DEPARTMENT OF HUMAN GENETICS

Men's Health and the Fragile X Premutation

A GUIDE FOR CARRIERS, CAREGIVERS, AND HEALTH PROFESSIONALS

FRAGILE X

EMORY
UNIVERSITY
SCHOOL OF
MEDICINE

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01 > Introduction

Up to 1 in 450 men are carriers of a genetic change called the fragile X premutation. Many people know about this type of genetic change in the fragile X gene because a family member may have fragile X syndrome, a type of intellectual disability. With increased awareness, more and more people are being diagnosed with the other disorders associated with the fragile X gene. Families are learning that they need to be aware of all of the health conditions that are related to the fragile X gene.

This brochure is focused on the health of men who carry a premutation. Both men and women can be affected by this genetic change, but in different ways. So it is important for men to learn about potential health problems that are specific to a fragile X premutation. It is a guide for carriers, for their families and health care providers.

This book is organized in a questions and answer format. It is divided into 4 main sections: 1) The Fragile X Premutation: What is it and how does it work?; 2) Fragile X Tremor/Ataxia Syndrome (FXTAS): What are the symptoms and what should I expect?; 3) Testing for Fragile X and FXTAS: Who should be tested and how?; and 4) How can I learn more about FXTAS?


Men who carry a fragile X premutation face specific health concerns.

02 > The Fragile X Premutation

What are genes?

Our bodies are made of trillions of cells. Within each cell is our genetic material, called DNA. DNA is made of a series of small molecules. We label these molecules A, C, G, or T.

Sections of DNA that contain the information needed for our bodies to work properly are called genes. It is sometimes helpful to think of our genes as a set of instructions for building proteins that our bodies need.

Our DNA is packaged into long strands called chromosomes. We have 23 pairs of chromosomes in each of our cells. We receive one set of 23 chromosomes from one parent, and one set from our other parent. One of these 23 pairs is different and is called the sex chromosomes. Females typically have two X chromosomes (XX) and males typically have one X and one Y chromosome (XY).

What happens if a gene is not working correctly?

We all have differences in our DNA. This is part of what makes us unique. If a difference occurs in one of our genes, it can sometimes affect the way that gene works.

These errors are sometimes called "mutations." The errors may act like a misspelling, or a missing step, or even extra words in the instructions. Each of these types of changes can affect how the protein is built.

If a protein is built differently due to a genetic change, it may cause a single health problem, or it may cause many health problems. This all depends on the gene that is not working properly and the type of mutation in that gene. Mutations in the fragile X gene are types of changes that can affect different functions in the body.

What is a fragile X premutation?

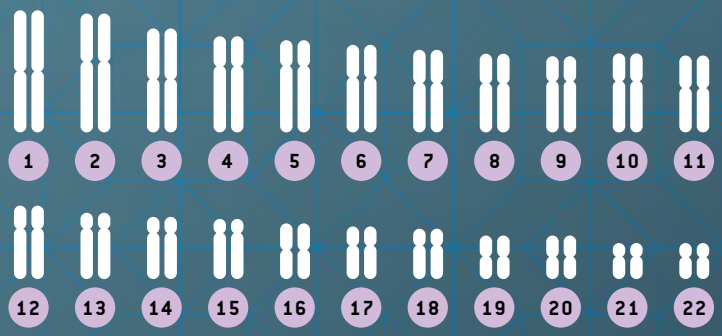
The fragile X premutation is one type of mutation within the fragile X gene. The fragile X gene is sometimes called the *FMR1* gene. It is located on the X chromosome, one of the sex chromosomes. This gene contains a short section of repeated letters of the DNA alphabet (or code). The repeated letters are "CGG" and are part of the beginning of the gene. The term "repeat expansion" refers to an extra number of repeated CGGs in this part of the gene.

Everyone has some number of repeats within this gene. Most people have between 20 and 35 repeated copies of three letters (CGG) within their *FMR1* gene. People who have a fragile X premutation carry between 55 and 200 copies of this CGG repeat. Up to 1 in 450 men carry a fragile X premutation. People who have a premutation have a higher chance to get specific health problems. The number of repeats in this premutation range is related to the chance to develop certain health conditions. For more information about premutation-related health conditions turn to page 10.

