

HEALTH & SAFETY: FRAGILE X SYNDROME

BQIS/Outreach Fact Sheets provide a general overview on topics important to supporting an individual's health and safety and to improving their quality of life.

Objectives

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

Definitions

Fragile X Syndrome: The most common inherited cause of developmental disability.

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 more severe in males
 - Up to 1 in 250 women are Fragile X Syndrome carriers. Up to 1 in 4,000 persons has Fragile X Syndrome.
- 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:
 - All males and about half of all females with Fragile X Syndrome have mild to moderate intellectual disability
 - Many children with Fragile X Syndrome suffer from Attention Deficit Hyperactivity Disorder (ADHD) symptoms
 - By grade school, many persons with Fragile X Syndrome suffer from significant anxiety
 - By adolescence, many persons with Fragile X Syndrome suffer from irritable behavior including aggression and self-injury
- Conditions often occurring in persons with Fragile X Syndrome:
 - Autism spectrum disorders affect 2 in 3 persons with Fragile X Syndrome
 - Seizures affect 2 in 10 persons with Fragile X Syndrome
- Cause of Fragile X Syndrome:
 - Persons 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 is directed towards symptoms displayed such as ADHD symptoms, anxiety, and irritable behavior
 - Treatments are in development that may be able to reverse the underlying cause of Fragile X Syndrome

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. Document 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. Ensure applicable data is presented to physician/psychiatrist during scheduled appointments

Learning Assessment

Questions that can be used to verify a person's competency in 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 persons with Fragile X Syndrome include:
 - A. Anxiety
 - B. Irritability
 - C. Attention Deficit Hyperactivity Disorder (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. Document 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

National Institute of Mental Health www.ninds.nih.gov/disorders/neuroleptic_syndrome/neuroleptic_syndrome.htm

American Academy of Child & Adolescent Psychiatry www.aacap.org/cs/forFamilies

The National Fragile X Foundation www.nfxf.org

Craig A. Erickson, M.D. Assistant Professor of Psychiatry; Chief, Fragile X Syndrome Clinic Indiana University School of Medicine; Riley Hospital Child & Adolescent Psychiatry Clinic

Related Resources

National Institute of Mental Health www.ninds.nih.gov/disorders/neuroleptic_syndrome/neuroleptic_syndrome.htm

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Learning Assessment Answers

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

Outreach Services

1-866-429-5290 • outreach@fssa.in.gov • DDRSOutreach.IN.gov

As a service for persons supporting individuals with intellectual/developmental disabilities, Outreach and the Indiana Institute on Disability and Community developed the Outreach Fact Sheet Library. The information provided is designed to enhance the understanding of the topic and does not replace other professional or medical instructions or individually developed plans. For more fact sheets and information, please visit DDRSOutreach.IN.gov and www.iidc.indiana.edu/training.



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