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Fragile X Syndrome: Identification and Treatment



Introduction

Patients suffering from Fragile X syndrome may have special needs and requirements. Therefore, it is essential for health care professionals to effectively identify those patients with Fragile X syndrome to ensure the safe and effective administration of health care. This course will review information regarding Fragile X syndrome and the treatment of Fragile X syndrome to provide health care professionals with the necessary insight to administer safe and effective health care to those patients with Fragile X syndrome.

Section 1: Fragile X Syndrome

Case Study 1

A 16-year-old male patient presents with complaints of anxiety. The patient has no known drug allergies and is not currently taking any medications. Upon questioning, the patient reports that he has been "very anxious of late" and has not been sleeping well for the past several months. The patient also reports that he has been "having trouble in school" and "trouble making friends." Further questioning reveals that the patient has "always had trouble learning new things" and often avoids social situations. As the patient is talking, a health care professional observes that the patient is avoiding eye-contact and is repeatedly flapping his right hand. The health care professional also observes that the patient is having trouble articulating what he is feeling and speaking clearly. During a physical examination, the health care professional notes that the patient has a large head, large ears, and a prominent jaw. The health care professional documents the aforementioned observations. The patient's parents are called into the examination room, at the request of the patient, for a consultation.

Case Study 2

A mother and a father present with their 38-month-old son. Upon questioning, the patient's parents report that their son had difficulties learning to crawl, walk, and speak. They also reveal that their son seems to be "very sensitive" to the way things smell, taste, look, and feel and often avoids eye-contact, has trouble maintaining focus, and easily becomes aggressive. A health care professional listens to the mother and father and conducts a developmental screening. At the conclusion of the developmental screening, the health care professional refers the patient and his parents to a genetic counselor.

Case Study 3

A 20-year-old female patient presents with complaints of depression. Upon questioning, the patient reveals that she has been "extremely depressed lately" to the point where she cannot "get out of bed sometimes." The patient also reveals that she is "not taking" her Celexa. When asked why she stopped taking Celexa, the patient responded by saying "I don't know." Further questioning reveals that the patient recently had to "drop out of school" because it was "too hard" and her classes were "too much" for her. The patient also reports that she has a history of cutting herself and feels like she wants to hurt herself again. A health care professional documents that aforementioned patient information and the following patient symptoms: the patient appeared to avoid eye contact during questioning, the patient stuttered at times, and the patient appeared to have a narrow face, large ears, and a prominent jaw. The patient is admitted into the health care facility.

The three case studies presented above highlight patients that may be suffering from Fragile X syndrome. As previously mentioned, patients suffering from Fragile X syndrome may have special needs and requirements - therefore, it is essential for health care professionals to effectively identify those patients with Fragile X syndrome to ensure the safe and effective administration of health care. Due to the importance of identifying patients with Fragile X syndrome, this section of the course will provide insight into Fragile X syndrome. The information found in this section was derived from materials provided by the Centers for Disease Control and Prevention (CDC), the United States Department of Health & Human Services, the American Academy of Pediatrics, and the Joint Commission.^{1,2,3,4}

What is Fragile X syndrome?

Fragile X syndrome may refer to a genetic disorder that affects development.

Fragile X syndrome can lead to learning disabilities, poor communication skills, cognitive impairment, and physical abnormalities.

What causes Fragile X syndrome?

It is believed, that Fragile X syndrome is caused by a mutation of the Fragile X Mental Retardation 1 (FMR1) gene found on the X chromosome. The term mutation may refer to a change in the structure of a gene.

Health care professionals should note the following: Fragile X syndrome is inherited, which means it is passed down from parents to their children; any individual with the FMR1 gene mutation can pass it to their children; males will pass it down to all of their daughters and not to their sons; females have a fifty percent chance to pass it

along to both their sons and daughters; Fragile X syndrome can affect both males and females, however, research indicates that Fragile X syndrome is more common in males; research also indicates males with Fragile X syndrome, typically, experience severer symptoms when compared to females with Fragile X syndrome.

Health care professionals should also note that the major symptoms of Fragile X syndrome fall into the following categories: intelligence and learning symptoms, physical symptoms, behavioral , social, and emotional symptoms, speech and language symptoms, and sensory symptoms.

What are the intelligence and learning symptoms associated with Fragile X syndrome?

The intelligence and learning symptoms associated with Fragile X syndrome include the following:

- Learning disorders
- An inability to learn new skills
- An inability to efficiently or effectively reason
- An inability to efficiently or effectively solve problems
- Intellectual disability (intellectual disability may refer to a disability characterized by significant limitations in both intellectual functioning and in adaptive behavior, which limits an individual's ability to learn at an expected level and function in daily life).

Health care professionals should note the following when considering the intelligence and learning symptoms associated with Fragile X syndrome: males who have Fragile X syndrome usually have some degree of intellectual disability that can range from mild to severe, while females with Fragile X syndrome can have normal intelligence or some degree of intellectual disability.

What are the physical symptoms associated with Fragile X syndrome?

The physical symptoms associated with Fragile X syndrome include the following:

- Large head
- Narrow face

- Large ears
- Prominent jaw
- Prominent forehead
- Flexible joints
- Flat feet
- Enlarged testicles in males
- Scoliosis

Health care professionals should note the following when considering the physical symptoms associated with Fragile X syndrome: most infants and younger children with Fragile X syndrome do not often exhibit physical symptoms; the physical symptoms associated with Fragile X syndrome typically develop when an individual goes through puberty.

What are the behavioral, social, and emotional symptoms associated with Fragile X syndrome?

The behavioral, social, and emotional symptoms associated with Fragile X syndrome include the following:

- Hyperactivity (e.g., very active)
- Impulsivity (e.g., acting without thinking)
- Anxiety
- Aggression
- Short attention span
- Avoids eye-contact
- Causes self injury
- Displays unusual sleeping habits
- Exhibits repetitive motions (the term repetitive motion, as it relates to Fragile X syndrome, may refer to any action that is consistently repeated for no apparent reason; examples of respective motions may include the

following: hand flapping, arm flapping, body rocking, and/or consistently spinning oneself in circles)

Health care professionals should note the following: individuals with Fragile X syndrome may appear shy or resistant. Health care professionals should also note that the behavioral, social, and emotional symptoms associated with Fragile X syndrome often lead to dramatic problems that impact everyday life.

What are the speech and language symptoms associated with Fragile X syndrome?

The speech and language symptoms associated with Fragile X syndrome include the following:

- Delayed speech and language skills
- Trouble speaking clearly
- May leave out parts of words when speaking
- Stuttering
- An inability to understand body language
- An inability to understand social cues (e.g., an individual with Fragile X syndrome may not be able to differentiate different speaking tones and/or tones signifying specific cues)
- Echolalia (the term echolalia may refer to reparative speech patterns; respective word use)

Health care professionals should note the following: individuals with Fragile X syndrome may begin speaking much later when compared to those without Fragile X syndrome; some individuals with Fragile X syndrome may not be able to speak at all.

What are the sensory symptoms associated with Fragile X syndrome?

The sensory symptoms associated with Fragile X syndrome include the following:

- Hypersensitivity to sounds and light
- Hypersensitivity to the way things smell, taste, look, or feel

- Unusual reactions to the way things sound, smell, taste, look, or feel (e.g., an individual with Fragile X syndrome may be extremely bothered by the way a piece of clothing feels on his or her body)

Health care professionals should note the following: often the sensory symptoms of Fragile X syndrome lead to behavioral issues (e.g., an individual with Fragile X syndrome may be bothered by a sound and/or light to the point where it causes the individual to act out or become aggressive).

When do individuals typically begin to exhibit Fragile X syndrome symptoms?

Some of the symptoms of Fragile X syndrome appear early in development; often individuals show symptoms of Fragile X syndrome by 36 months to 42 months of age or even earlier (the physical symptoms associated with Fragile X syndrome typically develop when an individual goes through puberty).

The early signs and symptoms of Fragile X syndrome include the following:

- Problems with eye contact
- Problems with nonverbal communication
- Delayed speech and language skills
- Delayed motor skills/problems with motor skills (e.g., an individual with Fragile X syndrome may have problems learning to walk)
- Emotional and social difficulties
- Displays unusual reactions to the way things sound, smell, taste, look, or feel

How does Fragile X syndrome affect development?