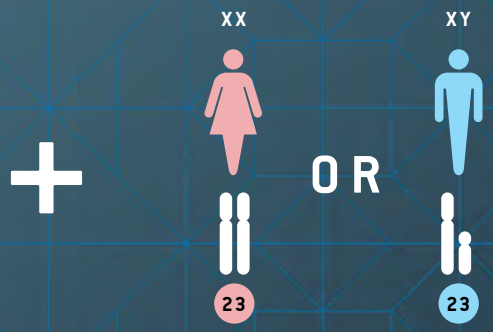
Individuals with over 200 repeats have fragile X syndrome. This type of mutation is known as a "full mutation." The full mutation stops the fragile X protein from being made. The full mutation causes very different changes in the cells compared with the premutation. See page 14 for more information about the cell changes related to a premutation.

Everyone inherits 23 pairs of chromosomes.

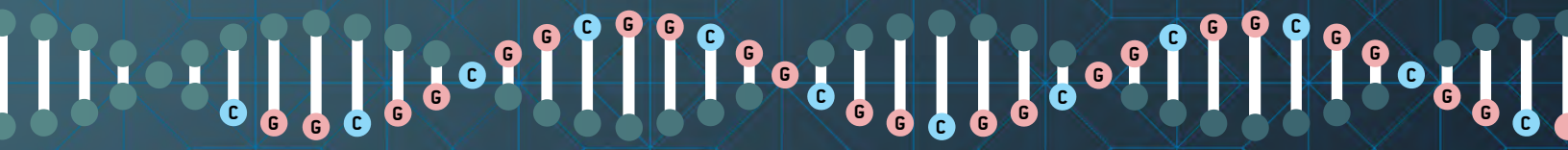
The chromosomes numbered 1 to 22 are called **autosomes**…



…and the last pair are the **sex chromosomes**.

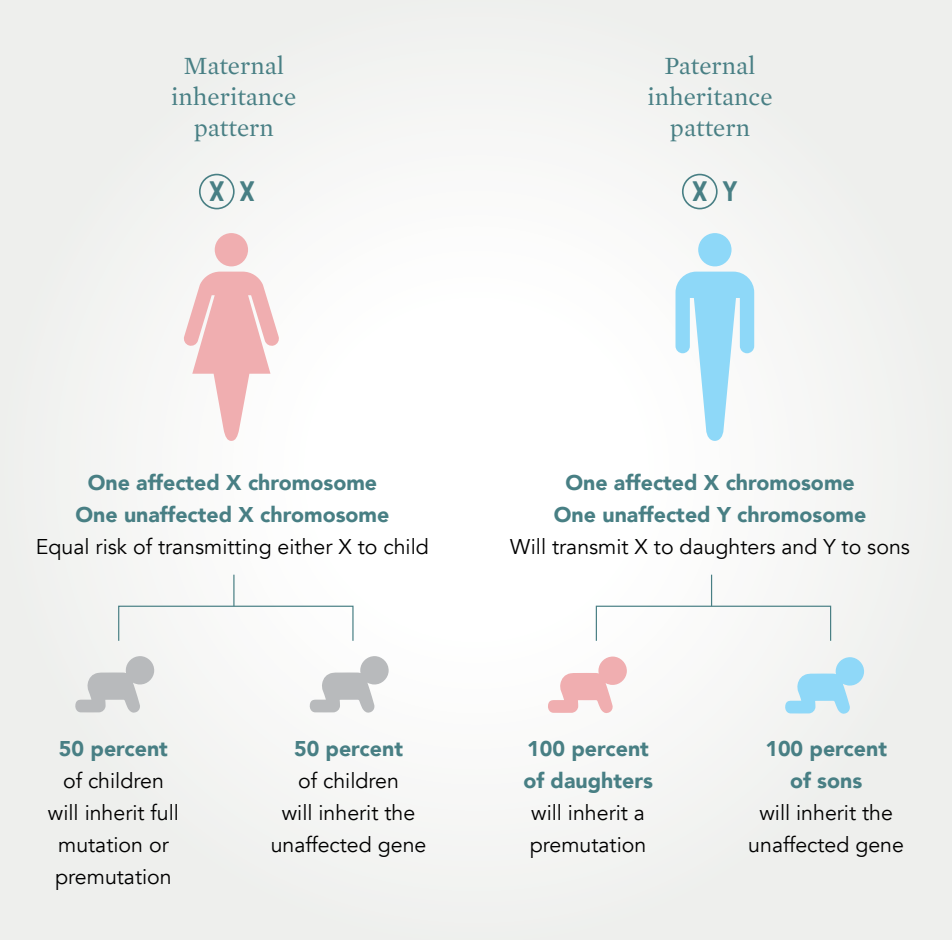


Fragile X carriers have a mutation on their X chromosome that causes an unusual number of repeats of CGG within the code that makes up their *FMR1* gene.



