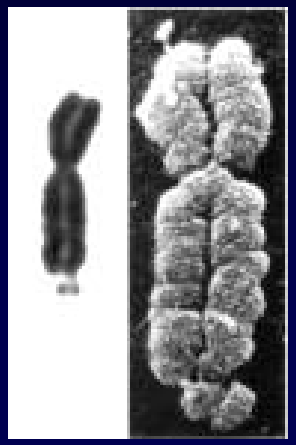
Fragile X Syndrome

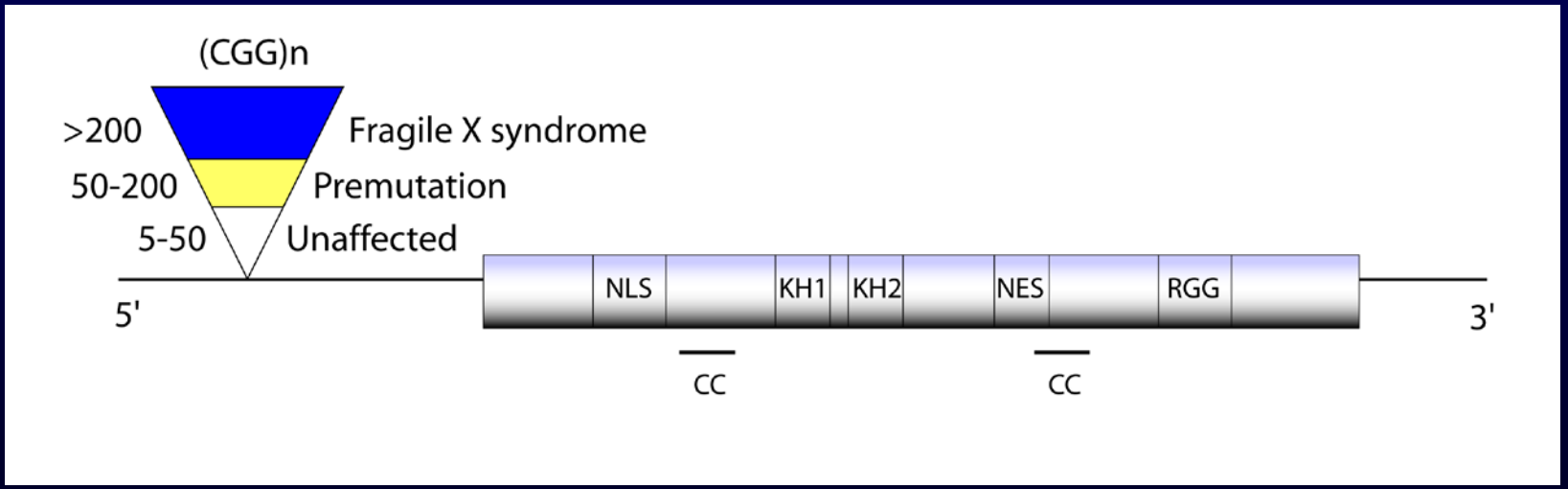
- Moderate intellectual deficiency (ID)
- Relatively large cranium
- Large ears
- Long face, prominent jaw
- Hyperextensibility
- Macroorchidism: pubertal males

- Inheritance: X-linked, females can have ID
- Gene: FMR-1 (Xq27.3). Mutation is a CGG repeat. Affected individuals have >200 repeats.

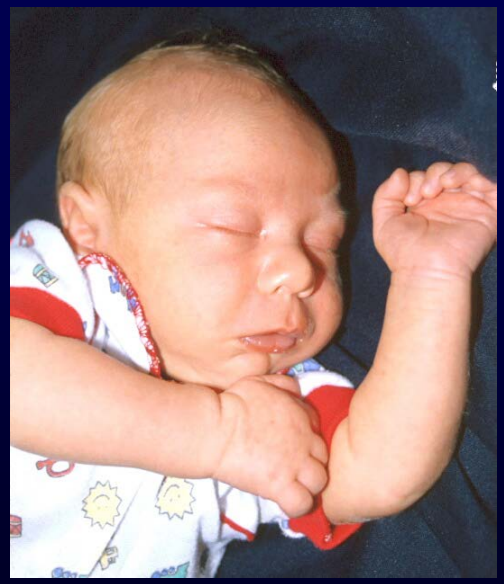
Genetics of Fragile X Syndrome

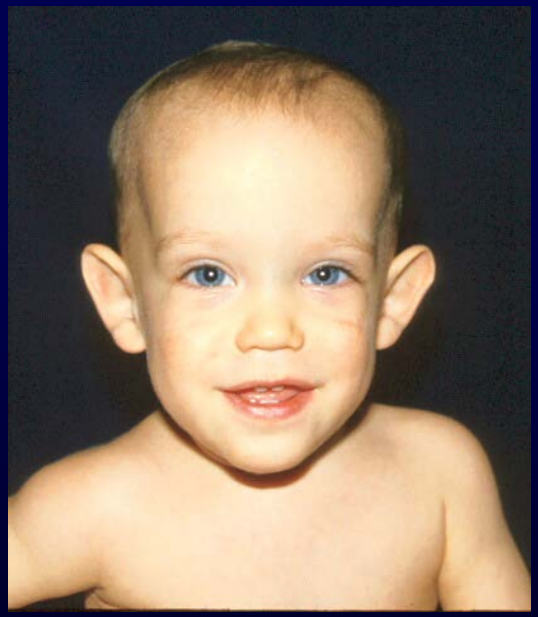
- Affects 1/2000 males
- X-linked, Xq27.3
- CGG repeat
- Normal 7-52
- Affected 230+
- Maternal expansion





- Large ears may be seen
- Babies may appear normal
- Macro-orchidism after puberty
- Intellectual deficiency





Fragile X Syndrome in Adults

- May have relatively long face
- Ears may prominent
- Testicles usually exceed 80-100 cc volume