Fragile X syndrome may affect some individuals' ability to reach age-related developmental milestones. An age-related developmental milestone may refer to any significant change typically associated with growth and aging. Age-related developmental milestones traditionally vary depending on the stage of development (i.e., the age-related developmental milestones associated with a 2 month old infant include the following: a 2 month old infant should begin to smile at people, pay attention to faces, and react to sound).

Health care professionals should also note that Fragile X syndrome may also affect the development of motor skills. For example, an individual with Fragile X syndrome may have problems learning to crawl or walk.

How may patients with Fragile X syndrome present?

Patients with Fragile X syndrome may present at different stages of development and with various symptoms from the different symptom categories associated with Fragile X syndrome. For example, a 40-month-old child with Fragile X syndrome may present with the following symptoms: problems with eye contact, hypersensitivity to way things smell, taste, look, or feel, emotional instability, and delayed age-related developmental milestones; while a 17-year-old patient may present with the following symptoms: stuttering, hyperactivity, large head, narrow face, large ears, and prominent jaw. Essentially, individuals with Fragile X syndrome may present at different ages and with a broad array of varied Fragile X syndrome-related symptoms.

Due to the potential for patients with Fragile X syndrome to present at different ages and with a broad array of varied Fragile X syndrome symptoms, it is important for health care professionals to employ effective patient observation when examining or attempting to identify patients with suspected Fragile X syndrome. Patient observation can be essential to identifying specific characteristics and symptoms of Fragile X syndrome. Patient observation can also be instrumental in identifying the special needs and requirements associated with Fragile X syndrome patients.

Along with patient observation, health care professionals should also be sure to complete effective health care documentation when examining and/or administering care to patients with Fragile X syndrome or suspected Fragile X syndrome. Health care documentation can be essential to communicating potential Fragile X syndrome symptoms and to the safe administration of health care to those patients with Fragile X syndrome. Additionally, effective health care documentation may be used as a method to review patient cases and to ensure all aspects of an individual patient's health care are noted and evaluated to maximize therapeutic outcomes. Health care professionals should note the following: in order for health care documentation to be considered effective, it must function as a viable form of communication, as well as a means to establish a detailed record of health care administration.

How is Fragile X syndrome diagnosed?

The diagnostic process for Fragile X syndrome typically involves two major stages or steps, which include developmental screening and a comprehensive diagnostic evaluation.

Developmental screening - developmental screening is often the first step to diagnosing Fragile X syndrome. Developmental screening may refer to the process of determining if an individual is progressing at a desired rate (e.g., is a child learning basic skills when he or she should). The essential elements of a typical developmental screening include: a parent interview, a child interview, and the use of a related questionnaire and/or the use of a specific developmental screening tool for Fragile X syndrome. If a health care professional determines, through a developmental screening, that a patient may, potentially, have Fragile X syndrome, he or she may recommend that the patient move to the second stage or step of the Fragile X syndrome diagnostic process, which consists of a comprehensive diagnostic evaluation.

Comprehensive diagnostic evaluation - a comprehensive diagnostic evaluation, as it relates to Fragile X syndrome, may refer to a diagnostic procedure used to determine if an individual meets the criteria necessary for Fragile X syndrome diagnosis. A comprehensive diagnostic evaluation for Fragile X syndrome may include: a parent interview, a child interview, and a genetic test to confirm the presence of Fragile X syndrome.

What conditions are typically associated with Fragile X syndrome?

Unfortunately, an array of physical and mental-health conditions frequently accompany Fragile X syndrome. The physical and mental-health conditions that frequently accompany Fragile X syndrome include the following: anxiety, depression, attention-deficit/hyperactivity disorder (ADHD), autism spectrum disorder (ASD), and intellectual disability. Specific information regarding each of the aforementioned conditions/disorders and how they relate to Fragile X syndrome may be found below.

Anxiety may refer to a mental health disorder characterized by prolonged periods of persistent, excessive worry about a number of events or activities, which causes clinically significant distress or impairment in social, occupational, or other important areas of functioning. There are several types of anxiety disorders. The most common type of anxiety disorder is referred to as generalized anxiety disorder. Generalized anxiety disorder may refer to a mental health disorder characterized by excessive anxiety and worry occurring more days than not for at least 6 months, about a number of events or activities (such as work or school performance), which is difficult to control and leads to clinically significant distress or impairment in social, occupational, or other important areas of functioning. Symptoms of generalized anxiety disorder may include: excessive anxiety, excessive worry, restlessness, persistent feelings of being keyed up or on edge, easily fatigued, difficulty

concentrating, mind feeling blank at times (mind going blank), irritability, muscle tension, and sleep difficulties.

Individuals suffering from generalized anxiety disorders may present in a variety of different states. They may appear anxious, worried, fearful, terrified, troubled, distracted and/or helpless. Also, they may report experiencing sleep problems and/or muscle tension and stiffness. Additionally, they may exhibit behaviors that may seem odd or inconsistent with other patient populations. Individuals potentially suffering from generalized anxiety disorders may also display body language indicating anxiety, worry, tension, and/or fear such as: consistently moving limbs, rubbing hands together, shaking, appearing tense or stiff as well as excessive finger nail biting and/or lip biting.

Health care professionals should note the following when considering Fragile X syndrome associated anxiety: anxiety can be triggered by different activities, including activities that were previously considered enjoyable. Health care professionals should also note the following: individuals with Fragile X syndrome may also experience separation anxiety disorder. Separation anxiety disorder may refer to a form of an anxiety disorder characterized by excessive worry and/or fear centered around being apart from select individuals.

Major depressive disorder may refer to a form of depression that occurs most days of the week for a period of two weeks or longer leading to clinically significant distress or impairment in social, occupational, or other important areas of functioning. Symptoms of major depressive disorder may include: depressed mood, a loss of interest in previously enjoyable activities, appetite changes, weight changes, sleep difficulties, psychomotor agitation or retardation, fatigue or loss of energy, diminished ability to think or concentrate, feelings of worthlessness or excessive guilt, and suicidality.

Individuals suffering from major depressive disorders may present in a variety of different states. They may appear untidy or disheveled. Also, their personal hygiene may be lacking. Additionally, they may appear troubled or distracted. Furthermore, individuals suffering from major depressive disorders may exhibit behaviors that may seem odd or inconsistent with other patient populations and may display body language indicating a depressed mood (e.g., moving slowly, head tilting down, arms crossed, and slouching).

Health care professionals should note that Fragile X syndrome associated depression may lead to suicidal ideation. Suicidal ideation may refer to thoughts of suicide and/or thoughts of planning suicide. Health care professionals should be very aware that individuals suffering from Fragile X syndrome associated depression may be suicidal.

Health care professionals should make every effort to identify the potential for suicide and prevent patient suicide when applicable. Health care professionals should also note the following: the suicide of a patient while in a staffed, round-the-clock care setting is a frequently reported type of sentinel event (the term sentinel event may refer to an unanticipated event in a health care setting that results in death or serious physical or psychological injury to a patient(s), not related to the natural course of the patient's illness); identification of individuals at risk for suicide while under the care of or following discharge from a health care organization is an important step in protecting these at-risk individuals; regarding patients at risk for suicide, health care professionals and health care organizations should follow the following recommendations: identify patients at risk for suicide, conduct a risk assessment that identifies specific patient characteristics and environmental features that may increase or decrease the risk for suicide, address the patient's immediate safety needs and most appropriate setting for treatment, and when a patient at risk for suicide leaves the care of the health care facility, provide suicide prevention information (such as relevant crisis hotline information) to the patient and his or her family.

Attention-deficit/hyperactivity disorder (ADHD) may refer to a type of brain disorder which is marked by an ongoing pattern of inattention and/or hyperactivity-impulsivity that interferes with functioning or development. ADHD may be very common in Fragile X syndrome patient populations. Symptoms associated with ADHD may include the following: an inability to maintain focus, an inability to maintain sustained mental effort for long periods of time, a capacity to consistently make mistakes, disorganization, forgetfulness, and restlessness. Health care professionals should note the following when considering Fragile X syndrome associated ADHD: symptoms of ADHD may overlap with those of Fragile X syndrome.