People who have a fragile X premutation carry between **55 and 200 copies** of this CGG repeat.

Fragile X syndrome occurs when the repeat expansion of the *FMR1* gene is over 200 repeats. This is called the “full mutation.” Fragile X syndrome includes intellectual disability and behavioral challenges like autism, ADHD and social anxiety. Fragile X syndrome is a developmental disorder (meaning it starts early in life). It is more severe in males compared with females.



How did I get the premutation?

The repeat expansion is passed through families. The *FMR1* gene is located on the X chromosome. The X and Y chromosomes are called the sex chromosomes. This is because they are involved in determining biological sex. Males inherit an X chromosome from their mother and a Y chromosome from their father. Females inherit an X chromosome from each of their parents. This is called "X-linked" inheritance. Because the *FMR1* gene is on the X chromosome, there is a different inheritance pattern for men and women.

Let's start with a woman who carries a premutation. Typically, a carrier woman has a premutation on only one of her X chromosomes and a normal number of repeats on the other. So, when a woman who carries a premutation has children, she will either pass on the X chromosome that carries a premutation or the X chromosome that does not carry the premutation. There is a 50% chance of passing on either X chromosome. This is true for each pregnancy. If she passes on the X chromosome that carries a premutation, there is a chance that it will expand to a higher number of repeats. It can expand to a larger premutation or to a full mutation. The size of the expansion will differ with each pregnancy. Remember, fragile X syndrome is caused by the number of repeats expanding to greater than 200.

A man who carries a premutation only has one X chromosome. This means he will pass on his X chromosome (which carries the premutation) to all of his daughters and to none of his sons. His sons will receive his Y chromosome. When a man passes on his X chromosome that carries a premutation to his daughters, it rarely, if ever, expands to the full mutation. This means that a man who carries a premutation will not have a daughter with fragile X syndrome. However, his daughter will carry a premutation. The daughter's premutation can sometimes be larger and sometimes be smaller than her father's premutation.

What factors lead to the expansion of the premutation from parent to child?

The chance that the number of repeats expands to a larger number when it is passed down from parent to child depends on a few factors. First, it depends on the number of repeats in the premutation of the parent. A premutation with more repeats will have a higher chance of expanding in the next generation. Second, the CGG repeat sometimes includes a different repeat, an "AGG." This interruption of the CGGs by an AGG reduces the chance of a large expansion. The more AGG interruptions a person has, the smaller that chance of expanding to a larger number of repeats. Genetic counselors can provide information about the chance that a premutation will expand when passed from parent to child. Also, see genetic testing on page 24 for more details.

It is important to talk about your test results with your relatives who may also be at risk for carrying a fragile X mutation. A genetic counselor can help you understand who might be at risk in your family and can also help you talk with your family about this information.

