Fragile X Syndrome

- **What is Fragile X Syndrome?**
  - Fragile X Syndrome (FXS) is a genetic condition that causes intellectual disability, behavioral and learning challenges, and various physical characteristics. Though FXS occurs in both genders, males are more frequently affected than females. Generally, there are no life-threatening health concerns associated with the condition.
- **What are the signs/symptoms?**
  - Behavioral Characteristics:
    - ADD/ADHD
    - Autism Spectrum Disorder
    - Social anxiety
    - Hand-biting and or flapping
    - Poor eye contact
    - Sensory disorders
    - Increased risk for aggression
  - Intellectual Disabilities
    - Intellectual disabilities in FXS include a range from moderate to severe learning disabilities.
  - Physical Features
    - Large ears
    - Long face
    - Soft skin
    - Large testicles in post-pubertal males
  - Connective Tissue Problems
    - Ear infections
    - Flat feet
    - High arched palate
    - Double-jointed fingers
    - Hyper-flexible joints
- **What is Fragile X Syndrome a result of?**
  - Changes in the FMR1 gene. This gene usually makes a protein called FMRP. FMRP is needed for brain development. FXS is inherited in a way that is known as 'X-linked', as the changed gene is on the X chromosome. Males with Fragile X syndrome are often more severely affected than women because males only have one X chromosome, whereas females have two X chromosomes, only one of which is changed.
- **What are the complications of Fragile X Syndrome?**
  - Vary depending on the type and severity of symptoms but they may include
    - Recurrent ear infections in children
    - Seizure disorder

References:

https://medlineplus.gov/ency/article/001668.htm

https://fragilex.org/understanding-fragile-x/fragile-x-syndrome/