Autism spectrum disorder (ASD) may refer to a complex developmental disorder that affects how an individual behaves, interacts with others, communicates, and learns. ASD may be very common in Fragile X syndrome patient populations. Symptoms associated with ASD may include the following: eye-contact avoidance, resistant to physical contact, trouble understanding other individual's feelings, trouble talking about his or her own feelings, gives unrelated answers to questions, unusual language use, does not point or respond to pointing, uses few or no gestures (e.g., does not wave hello or goodbye), repetitive motions, talks in a flat, robot-like, or sing-song voice, often gets upset by minor changes, has obsessive interests, and has to follow certain routines.

Health care professionals should note that ASD is referred to as a spectrum disorder because there is a wide variation in the type and severity of ASD symptoms. Health care professionals should also note the following: patients with ASD may present with

a range of ASD symptoms that differ in both type and severity; the symptoms of ASD may overlap with those of Fragile X syndrome.

Intellectual disability may refer to a disability characterized by significant limitations in both intellectual functioning and in adaptive behavior, which limits an individual's ability to learn at an expected level and function in daily life. As previously mentioned individuals with Fragile X syndrome often suffer from intellectual disability. Individuals with intellectual disability often have a below-average intelligence quotient (IQ) and/or intelligence as well as limitations in the ability to function in areas of daily life, such as communication, self-care, and getting along in social situations and school activities (IQ may refer to a number representing an individual's intelligence and ability to reason, which is derived from a specific, related test or tests). Health care professionals should note the following: individuals with intellectual disability can and do learn new skills, but they develop more slowly than children with average intelligence and adaptive skills; there are different degrees of intellectual disability; an individual's level of intellectual disability can be defined by their IQ, or by the types and amount of support they need. Additional information regarding intellectual disability may be found below.

- Signs of an intellectual disability include the following: delays in age-related developmental milestones, difficulty understanding social rules, difficulty seeing the consequences of actions, difficulty thinking logically, and poor problem solving skills.
- There are different degrees of intellectual disability, ranging from mild to severe; individuals with more severe forms of intellectual disability typically require support; individuals with milder forms of intellectual disability can gain some independent skills which may allow them to function and carry out daily activities on their own.
- In order to be diagnosed with an intellectual disability, an individual must have both a significantly low IQ and considerable problems in everyday functioning.
- A normal IQ score is around 100; individuals with an intellectual disability, typically, have an IQ score between 70 and 55 or lower.
- As previously mentioned, an individual must have considerable problems in everyday functioning to be diagnosed with an intellectual disability. To examine an individual's everyday functioning, adaptive behavior should be evaluated. Adaptive behavior may refer to any age-appropriate behavior that allows individuals to live independently and to function well in daily life.

Examples of the types of adaptive behaviors that should be evaluated when attempting to identify an individual with intellectual disability include the following: personal care skills (e.g., the ability to get dressed alone, the ability of an individual to engage in effective personal hygiene), communication and social skills (e.g., the ability of an individual to have a conversation with another individual), school or work skills, learning routines, the ability to engage in activities in a safe manner, the ability to ask for assistance, and the ability to understand the concept of money.

Health care professionals should make a concerted effort to identify and manage any additional issues or concerns that may arise when administering health care to patients with Fragile X syndrome, especially when potential additional issues or concerns center around suicidal ideation.

Section 1: Summary

Fragile X syndrome may refer to a genetic disorder that affects development. The major symptoms of Fragile X syndrome fall into the following categories: intelligence and learning symptoms, physical symptoms, behavioral, social, and emotional symptoms, speech and language symptoms, and sensory symptoms. The diagnostic process for Fragile X syndrome typically involves two major stages or steps, which include developmental screening and a comprehensive diagnostic evaluation. The physical and mental-health conditions typically associated with Fragile X syndrome include the following: anxiety, depression, ADHD, ASD, and intellectual disability. To best serve patients with Fragile X syndrome, health care professionals should possess an understanding of Fragile X syndrome as well as its presentation, diagnosis, and associated conditions.

Section 1: Key Concepts

- Fragile X syndrome can lead to learning disabilities, poor communication skills, cognitive impairment, and physical abnormalities.
- It is believed, that Fragile X syndrome is caused by a mutation of the FMR1 gene found on the X chromosome.
- Fragile X syndrome is inherited, which means it is passed down from parents to their children; any individual with the FMR1 gene mutation can pass it to their children.
- Fragile X syndrome can affect both males and females, however, research indicates that Fragile X syndrome is more common in males.

- The major symptoms of Fragile X syndrome fall into the following categories: intelligence and learning symptoms, physical symptoms, behavioral, social, and emotional symptoms, speech and language symptoms, and sensory symptoms.
- Fragile X syndrome may affect some individual's ability to reach age-related developmental milestones.
- Individuals with Fragile X syndrome may present at different ages and with a broad array of varied Fragile X syndrome-related symptoms.
- The diagnostic process for Fragile X syndrome typically includes a genetic test to confirm the presence of Fragile X syndrome.
- The physical and mental-health conditions typically associated with Fragile X syndrome include the following: anxiety, depression, ADHD, ASD, and intellectual disability.
- Health care professionals should make a concerted effort to identify and manage any additional issues or concerns that may arise when administering health care to patients with Fragile X syndrome, especially when potential additional issues or concerns center around suicidal ideation.

Section 1: Key Terms

Fragile X syndrome - a genetic disorder that affects development

Mutation - a change in the structure of a gene

Intellectual disability - a disability characterized by significant limitations in both intellectual functioning and in adaptive behavior, which limits an individual's ability to learn at an expected level and function in daily life

Repetitive motion (as it relates to Fragile X syndrome) - any action that is consistently repeated for no apparent reason

Echolalia - reparative speech patterns; respective word use

Age-related developmental milestone - any significant change typically associated with growth and aging

Developmental screening - the process of determining if an individual is progressing at a desired rate

Comprehensive diagnostic evaluation (as it relates to Fragile X syndrome) - a diagnostic procedure used to determine if an individual meets the criteria necessary for Fragile X syndrome diagnosis

Anxiety - a mental health disorder characterized by prolonged periods of persistent, excessive worry about a number of events or activities, which causes clinically significant distress or impairment in social, occupational, or other important areas of functioning

Generalized anxiety disorder - a mental health disorder characterized by excessive anxiety and worry occurring more days than not for at least 6 months, about a number of events or activities (such as work or school performance), which is difficult to control and leads to clinically significant distress or impairment in social, occupational, or other important areas of functioning

Separation anxiety disorder - a form of an anxiety disorder characterized by excessive worry and/or fear centered around being apart from select individuals

Major depressive disorder - a form of depression that occurs most days of the week for a period of two weeks or longer leading to clinically significant distress or impairment in social, occupational, or other important areas of functioning

Suicidal ideation - thoughts of suicide and/or thoughts of planning suicide

Sentinel event - an unanticipated event in a health care setting that results in death or serious physical or psychological injury to a patient(s), not related to the natural course of the patient's illness

Attention-deficit/hyperactivity disorder (ADHD) - a type of brain disorder which is marked by an ongoing pattern of inattention and/or hyperactivity-impulsivity that interferes with functioning or development

Autism spectrum disorder (ASD) - a complex developmental disorder that affects how an individual behaves, interacts with others, communicates, and learns

Intelligence quotient (IQ) - a number representing an individual's intelligence and ability to reason, which is derived from a specific, related test or tests

Adaptive behavior - any age-appropriate behavior that allows individuals to live independently and to function well in daily life

Section 1: Personal Reflection Question

How can health care professionals effectively identify an individual with Fragile X syndrome?

Section 2: Fragile X Syndrome Treatment

It is important for health care professionals to possess insight into Fragile X syndrome in order to best serve patients in need. With that said, health care professionals should also possess insight into Fragile X syndrome treatment options. Treatment for Fragile X syndrome can come in many different forms including both non-pharmacological and pharmacological options. The beginning of this section of the course will focus on some of the more common non-pharmacological treatment options for Fragile X syndrome.