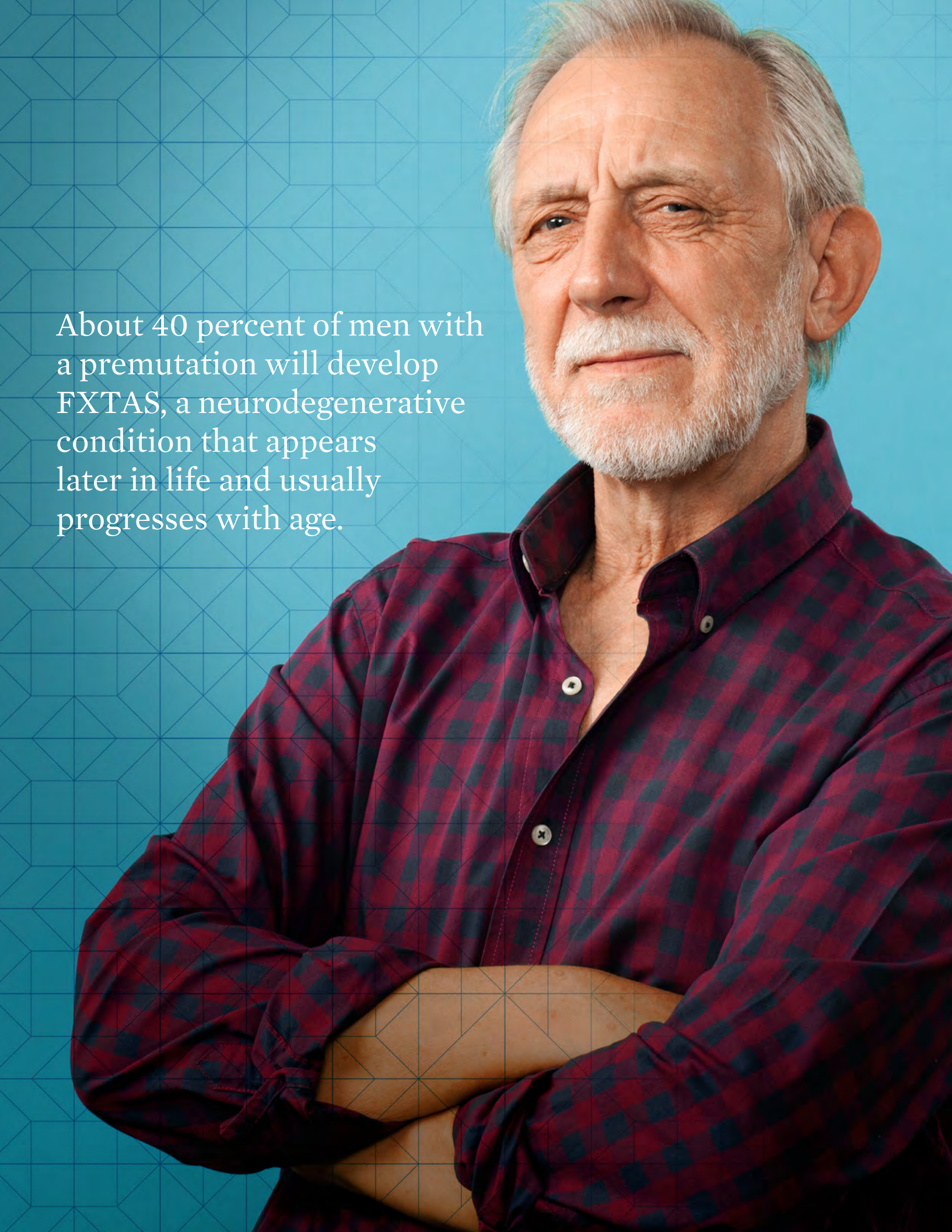
How do I know if I have a premutation?

A fragile X premutation can be detected through genetic testing, usually done with a simple blood test. Your doctor or genetic counselor can order a specific type of genetic test that looks at the number of repeats in the *FMR1* gene. Your genetic test report will have information about the name of the test, the lab that performed the test, and the method used in the test. The "results" section of your genetic test will show how many repeats you have in your *FMR1* gene. If your repeat number falls within 55–200 repeats, this means that you have a premutation.

Why do I need to know if I have a premutation?

Men who have a premutation will pass it on to all of their daughters and none of their sons. It is important for the family to understand this inheritance pattern. Also, if you carry a premutation, you may be at risk for specific health conditions. Knowing if you are a carrier will help you and your doctor build a health plan.

Fragile X is a family diagnosis.


About 40 percent of men with a premutation will develop FXTAS, a neurodegenerative condition that appears later in life and usually progresses with age.

03 > Fragile X Tremor/Ataxia Syndrome (FXTAS)

What health concerns should I be aware of?

Men who have a premutation have a chance to develop a condition called fragile X-associated tremor/ataxia syndrome, also known as FXTAS (pronounced FAX-tas).

FXTAS is a neurodegenerative condition. This means that it affects the brain and typically gets worse over time. Symptoms of FXTAS appear later in life and usually progress with age. The three areas that are affected by FXTAS are: 1) movement, 2) mental health, and 3) the nervous system.

Not everyone with a premutation develops FXTAS. Also, not everyone with FXTAS will have the same symptoms or the same severity. Someone with FXTAS may only have some of the symptoms while others might have many.

Why do some premutation carriers develop FXTAS and others do not?

About 40% of men with a premutation develop FXTAS. This means that around 60% of men who have a premutation do not develop FXTAS. Researchers are working hard to figure out why some premutation carriers do develop FXTAS, while others don't. They do know that individuals with a larger number of premutation repeats, usually greater than 70 repeats, have a higher chance of developing FXTAS.

Women who carry a premutation can also develop FXTAS. Their symptoms may be milder or may differ from those seen in men. Up to 17% of women with a premutation may develop FXTAS. The lower risk is because they have another X chromosome with a normal number of repeats. This helps reduce the risk of FXTAS-related symptoms.

Knowing about the potential health risks of FXTAS is important for your health care plan, even if you do not currently have any symptoms.

What are the symptoms of FXTAS?

