrome (FXTAS): a neurological condition more common in men over the age of 50 who carry *FMR1* premutation alleles. Clinical features include intention tremor, cerebellar ataxia, Parkinsonism, and peripheral neuropathy.

Fragile X-associated premature ovarian insufficiency (FXPOI): early-onset (under the age of 40) menopause more common for women who carry *FMR1* premutation alleles.

†McConkie-Rosell A et al. Genetic counseling for Fragile X syndrome: updated recommendations of the national society of genetic counselors. *Journal of Genetic Counseling* 2005; 14(4):249-70.

Sherman S et al. Fragile X syndrome: diagnostic and carrier testing. *Genetics in Medicine* 2005; Oct: 7(8): 584-7.