Non-Pharmacological Treatment Options for Fragile X Syndrome

Physical Therapy

Physical therapy may not be the first non-pharmacological treatment option that comes to mind, however some patients with Fragile X syndrome may require physical therapy. Physical therapy may refer to the practice of treating a disease, condition, disorder, and/or injury through physical means.² The goal of physical therapy, when used to treat patients with Fragile X syndrome, is often to build motor skills and improve strength, posture, and balance.²

Speech-Language Therapy

Speech-language therapy may refer to a form of therapy that addresses issues with speech, language, and communication.² The goal of speech-language therapy, when used to treat patients with Fragile X syndrome, is to improve upon the pronunciation of words, slow down speech patterns, and improve upon the use of language to express ideas and emotions.²

Psychotherapy

Psychotherapy, also known as talk therapy, may refer to the use of psychological techniques and/or psychotherapeutic approaches to help individuals overcome problems and develop healthier habits.⁵ Health care professionals should note that many different forms of psychotherapy may be used to treat patients with Fragile X syndrome.

Cognitive Behavioral Therapy

Cognitive behavioral therapy may refer to a form of psychotherapy which focuses on helping individuals solve problems and create positive outcomes by changing unrealistically negative patterns of thought and behavior.⁶ In other words, cognitive behavioral therapy works to identify unrealistically negative thoughts and their relationship to negative behavior patterns and outcomes in order to develop constructive ways of thinking that will ultimately lead to more positive behavior patterns and outcomes.

Support Groups

Support groups may also be used as a therapeutic option for those with Fragile X syndrome. Support groups can be used to help those with Fragile X syndrome avoid isolation and make connections with other individuals to improve upon symptoms and quality of life. Health care professionals should be aware that various types of support groups exist and that an individual may participate in one or more support group at a time to cope with or manage his or her Fragile X syndrome.

Sleep Routines

Some individuals with Fragile X syndrome may suffer from sleep disturbances and/or have problems sleeping. Therefore, developing routines involving sleep or bedtimes may be beneficial to patients suffering from Fragile X syndrome. Specific information regarding age-related sleep recommendations may be found in Figure 1.

FIGURE 1: SLEEP RECOMMENDATIONS¹

Sleep Recommendations for Newborns

- Newborns should sleep 14 - 17 hours per 24 hours.

Sleep Recommendations for infants

- Infants should sleep 12 - 16 hours (including naps) per 24 hours.

Sleep Recommendations for individuals 1 - 2 years old

- Individuals 1 - 2 years old should sleep 11 - 14 hours (including naps) per 24 hours.

Sleep Recommendations for individuals 3 - 5 years old

- Individuals 3 - 5 years old should sleep 10 - 13 hours (including naps) per 24 hours.

Sleep Recommendations for individuals 6 - 12 years old

- Individuals 6 - 12 years old should sleep 9 - 12 hours per 24 hours.

Sleep Recommendations for individuals 13 - 18 years old

- Individuals 13 - 18 years old should sleep 8 - 10 hours per 24 hours.

Sleep Recommendations for individuals 18 - 60 years old

- Individuals 18 - 60 years old should sleep 7 or more hours per night.

Sleep Recommendations for individuals 61 - 64 years old

- Individuals 61 - 64 years old should sleep 7 - 9 hours.

Sleep Recommendations for individuals 65 years old or older

- Individuals 65 years old or older should sleep 7 - 8 hours.

Pharmacological Treatment

As previously mentioned, pharmacological treatment options may be used as care for individuals with Fragile X syndrome. That being the case, the rest of this section will focus on some of the most widely prescribed medications used to treat individuals with Fragile X syndrome. The medications highlighted in this section will be presented in informational segments. The information found below was derived from materials provided by the United States Department of Health & Human Services and the United States Food and Drug Administration (FDA).^{2,7} When reviewing the highlighted medications, health care professionals should keep in mind that, currently, the FDA has not approved any medications for the specific treatment of Fragile X syndrome. That being said, patients with Fragile X syndrome may be treated in some capacity with the following medications. Health care professionals should also keep in mind that the following medications may be used alone or in combination with other therapeutic options to treat individuals with Fragile X syndrome.

Citalopram (Celexa)

Medication notes - Patients with Fragile X syndrome may be treated in some capacity with selective serotonin reuptake inhibitors (SSRIs). Celexa is an example of a SSRI. Celexa is an orally administered medication. Celexa doses above 40 mg/day are not recommended due to the risk of QT prolongation. Common side effects of Celexa may

include: nausea, dry mouth, somnolence, insomnia, increased sweating, diarrhea, tremor and sexual dysfunction.

Safety notes - Celexa is contraindicated in patients with a hypersensitivity to citalopram or any of the inactive ingredients in Celexa. Concomitant use in patients taking monoamine oxidase inhibitors (MAOIs) and/or pimozide is also contraindicated. Warnings associated with Celexa include the following: patients with major depressive disorder, both adult and pediatric, may experience worsening of their depression and/or the emergence of suicidal ideation and behavior (suicidality) or unusual changes in behavior, whether or not they are taking antidepressant medications, and this risk may persist until significant remission occurs. Suicide is a known risk of depression and certain other psychiatric disorders, and these disorders themselves are the strongest predictors of suicide. There has been a long-standing concern, however, that antidepressants may have a role in inducing worsening of depression and the emergence of suicidality in certain patients during the early phases of treatment. All patients being treated with antidepressants for any indication should be monitored appropriately and observed closely for clinical worsening, suicidality, and unusual changes in behavior, especially during the initial few months of a course of drug therapy, or at times of dose changes, either increases or decreases.

Considerations for special patient populations - No dosage adjustment is necessary for patients with mild or moderate renal impairment. Celexa should be used with caution in patients with severe renal impairment. Celexa falls in Pregnancy Category C.

Fluoxetine (Prozac)

Medication notes - Prozac is another example of an SSRI. A typical adult starting dose for Prozac is 20 mg daily. Common side effects of Prozac include: nausea, diarrhea, tremor, dry mouth, sweating, headaches, dizziness and weakness.

Safety notes - Prozac is contraindicated in patients with a hypersensitivity to Prozac or any of the inactive ingredients in Prozac. Additional Prozac contraindications include the following: concurrent use of MAOIs intended to treat psychiatric disorders with Prozac or within five weeks of stopping treatment with Prozac; do not use Prozac within 14 days of stopping an MAOI intended to treat psychiatric disorders; in addition, do not start Prozac in a patient who is being treated with linezolid or intravenous methylene blue; do not use Prozac with pimozide or thioridazine due to risk of QT prolongation and drug interaction; do not use thioridazine within five weeks of discontinuing Prozac. Warnings and precautions associated with Prozac include the following: serotonin syndrome has been reported with SSRIs and SNRIs, including Prozac, both when taken alone, but especially when co-administered with other

serotonergic agents (including triptans, tricyclic antidepressants, fentanyl, lithium, tramadol, tryptophan, buspirone, amphetamines, and St. John's Wort). If such symptoms occur, discontinue Prozac and initiate supportive treatment. If concomitant use of Prozac with other serotonergic drugs is clinically warranted, patients should be made aware of a potential increased risk for serotonin syndrome, particularly during treatment initiation and dose increases. Patients should also be screened for bipolar disorder and monitored for mania/hypomania due to potential activation of mania/hypomania. Prozac should be used cautiously in patients with a history of seizures or with conditions that potentially lower the seizure threshold. Prozac may lead to altered appetite and significant weight loss. Prozac may increase the risk of bleeding when used with NSAIDs, aspirin, warfarin, or other drugs that affect coagulation/may potentiate the risk of gastrointestinal or other bleeding; angle-closure glaucoma has occurred in patients with untreated anatomically narrow angles treated with antidepressants; hyponatremia has been reported with Prozac in association with syndrome of inappropriate antidiuretic hormone (SIADH), consider discontinuing if symptomatic hyponatremia occurs; anxiety and insomnia may occur; QT prolongation and ventricular arrhythmia including Torsades de Pointes have been reported with Prozac use, use with caution in conditions that predispose to arrhythmias or increased fluoxetine exposure, use cautiously in patients with risk factors for QT prolongation; Prozac has potential to impair judgment, thinking, and motor skills. Other warnings associated with Prozac include the following: Prozac may lead to increased risk of suicidal thinking and behavior in children, adolescents, and young adults taking antidepressants; monitor for worsening and emergence of suicidal thoughts and behaviors.