As mentioned earlier, FXTAS-related symptoms vary widely. If they appear, it is usually not until after age 60, but can start in the 50's. Sometimes symptoms get worse (progress) slowly, but sometimes they may progress quickly. At this point, the reason for these differences is not known.

Here we provide a list of symptoms that may occur and a general description of each.

Movement-related symptoms

ACTION TREMOR

Tremor is involuntary muscle movement. This means that your muscles move when you are not trying to make them move. Action tremor (sometimes called intention tremor) happens when you begin to do an action (such as trying to write with a pencil). Resting tremor occurs when the muscles are at rest and you are not trying to move. Most often, people with FXTAS have action tremors.

ATAXIA

Ataxia is a loss of balance and coordination. Balance problems may lead to needing support when walking or going up or down stairs, being unstable, and sometimes falling. To gain some stability, sometimes people with ataxia will have a wide-based gait (that is, walk with the feet wider apart). Many symptoms of ataxia can look like being drunk like slurred speech, stumbling, falling, and incoordination.

PARKINSONISM

Parkinsonism is describes a combination of the movement problems commonly seen in Parkinson's disease. These may include resting tremor, slow movements, and tensed muscles also called rigidity.

Mental health-related symptoms

APATHY

Apathy is a lack of feelings, emotions, interest or concern. Apathy may affect your behavior and your ability to complete daily activities.

DISINHIBITION

Disinhibition leads to the loss of ability to control your behavior. Someone with this condition may have a lack of impulse control and the inability to "filter" inappropriate comments.

IRRITABILITY AND PERSONALITY CHANGES

Someone with FXTAS may have unstable moods. They may become more irritable or have changes in personality.

DEPRESSION

Depression is a period of sadness lasting for more than two weeks.

COGNITIVE DECLINE OR DEMENTIA

A person with FXTAS may experience a decline in their mental ability. This typically happens in the later stage of the disease. It can include loss of memory (especially short term), difficulty with language, loss of focus, or difficulty with visual perception (the brain's ability to make sense of what the eyes see). Cognitive decline may affect a person's ability to do everyday tasks such as reading or balancing the check book.

Nervous system-related symptoms

NEUROPATHY

Neuropathy means damage to your nerves. This can lead to weakness, numbness, or pain in parts of your body.

ERECTILE DYSFUNCTION

Neuropathy to the nerves of the penis can make it difficult to get or maintain an erection.

BLADDER OR GASTROINTESTINAL (GI) PROBLEMS

Some people with FXTAS have issues with the nerves that control the bladder or bowels, which may lead to incontinence.

ORTHOSTATIC HYPOTENSION

Orthostatic hypotension leads to a feeling of dizziness or lightheaded when they get up from a seated or laying position.

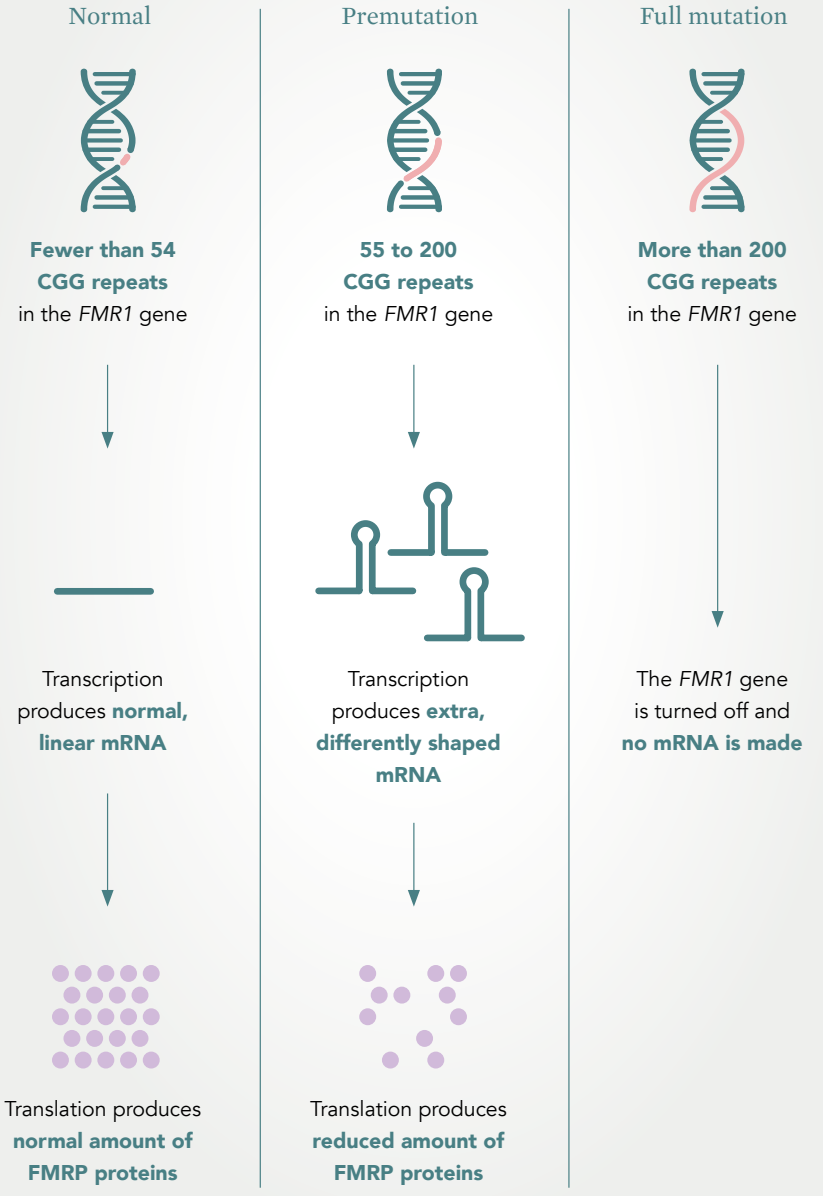
What causes FXTAS?

