Fragile X Syndrome

BQIS Fact Sheets provide a general overview on topics important to supporting an individual’s health and safety and to improving their quality of life. This document provides general information on the topic and is not intended to replace team assessment, decision-making, or medical advice.

Intended Outcomes

Readers will be knowledgeable of the characteristics, cause, and treatments of Fragile X Syndrome.

Definitions

**Fragile X Syndrome**: A genetic condition that causes a range of developmental problems including learning disabilities and cognitive impairment. Males are usually more severely affected by this disorder than females.

**Glutamate**: An amino acid necessary for transmitting an impulse from a nerve cell to another nerve, muscle, or organ.

Facts

- Genetic characteristics of Fragile X Syndrome:
  - Fragile X Syndrome is inherited from carrier mothers.
  - Carrier mothers have mild, if any, symptoms and most likely do not know they are carriers.
  - The genetic defect in Fragile X Syndrome is on the X chromosome. Males have one X and females have two X’s.
  - As an X-linked disorder, Fragile X Syndrome occurs more often in males and symptoms are typically more severe in males.

- Physical features (more commonly seen in males with Fragile X Syndrome):
  - Long, narrow face
  - Enlarged ears
  - Large testicles

- Cognitive and behavioral features of Fragile X Syndrome:
  - Most males with Fragile X Syndrome have significant intellectual disabilities, while only one-third of females have significant intellectual disabilities.
Behavioral characteristics of individuals with Fragile X Syndrome include attention deficit disorder, attention deficit hyperactivity disorder (ADHD), autism or autistic behaviors, social or general anxiety, hand-biting, poor eye contact, sensory disorders, and increased risk for aggression.

- Conditions often occurring in persons with Fragile X Syndrome:
  - Autism spectrum disorders affect approximately 46 percent of males and 16 percent of females with Fragile X Syndrome, according to the Centers for Disease Control and Prevention.
  - Seizures affect approximately 25 percent of individuals with Fragile X Syndrome.

- Cause of Fragile X Syndrome:
  - Individuals with Fragile X Syndrome do not make Fragile X Mental Retardation Protein (FMRP).
  - Without FMRP in their brains, persons with Fragile X Syndrome develop excess activity of the brain transmitter glutamate.
  - Excessive glutamate activity has toxic effects on the brain resulting in the behaviors and characteristics of Fragile X Syndrome.

- Treatment:
  - Currently, treatment consists of education, therapy, medication, and support directed toward symptoms displayed, such as ADHD symptoms, anxiety, and irritable behavior. There is no cure for Fragile X Syndrome.
  - People with Fragile X Syndrome have a higher incidence rate of other medical and health issues.

**Recommended Actions and Prevention Strategies**

1. Administer medications and other treatments as prescribed.

2. Watch for anything different/change in status about the person and for signs and symptoms of side effects of medications.

3. Write down noted signs and symptoms of side effects of medications and changes in status of the person.

4. Communicate noted signs and symptoms of side effects of medications and changes in status of the person to supervisor/nurse immediately.
5. Monitor for effectiveness of treatment by documenting target behavior data that relate to diagnosis.

6. Make sure applicable data is presented to physician/psychiatrist during scheduled appointments.

**Learning Assessment**

The following questions can be used to verify a person's competency regarding the material contained in this Fact Sheet:

1. Which physical characteristic below is commonly seen in males with Fragile X Syndrome?
   - A. High muscle tone
   - B. Small heads
   - C. Large testicles
   - D. None of the above

2. How many persons with Fragile X Syndrome also have an autism spectrum disorder?
   - A. 2 in 3
   - B. 3 in 4
   - C. 1 in 10
   - D. 1 in 20

3. The genetic defect responsible for Fragile X Syndrome is inherited from?
   - A. Siblings
   - B. Grandmothers
   - C. Fathers
   - D. Mothers
4. Symptoms commonly seen in individuals with Fragile X Syndrome include:
   A. Anxiety
   B. Irritability
   C. ADHD symptoms
   D. All of the above

5. To help monitor the condition of someone with Fragile X Syndrome, it is important to do all of the following except:
   A. Watch for any changes in the person
   B. Write down observations of possible side effects of medications
   C. Report changes in status of the person to your supervisor on a monthly basis
   D. Take target behavior data to medical appointments for the physician/psychiatrist’s review.

References


Learning Assessment Answers

1. C
2. A
3. D
4. D
5. C