Considerations for special patient populations - Lower or less frequent dosing may be appropriate in patients with cirrhosis. Prozac should be used during pregnancy only if the potential benefit justifies the potential risks to the fetus. Breast feeding is not recommended.

Sertraline (Zoloft)

Medication notes - Zoloft is another example of an SSRI. The mechanism of action of Zoloft is presumed to be linked to its inhibition of CNS neuronal uptake of serotonin. Zoloft should be administered once daily, either in the morning or evening. Common side effects of Zoloft include: nausea, diarrhea/loose stool, tremor, dyspepsia, decreased appetite, hyperhidrosis, ejaculation failure and decreased libido.

Safety notes - Zoloft is contraindicated in patients with a hypersensitivity to sertraline or any of the inactive ingredients in Zoloft. Concomitant use in patients taking MAOIs is contraindicated. Concomitant use in patients taking pimozide is contraindicated. Additionally, Zoloft oral concentrate is contraindicated with

disulfiram (Antabuse) due to the alcohol content of the concentrate. Warnings and precautions associated with Zoloft include the following: increased risk of serotonin syndrome when co-administered with other serotonergic agents; increased risk of bleeding when used with aspirin, nonsteroidal anti-inflammatory drugs (NSAIDs), other antiplatelet drugs, warfarin, and other anticoagulants may increase risk; screen patients for bipolar disorder due to activation of mania/hypomania; use with caution in patients with seizure disorders and avoid use of antidepressants, including Zoloft, in patients with untreated anatomically narrow angles. Other warnings associated with Zoloft include the following: antidepressants increase the risk of suicidal thoughts and behaviors in pediatric and young adult patients, closely monitor for clinical worsening and emergence of suicidal thoughts and behaviors.

Considerations for special patient populations - Health care professionals should note the following: the use of Zoloft in patients with liver disease should be approached with caution. Health care professionals should also note the following: third trimester use of Zoloft may increase risk for persistent pulmonary hypertension and withdrawal in the neonate. Zoloft falls into Pregnancy Category C.

Risperidone (Risperdal)

Medication notes - Risperdal is an atypical antipsychotic agent indicated for the treatment of irritability associated with autistic disorder in children and adolescents aged 5 - 16 years. Risperdal is also indicated for the treatment of schizophrenia in adults and adolescents aged 13-17 years as well as other mental health disorders. Common side effects of Risperdal include: somnolence, appetite increases, fatigue, rhinitis, upper respiratory tract infection, vomiting, coughing, urinary incontinence, saliva increased, constipation, fever, Parkinsonism, dystonia, abdominal pain, anxiety, nausea, dizziness, dry mouth, tremor, rash, akathisia, and dyspepsia.

Safety notes - Contraindications associated with Risperdal include a known hypersensitivity to the product. Warnings associated with Risperdal include: elderly patients with dementia-related psychosis treated with antipsychotic drugs are at an increased risk of death; Risperdal is not approved for use in patients with dementia-related psychosis. Additional warnings associated with Risperdal include the following: leukopenia, neutropenia, and agranulocytosis have been reported with antipsychotics, including Risperdal; patients with a history of a clinically significant low white blood cell count (WBC) or a drug-induced leukopenia/neutropenia should have their complete blood count (CBC) monitored frequently during the first few months of therapy and discontinuation of Risperdal should be considered at the first sign of a clinically significant decline in WBC in the absence of other causative factors; Risperdal may lead to cognitive and motor impairment; Risperdal may lead to suicide.

Considerations for special patient populations - Health care professionals should note the following: safety and effectiveness not established for schizophrenia less than 13 years of age, for bipolar mania less than 10 years of age, and for autistic disorder less than 5 years of age; nursing mothers should not breast feed.

Olanzapine (Zyprexa)

Medication notes - Zyprexa is an atypical antipsychotic medication indicated for acute treatment of manic or mixed episodes associated with bipolar I disorder and maintenance treatment of bipolar I disorder as well as other types of mental health disorders. The typical oral adult starting dose of Zyprexa for the treatment of bipolar I disorder (manic or mixed episodes) is 10 or 15 mg once daily. Side effects of Zyprexa may include: postural hypotension, constipation, weight gain, dizziness, personality disorder and akathisia.

Safety notes - Warnings associated with Zyprexa include the following: elderly patients with dementia-related psychosis treated with antipsychotic drugs are at an increased risk of death. Zyprexa is not approved for the treatment of patients with dementia-related psychosis.

Considerations for special patient populations - Safety and effectiveness of Zyprexa in children < 13 years of age have not been established. Zyprexa should be used during pregnancy only if the potential benefit justifies the potential risk to the fetus. Breast-feeding is not recommended.

Ritalin

Medication notes - Ritalin is a central nervous system (CNS) stimulant. Ritalin may be used to help increase attention and decrease impulsiveness and hyperactivity in patients with Fragile X syndrome. The exact mechanism of action of Ritalin is unknown, but it is believed that Ritalin activates the brain stem arousal system and cortex to produce its stimulant effect. Dosage should be individualized according to the needs and responses of the patient. Side effects of Ritalin may include: nervousness, trouble sleeping, loss of appetite, weight loss, dizziness, headaches, nausea, and vomiting. Health care professionals should note that Ritalin is often used with other therapeutic options such as cognitive behavioral therapy.

Safety notes - Marked anxiety, tension, and agitation are contraindications to Ritalin, since the drug may aggravate the aforementioned symptoms. Ritalin is also contraindicated in patients known to be hypersensitive to the drug, in patients with glaucoma, and in patients with motor tics or with a family history or diagnosis of Tourette's syndrome. Additionally, Ritalin is contraindicated during treatment with monoamine oxidase inhibitors, and also within a minimum of 14 days following

discontinuation of a monoamine oxidase inhibitor (MAOI). Warnings associated with Ritalin include the following: sudden death has been reported in association with CNS stimulant treatment at usual doses in children and adolescents with structural cardiac abnormalities or other serious heart problems; sudden death, stroke, and myocardial infarction have been reported in adults taking stimulant drugs at usual doses for ADHD, stimulant medications cause a modest increase in average blood pressure and average heart rate. Additional warnings associated with Ritalin include: administration of stimulants may exacerbate symptoms of behavior disturbance and thought disorder in patients with a pre-existing psychotic disorder, aggressive behavior or hostility is often observed in children and adolescents with ADHD; Ritalin may affect growth.

Considerations for special patient populations - Ritalin should be given cautiously to patients with a history of drug dependence or alcoholism. Chronic abusive use can lead to marked tolerance and psychological dependence with varying degrees of abnormal behavior. Careful supervision is required during withdrawal from abusive use, since severe depression may occur. Withdrawal following chronic therapeutic use may unmask symptoms of the underlying disorder that may require follow-up. Ritalin falls in Pregnancy Category C.

Concerta

Medication notes - Concerta is a CNS stimulant. A typical starting dose for children and adolescents is 18 mg once daily. Side effects of Concerta may include the following: decreased appetite, headache, dry mouth, nausea, insomnia, anxiety, dizziness, weight loss, irritability, and hyperhidrosis.

Safety notes - Contraindications for Concerta include: known hypersensitivity to the product, marked anxiety, tension, or agitation, glaucoma, tics or a family history or diagnosis of Tourette's syndrome, use within 2 weeks of using an MAOI. Warnings associated with Concerta include: Concerta should be given cautiously to patients with a history of drug dependence or alcoholism; chronic abusive use can lead to marked tolerance and psychological dependence, with varying degrees of abnormal behavior; sudden death has been reported in association with CNS stimulant treatment at usual doses in children and adolescents with structural cardiac abnormalities or other serious heart problems; monitor patients for changes in heart rate and blood pressure and use with caution in patients for whom an increase in blood pressure or heart rate would be problematic.

Considerations for special patient populations - Caution should be exercised if administered to nursing mothers. Health care professionals should note the safety and

efficacy of Concerta has not been established in children less than six years old or elderly patients greater than 65 years of age.