The exact cause of FXTAS is still unknown. However, research tells us that having a premutation leads to two altered processes in the body. To understand, let's first review more about genes and how they make proteins. Remember that genes provide the DNA code, or instructions, to build a protein. This DNA code has to be transcribed from the DNA "language" to a different "language" that is stored in a molecule called the messenger RNA (mRNA).

mRNA takes the information from the DNA and gives it to machinery that "translates" the code into building blocks that make up proteins. The *FMR1* gene provides the code for its protein, called FMRP. This protein has many functions. One function is to regulate the "translation" of many proteins in the brain that help with development of neurons and their communication. The figure at right shows what happens to the *FMR1* DNA code, the mRNA and FMRP when there is an expansion of the CGG repeats. You can see that the premutation and the full mutation cause completely different results.

The long fragile X premutation CGG repeat is transcribed from the *FMR1* gene to the *FMR1* mRNA. This leads to extra *FMR1* mRNA being produced. Also, the mRNA that is produced looks different because of the long CGG repeat. Instead of remaing straight, it folds back on itself, creating a loop (sometimes called a "hairpin").

Differences in the number of CGG repeats in the DNA sequence cause differences in the way the body creates proteins needed for crucial processes.



This premutation mRNA may be "toxic"to the cell in two different ways. First, the folded mRNA can attract (or sequester) other proteins to bind on to it. This causes the proteins to clump together to the folded mRNA. The proteins then are not able to do their normal job in a cell.

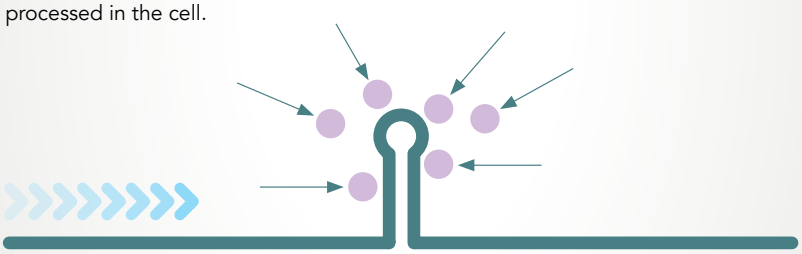
Second, that folded part of the mRNA can cause the "translation" machinery to start building a new protein at the wrong place. Instead of adding building blocks from the beginning of the instructions, the machinery starts building the protein from the wrong step. This means that new proteins are being made that should not be in the cell.

These two events (the attraction of proteins to the folded mRNA, and the building of incorrect proteins) cause the cell to have problems. This can cause the cell to die earlier than normal. Researchers are focusing on these events and how they may be targeted for prevention or for treatment.

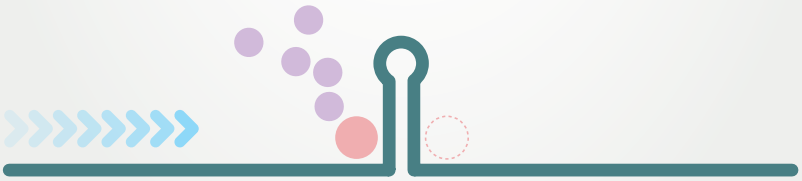
mRNA takes the information from the DNA and gives it to machinery that translates the code into building blocks that make up proteins.

