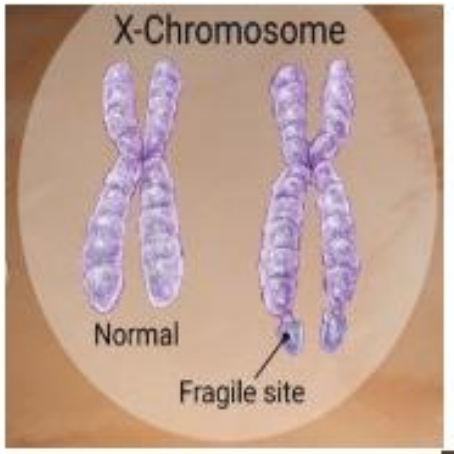
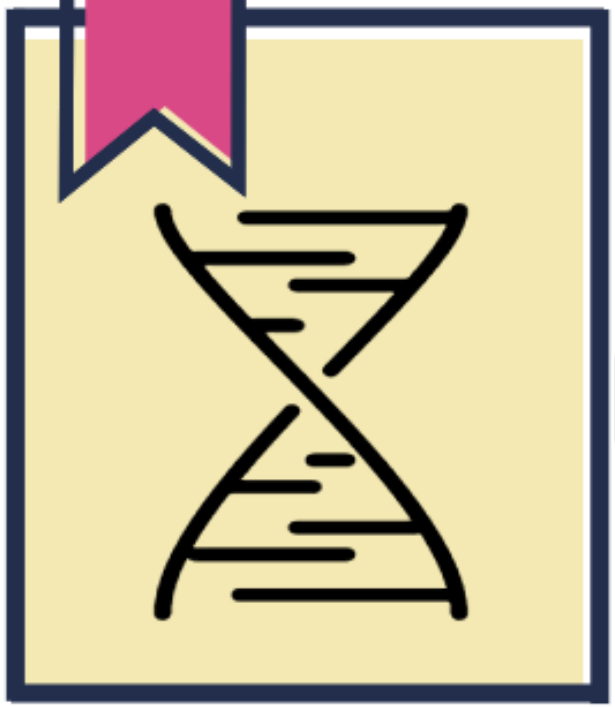
Women's Care
FLORIDA



FRAGILE X SYNDROME

Fragile X syndrome is associated with intellectual disability in males, characteristic appearance (large head, long face, prominent forehead and chin, protruding ears), joint laxity
Males are more severely affected than females

Fragile X syndrome is X-linked
If a woman is a carrier, her partner does not need to be tested



Repeat Size and Carrier Type

- 29-44 CGG repeats: Normal, not a carrier
- 45-54 CGG repeats: Fragile X intermediate carrier
- 55-200 CGG repeats: Fragile X pre-mutation carrier
  - Number of AGG repeats effects likelihood of expanding into full mutation in a generation
- >200 CGG repeats: Fragile X full mutation

Clinical Features Associated with Carrier Status

No clinical signs or symptoms

20% chance of premature ovarian failure (occurring in the 20s/30s)
8-16% chance of Fragile X-associated tremor-ataxia syndrome

Females with a full mutation can be as severely affected as males.

Interesting Facts

- Natera and Counsyl automatically reflex to AGG repeats when pre-mutation carrier is detected
- Prenatal testing through CVS or amniocentesis can be done ONLY if patient is a confirmed pre-mutation carrier
- If a patient tests negative, this reduces but does not eliminate the chance that they could have a child with the disease
- If a patient tests positive, family members should be informed since they are at a 50% chance to test positive