Adderall XR

Medication notes - Adderall XR is a CNS stimulant. A typical dose of Adderall XR for individuals ages 6 - 17 is 10 mg once daily in the morning. The maximum dose for children 6 - 12 is 30 mg once daily. A typical adult dose of Adderall XR is 20 mg once daily in the morning. Side effects of Adderall XR include: loss of appetite, insomnia, abdominal pain, emotional lability, vomiting, nervousness, nausea, and fever.

Safety notes - Adderall XR is contraindicated in patients with advanced arteriosclerosis, symptomatic cardiovascular disease, moderate to severe hypertension, hyperthyroidism, known hypersensitivity or idiosyncrasy to the sympathomimetic amines, glaucoma, agitated states, history of drug abuse, and during or within 14 days following the administration of an MAOI. Warnings for Adderall XR include the following: amphetamines have a high potential for abuse; prolonged administration may lead to dependence; misuse of amphetamines may cause sudden death and serious cardiovascular adverse reactions. Additional warnings associated with Adderall XR include: sudden death has been reported with usual doses of CNS stimulants in children and adolescents with structural cardiac abnormalities or other serious heart problems; sudden death, stroke, and myocardial infarction have been reported in adults taking CNS stimulants at usual doses; stimulant drugs should not be used in patients with known structural cardiac abnormalities, cardiomyopathy, serious heart rhythm abnormalities, coronary artery disease, or other serious heart problems; monitor blood pressure and pulse at appropriate intervals. Use with caution in patients for whom blood pressure increases may be problematic; stimulants may cause treatment emergent psychotic or manic symptoms in patients with no prior history, or exacerbation of symptoms in patients with pre-existing psychosis; evaluate for bipolar disorder prior to stimulant use; monitor for aggressive behavior; discontinue in the presence of seizures; may exacerbate tics; evaluate for tics and Tourette's syndrome prior to stimulant administration.

Considerations for special patient populations - Adderall XR should only be used during pregnancy if the potential benefit justifies the potential risk to the fetus. Nursing mothers should refrain from breastfeeding when taking Adderall XR.

Section 2: Summary

It is important for health care professionals to possess insight into Fragile X syndrome treatment options. Treatment options for Fragile X syndrome include: physical therapy, speech-language therapy, psychotherapy, cognitive behavioral therapy,

support groups, and the development of sleep routines as well as the use of medications. Some of the most widely prescribed medications used in Fragile X syndrome-related treatment include the following: Celexa, Prozac, Zoloft, Risperdal, Zyprexa, Ritalin, Concerta, and Adderall XR. Possessing insight into the aforementioned treatment options can help health care professionals safely and effectively administer health care to patients with Fragile X syndrome.

Section 2: Key Concepts

- It is vital for health care professionals to possess insight into Fragile X syndrome treatment options.
- Non-pharmacological treatment options for Fragile X syndrome may include: physical therapy, speech-language therapy, psychotherapy, cognitive behavioral therapy, support groups, and the development of sleep routines.
- Pharmacological Fragile X syndrome-related treatment options may include the following medications: Celexa, Prozac, Zoloft, Risperdal, Zyprexa, Ritalin, Concerta, and Adderall XR.

Section 2: Key Terms

Physical therapy - the practice of treating a disease, condition, disorder, and/or injury through physical means²

Speech-language therapy - a form of therapy that addresses issues with speech, language, and communication²

Psychotherapy (also known as talk therapy) - the use of psychological techniques and/or psychotherapeutic approaches to help individuals overcome problems and develop healthier habits⁵

Cognitive behavioral therapy - a form of psychotherapy which focuses on helping individuals solve problems and create positive outcomes by changing unrealistically negative patterns of thought and behavior⁶

Section 2: Personal Reflection Question

What therapeutic options may be used to treat patients with Fragile X syndrome?

Section 3: Case Studies Revisited

The three case studies presented at the beginning of this course will be revisited in this section to further explore the concepts found in this course. Each case study will be presented again below followed by a case study review. The case study review includes the types of questions health care professionals should ask themselves when considering Fragile X syndrome and how it relates to the administration of health care. Additionally, reflection questions will be posed, within the case study review, to encourage further internal debate and consideration regarding the presented case study and Fragile X syndrome. The information found within this section was derived from materials provided by the CDC, the United States Department of Health & Human Services, the American Academy of Pediatrics, the Joint Commission, and the FDA^{1,2,3,4,7}

Case Study 1

A 16-year-old male patient presents with complaints of anxiety. The patient has no known drug allergies and is not currently taking any medications. Upon questioning, the patient reports that he has been "very anxious of late" and has not been sleeping well for the past several months. The patient also reports that he has been "having trouble in school" and "trouble making friends." Further questioning reveals that the patient has "always had trouble learning new things" and often avoids social situations. As the patient is talking, a health care professional observes that the patient is avoiding eye-contact and is repeatedly flapping his right hand. The health care professional also observes that the patient is having trouble articulating what he is feeling and speaking clearly. During a physical examination the health care professional notes that the patient has a large head, large ears, and a prominent jaw. The health care professional documents the aforementioned observations. The patient's parents are called into the examination room, at the request of the patient, for a consultation.

Case Study 1 Review

What patient details may be relevant to the possible presence of Fragile X syndrome?

The following patient details may be relevant to the possible presence of Fragile X syndrome: the patient is 16 years old, the patient is male, the patient presents with complaints of anxiety, the patient reports that he has been "very anxious of late," the patient reports that he has not been sleeping well for the past several months, the patient reports that he has been "having trouble in school," the patient reports that he has "always had trouble learning new things," the patient reports that he has

trouble "making friends," the patient reports that he often avoids social situations, the patient avoids eye-contact, the patient repeatedly flaps his right hand, the patient has trouble articulating what he is feeling and speaking clearly, and the patient has a large head, large ears, and a prominent jaw.

Are there any other patient details that may be relevant to the possible presence of Fragile X syndrome; if so, what are they?

How are each of the aforementioned patient details relevant to the possible presence of Fragile X syndrome?

Each of the previously highlighted patient details may be potentially relevant to the possible presence of Fragile X syndrome. The potential relevance of each patient detail may be found below.

The patient is 16 years old - the patient's age is potentially relevant because the physical symptoms associated with Fragile X syndrome typically develop when an individual goes through puberty (males typically begin puberty between the approximate ages of 9 - 14; females typically begin puberty between the approximate ages of 8 - 13).

The patient is male - the patient's gender is relevant because research indicates that Fragile X syndrome is more common in males.

The patient presents with complaints of anxiety/the patient reports that he has been "very anxious of late" - the previous patient details are relevant because they may represent signs/symptoms of Fragile X syndrome.

The patient reports that he has not been sleeping well for the past several months - the previous patient detail is relevant because it may represent a sign/symptom of Fragile X syndrome.

The patient reports that he has been "having trouble in school"/the patient reports that he has "always had trouble learning new things" - the previous patient details are relevant because they may represent signs/symptoms of Fragile X syndrome (i.e., the previous patient details may represent some of the intelligence and learning symptoms associated with Fragile X syndrome; individuals with Fragile X syndrome often have learning disabilities and/or intellectual disability).

The patient reports that he has trouble "making friends"/the patient reports that he often avoids social situations - the previous patient details are relevant because they may represent signs/symptoms of Fragile X syndrome (i.e., the previous patient

details may represent some of the behavioral, social, and emotional symptoms associated with Fragile X syndrome).

The patient avoids eye-contact and is repeatedly flapping his right hand - the previous patient details are relevant because they may represent signs/symptoms of Fragile X syndrome (e.g., the repeated flapping of the right hand may be indicative of a Fragile X syndrome associated repetitive motion).

The patient has trouble articulating what he is feeling and speaking clearly - the previous patient details are relevant because they may represent signs/symptoms of Fragile X syndrome.

The patient has a large head, large ears, and a prominent jaw - the previous patient details are relevant because they may represent signs/symptoms of Fragile X syndrome (i.e., the previous patient details may represent some of the physical symptoms associated with Fragile X syndrome).

What other ways, if any, are the patient details relevant to the possible presence of Fragile X syndrome?

Is it possible the patient in the above case study has Fragile X syndrome?

Based on the information found in the above case study, it does appear possible that the patient may have Fragile X syndrome.