Normal mRNA forms in a straight line to present the code for translation.

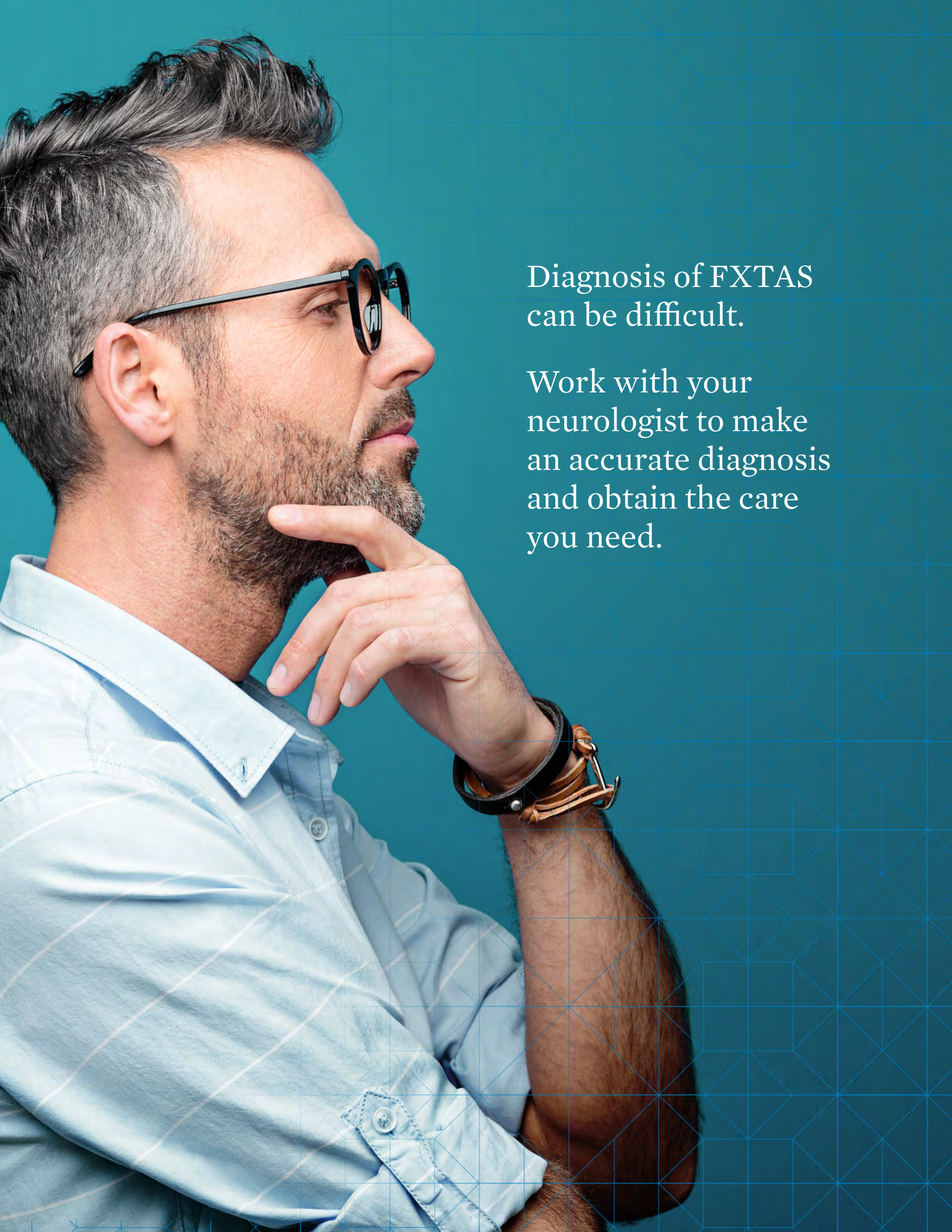
But the CGG repeats in the DNA causes the mRNA to **fold back onto itself in a "hairpin" formation**. This change in shape alters the way some proteins are processed in the cell.



For instance, the folded mRNA attracts, or **"sequesters" some proteins** away from other necesary cellular functions. The proteins then are not able to do their normal job.



Also, the folded mRNA can cause the translation machinery to build new proteins in **the wrong place on the mRNA strand**. These new proteins do not belong in the cell.



Diagnosis of FXTAS can be difficult.

Work with your neurologist to make an accurate diagnosis and obtain the care you need.

How do I know if I have FXTAS?