How can a health care professional potentially gather additional patient information to help confirm the possible presence of Fragile X syndrome?

How may a health care professional address any patient/parent questions and/or concerns regarding the possible presence of Fragile X syndrome?

There are a variety of strategies that may be used, by a health care professional, to address any patient/parent questions and/or concerns regarding the possible presence of Fragile X syndrome, including the ones found below.

Calmly and Clearly answer questions - many questions may arise in a parent/patient discussion regarding Fragile X syndrome. It is important for health care professionals to calmly and clearly answer questions that may develop in a Fragile X syndrome discussion to help avoid confusion among parents and/or patients.

Provide Fragile X syndrome-related educational information - parents/patients may not be familiar with Fragile X syndrome. Thus, health care professionals should consider providing parents/patients Fragile X syndrome-related educational information to help address any questions and concerns that may arise. Health care

professionals should note the following when providing Fragile X syndrome-related educational information: it is important not to overwhelm a parent/patient with Fragile X syndrome-related educational information. Finding out a child may have Fragile X syndrome can be overwhelming in and of itself. Thus, health care professionals should not further overwhelm parents with copious amounts of information. Health care professionals should observe parents/patients to ascertain their responses to Fragile X syndrome-related educational information and provide subsequent information accordingly.

Outline the Fragile X syndrome diagnostic process - outlining the Fragile X syndrome diagnostic process to a parent or a patient may help in addressing any future questions or concerns that may arise.

Outline Fragile X syndrome treatment options - some parents and/or patients may initially ask questions regarding Fragile X syndrome treatment options. Therefore, it may be appropriate, in some cases, for health care professionals to outline Fragile X syndrome treatment options. Health care professionals should note the following: when reviewing Fragile X syndrome treatment options, health care professionals should include both non-pharmacological and pharmacological therapies, when applicable.

Provide information regarding the "next step" for parents/patients - parents and/or patients may not know how to proceed with obtaining further Fragile X syndrome-related health care. Providing clear information regarding the "next step" for Fragile X syndrome-related health care can be both informational and comforting for parents and patients.

What other strategies may be used to address any patient/parent questions and/or concerns regarding the possible presence of Fragile X syndrome?

Case Study 2

A mother and a father present with their 38-month-old son. Upon questioning, the patient's parents report that their son had difficulties learning to crawl, walk, and speak. They also reveal that their son seems to be "very sensitive" to the way things smell, taste, look, and feel and often avoids eye-contact, has trouble maintaining focus, and easily becomes aggressive. A health care professional listens to the mother and father and conducts a developmental screening. At the conclusion of the developmental screening, the health care professional refers the patient and his parents to a genetic counselor.

Case Study 2 Review

What patient details may be relevant to the possible presence of Fragile X syndrome?

The following patient details may be relevant to the possible presence of Fragile X syndrome: the patient is 38 months old, the patient is male, the patient's parents report that their son had difficulties learning to crawl, walk, and speak, the patient's parents report that their son seems to be "very sensitive" to the way things smell, taste, look, and feel, the patient's parents report that their son often avoids eye-contact, the patient's parents report that their son has trouble maintaining focus, and the patient's parents report that their son easily becomes aggressive.

Are there any other patient details that may be relevant to the possible presence of Fragile X syndrome; if so, what are they?

How are each of the aforementioned patient details relevant to the possible presence of Fragile X syndrome?

Each of the previously highlighted patient details may be potentially relevant to the possible presence of Fragile X syndrome. The potential relevance of each patient detail may be found below.

The patient is 18 months old - the patient's age is potentially relevant because the symptoms of Fragile X syndrome typically appear early in development; often individuals show symptoms of Fragile X syndrome by 36 months to 42 months of age or even earlier.

The patient is male - the patient's gender is relevant because research indicates that Fragile X syndrome is more common in males.

The patient's parents report that their son had difficulties learning to crawl, walk, and speak - the aforementioned patient details may be relevant because it suggests that the patient is having difficulties reaching age-related developmental milestones, which can be an indication of the possible presence of Fragile X syndrome. The aforementioned patient detail may also be relevant because it may indicate the possible presence of some of the earliest signs/symptoms of Fragile X syndrome.

The patient's parents report that their son seems to be "very sensitive" to the way things smell, taste, look, and feel - the previous patient detail is relevant because it may represent a sign/symptom of Fragile X syndrome.

The patient's parents report that their son often avoids eye-contact - the previous patient detail is relevant because it may represent a sign/symptom of Fragile X syndrome.

The patient's parents report that their son has trouble maintaining focus - the previous patient detail is relevant because it may represent a sign/symptom of Fragile X syndrome.

The patient's parents report that their son easily becomes aggressive - the previous patient detail is relevant because it may represent a sign/symptom of Fragile X syndrome.

What other ways, if any, are the patient details relevant to the possible presence of Fragile X syndrome?

Is it possible the patient in the above case study has Fragile X syndrome?

Based on the information found in the above case study, it does appear possible that the patient may have Fragile X syndrome.

How can a health care professional potentially gather additional patient information to help confirm the possible presence of Fragile X syndrome?

How may a health care professional address any patient/parent questions and/or concerns regarding the possible presence of Fragile X syndrome?

There are a variety of strategies that may be used, by a health care professional, to address any patient/parent questions and/or concerns regarding the possible presence of Fragile X syndrome, including the ones found below.

Provide patient education regarding a developmental screening - in this particular case, study a health care professional conducts a developmental screening. Therefore, it may be advantageous for the health care professionals to provide the patient's parents with education regarding the developmental screening and/or the results of the developmental screening to address any questions and/or concerns that may arise.

Provide patient education regarding the role and relevance of a genetic counselor - in this particular case study, a health care professional refers the patient and his parents to a genetic counselor. Therefore, the health care professional should provide some educational information to the patient's parents regarding the role and relevance of a genetic counselor to address any questions and/or concerns that may arise. Health care professionals should note the following: a genetic counselor may refer to an individual who specializes in genetics and provides patients with

counseling, education, and assistance in making decisions regarding genetic-related health care; genetic counselors are often used in Fragile X syndrome diagnostic procedures to provide support to other health care professionals and patients; a genetic counselor may also be called upon to conduct related genetic testing and/or to provide insight into the results of genetic testing.

What other strategies may be used to address any patient/parent questions and/or concerns regarding the possible presence of Fragile X syndrome?

Case Study 3

A 20-year-old female patient presents with complaints of depression. Upon questioning, the patient reveals that she has been "extremely depressed lately" to the point where she cannot "get out of bed sometimes." The patient also reveals that she is "not taking" her Celexa. When asked why she stopped taking Celexa, the patient responded by saying "I don't know." Further questioning reveals that the patient recently had to "drop out of school" because it was "too hard" and her classes were "too much" for her. The patient also reports that she has a history of cutting herself and feels like she wants to hurt herself again. A health care professional documents that aforementioned patient information and the following patient symptoms: the patient appeared to avoid eye contact during questioning, the patient stuttered at times, and the patient appeared to have a narrow face, large ears, and a prominent jaw. The patient is admitted into the health care facility.

Case Study 3 Review

What patient details may be relevant to the possible presence of Fragile X syndrome?

The following patient details may be relevant to the possible presence of Fragile X syndrome: the patient is 20 years old, the patient is female, the patient reports that she has been "extremely depressed lately" to the point where she cannot "get out of bed sometimes," the patient reports that she is "not taking" her Celexa, when asked why she stopped taking Celexa, the patient responded by saying "I don't know," the patient reports that she recently had to "drop out of school" because it was "too hard" and her classes were "too much" for her, the patient reports that she has a history of cutting herself and feels like she wants to hurt herself again, and a health care professional observes the following patient symptoms: the patient appeared to avoid eye contact during questioning, the patient stuttered at times, and the patient appeared to have a narrow face, large ears, and a prominent jaw.

Are there any other patient details that may be relevant to the possible presence of Fragile X syndrome; if so, what are they?

How are each of the aforementioned patient details relevant to the possible presence of Fragile X syndrome?

Each of the previously highlighted patient details may be potentially relevant to the possible presence of Fragile X syndrome. The potential relevance of each patient detail may be found below.

The patient is 20 years old - the patient's age is potentially relevant because the physical symptoms associated with Fragile X syndrome typically develop when an individual goes through puberty (males typically begin puberty between the approximate ages of 9 - 14; females typically begin puberty between the approximate ages of 8 - 13).

The patient is female - the patient's gender is relevant because research indicates that males who have Fragile X syndrome usually have some degree of intellectual disability that can range from mild to severe, while females with Fragile X syndrome can have normal intelligence or some degree of intellectual disability (i.e., Fragile X syndrome may have less impact on a female's intelligence, and allow for a potential Fragile X syndrome diagnosis later in development).

The patient reports that she has been "extremely depressed lately" to the point where she cannot "get out of bed sometimes" - the previous patient detail may be relevant because it may support the presence of depression, which is often associated with Fragile X syndrome. Health care professionals should note the following when considering Fragile X syndrome associated depression: major depressive disorder may refer to a form of depression that occurs most days of the week for a period of two weeks or longer leading to clinically significant distress or impairment in social, occupational, or other important areas of functioning; symptoms of major depressive disorder may include: depressed mood, a loss of interest in previously enjoyable activities, appetite changes, weight changes, sleep difficulties, psychomotor agitation or retardation, fatigue or loss of energy, diminished ability to think or concentrate, feelings of worthlessness or excessive guilt, and suicidality.

The patient reports that she is "not taking" her Celexa, when asked why she stopped taking Celexa, the patient responded by saying "I don't know" - the previous patient detail may be relevant because it may further support the presence of depression, which is often associated with Fragile X syndrome. Health care professionals should note that Celexa is a SSRI that may be used to treat depression; patients with Fragile X syndrome may be treated in some capacity with SSRIs.

The patient reports that she recently had to "drop out of school" because it was "too hard" and her classes were "too much" for her - the previous patient detail is

relevant because it may point to the presence of signs/symptoms of Fragile X syndrome (i.e., the previous patient detail may represent the presence of some of the intelligence and learning symptoms associated with Fragile X syndrome; individuals with Fragile X syndrome often have learning disabilities and/or intellectual disability).

The patient reports that she has a history of cutting herself and feels like she wants to hurt herself again - the previous patient detail is relevant because it may represent a behavioral, social, and emotional symptom associated with Fragile X syndrome (e.g., causes self injury).

The previous patient detail may also be relevant because it may support the presence of suicidal ideation. Suicidal ideation may refer to thoughts of suicide and/or thoughts of planning suicide. Health care professionals should be very aware that individuals suffering from Fragile X syndrome associated depression may be suicidal. Health care professionals should make every effort to identify the potential for suicide and prevent patient suicide when applicable. Health care professionals should also note the following: the suicide of a patient while in a staffed, round-the-clock care setting is a frequently reported type of sentinel event; identification of individuals at risk for suicide while under the care of or following discharge from a health care organization is an important step in protecting these at-risk individuals; regarding patients at risk for suicide, health care professionals and health care organizations should follow the following recommendations: identify patients at risk for suicide, conduct a risk assessment that identifies specific patient characteristics and environmental features that may increase or decrease the risk for suicide, address the patient's immediate safety needs and most appropriate setting for treatment, and when a patient at risk for suicide leaves the care of the health care facility, provide suicide prevention information (such as a crisis hotline) to the patient and his or her family.

A health care professional observes the following patient symptoms: the patient appeared to avoid eye contact during questioning, the patient stuttered at times, and the patient appeared to have a narrow face, large ears, and a prominent jaw - the previous patient details are relevant because they may represent symptoms of Fragile X syndrome (i.e., the previous patient details may represent some of the behavioral, social, and emotional symptoms, speech and language symptoms, and physical symptoms associated with Fragile X syndrome).

What other ways, if any, are the patient details relevant to the possible presence of Fragile X syndrome?

Is it possible the patient in the above case study has Fragile X syndrome?

Based on the information found in the above case study, it does appear possible that the patient may have Fragile X syndrome.

How can a health care professional potentially gather additional patient information to help confirm the possible presence of Fragile X syndrome?

How may a health care professional address any patient questions and/or concerns regarding the possible presence of Fragile X syndrome?

There are a variety of strategies that may be used, by a health care professional, to address any patient questions and/or concerns regarding the possible presence of Fragile X syndrome, including the ones found below.

Provide patients with information regarding relevant Fragile X syndrome associated conditions/disorders (e.g., depression) - in this particular case study the patient may be suffering from Fragile X syndrome associated depression. Thus, it may be advantageous to provide the patient with relevant information regarding depression.

Provide patients with information regarding medication adherence - in this particular case study the patient stopped taking a prescribed medication. Therefore, it may be advantageous to the patient's treatment to address the importance of medication adherence.

Address potential patient suicidal ideation - in this particular case study the patient made comments that reflect potential suicidal ideation. Thus, health care professionals involved in the patient's care should follow the following recommendations: identify patients at risk for suicide, conduct a risk assessment that identifies specific patient characteristics and environmental features that may increase or decrease the risk for suicide, address the patient's immediate safety needs and most appropriate setting for treatment, and when a patient at risk for suicide leaves the care of the health care facility, provide suicide prevention information (such as a crisis hotline) to the patient and his or her family.

What other strategies may be used to address any patient questions and/or concerns regarding the possible presence of Fragile X syndrome?

Section 3: Summary

Health care professionals should work to identify those individuals who may have Fragile X syndrome. When attempting to identify individuals with Fragile X syndrome,

health care professionals should differentiate patient details that may be relevant to the possible presence of Fragile X syndrome, observe patients and patient symptoms, ask relevant questions, work to recognize the presence of conditions/disorders typically associated with Fragile X syndrome, effectively document patient information, and address any questions and/or concerns patients/parents may have. Health care professionals should note that, often, one of the main goals of identifying individuals with Fragile X syndrome is to facilitate the safe and effective administration of health care.

Section 3: Key Concepts

- Health care professionals should work to identify those individuals who may have Fragile X syndrome.
- When attempting to identify individuals with Fragile X syndrome, health care professionals should differentiate patient details that may be relevant to the possible presence of Fragile X syndrome, observe patients and patient symptoms, ask relevant questions, work to recognize the presence of conditions typically associated with Fragile X syndrome, effectively document patient information, and address any questions and/or concerns patients/parents may have.
- Strategies that may be use, by a health care professional, to address any patient/parent questions and/or concerns regarding the possible presence of Fragile X syndrome, my include: calmly and clearly answer questions, provide Fragile X syndrome-related educational information, outline the Fragile X syndrome diagnostic process, outline Fragile X syndrome treatment options, provide patient education regarding the role and relevance of a genetic counselor, provide patients with information relevant Fragile X syndrome associated conditions/disorders (e.g., depression), provide patients with information regarding medication adherence, address potential patient suicidal ideation.
- One of the main goals of identifying individuals with Fragile X syndrome is to facilitate the safe and effective administration of health care.

Section 3: Key Terms

Genetic counselor - an individual who specializes in genetics and provides patients with counseling, education, and assistance in making decisions regarding genetic-related health care

Section 3: Personal Reflection Question

Why is it important for health care professionals to recognize relevant patient details when attempting to identify individuals with Fragile X syndrome?

Conclusion

Fragile X syndrome may refer to a genetic disorder that affects development. The major symptoms of Fragile X syndrome fall into the following categories: intelligence and learning symptoms, physical symptoms, behavioral, social, and emotional symptoms, speech and language symptoms, and sensory symptoms. The diagnostic process for Fragile X syndrome typically involves two major stages or steps, which include a developmental screening and a comprehensive diagnostic evaluation. The physical and mental-health conditions typically associated with Fragile X syndrome include the following: anxiety, depression, ADHD, ASD, and intellectual disability. Non-pharmacological treatment options for Fragile X syndrome may include: physical therapy, speech-language therapy, psychotherapy, cognitive behavioral therapy, support groups, and the development of sleep routines. Pharmacological Fragile X syndrome-related treatment options may include the following medications: Celexa, Prozac, Zoloft, Risperdal, Zyprexa, Ritalin, Concerta, and Adderall XR. Finally, health care professionals should work to identify those individuals who may have Fragile X syndrome in order to facilitate the safe and effective administration of health care.



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