For an adult who carries a premutation, FXTAS can be diagnosed by a neurologist by checking a list of specific symptoms. The neurologist will need to take a family history, conduct a neurological examination, and request a magnetic resonance image (MRI) to be done. Symptoms related to FXTAS are divided into two groups: "major" and "minor" symptoms. Major symptoms are almost always seen in individuals with FXTAS. Minor symptoms might be seen in individuals with FXTAS, but are also sometimes part of normal aging in everyone.

	MINOR SYMPTOMS	MAJOR SYMPTOMS
CLINICAL FINDINGS	Moderate to severe short-term memory deficit Executive function deficit Neuropathy Parkinsonism	**Intention tremor** **Gait ataxia**
BRAIN FINDINGS (MRI)	MRI lesions involving cerebral white matter Moderate to severe generalized brain atrophy (loss of cells in the brain)	**MRI white matter lesions in the middle cerebellar peduncles** **MRI white matter lesions in the splenium of the corpus callosum**

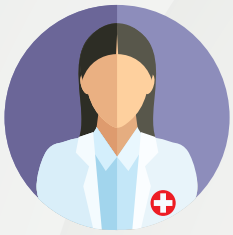
The diagnosis can sometimes be hard because there are many reasons for symptoms, including natural aging. Because of this, the FXTAS diagnosis maybe be "definite", "probable" or "possible". This table describes the diagnostic criteria that relates to the confidence of a diagnosis. As we gain more knowledge about FXTAS and how it presents, these criteria may change. It is important for you and your neurologist to work together to make sure you have an accurate diagnosis and supportive care.

	DIGNOSTIC CRITERIA
POSSIBLE	Presence of 1 major radiological sign plus 1 major clinical symptom
PROBABLE	**Presence of either 1 major radiological sign plus 1 major clinical symptom or 2 major clinical symptoms**
DEFINITE	**Presence of 1 major radiological sign plus 1 major clinical symptom**

What should I do if I am experiencing any of these symptoms?

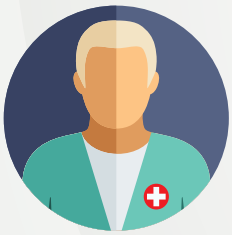
Individuals with FXTAS can benefit from having a healthcare team made up of various specialists. All individuals with FXTAS should see a neurologist. A neurologist with experience in movement disorders can help treat some of the symptoms of FXTAS.

Your healthcare team will depend on your specific symptoms and diagnosis, and may include specialists in the following fields:



GENETIC COUNSELOR

Genetic counselors are healthcare providers who can help individuals and their families understand the genetics of the fragile X mutation and adapt to the medical, psychological and familial aspects of the associated disorders.



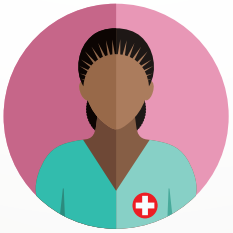
PHYSICAL THERAPIST

Physical therapists are experts in physical movement. They will help to improve quality of life through exercise, hands-on care, and patient education.

NEUROLOGIST

All individuals with FXTAS should see a neurologist. A neurologist with experience in movement disorders can help treat some of the symptoms of FXTAS.

YOU



OCCUPATIONAL THERAPIST

Occupational therapists help people fully take part in daily life. This includes activites at home, at work and at play. Their goal is to promote a person's independence as much as possible.

SPEECH AND LANGUAGE THERAPIST

Speech and language therapists can help prevent and treat problems with speech, language, and swallowing.

SOCIAL WORKER

Social workers help individuals, families, and their communities develop skills to use their resources to solve problems in their own life and in their social lives. Their goal is to increase overall well-being.

What can I expect if I am diagnosed with FXTAS?

FXTAS is a progressive disease. This means that the symptoms may get worse over time. Not everyone with FXTAS will have the same experience. Some people may have a very slow and stable progression of the disease. Others may decline more quickly.

The first symptom of FXTAS is usually action tremors in their upper limbs. Tremor means that your muscles move without you telling them to. Some signs of tremor might be hard to notice at first. You may have difficulty keeping your hands steady when you do an activity. As the tremor gets worse, people may have a hard time with activities of daily living. These may include writing, eating, dressing yourself, or bathing yourself. If the tremor gets much worse, people may need help with their daily activities and tasks.

Following the presence of tremors, people may have trouble with balance and coordination (called ataxia). Sometimes, balance problems are noticed before tremors. Ataxia may cause a person to walk differently or to be less coordinated when doing physical activities. Balance problems can lead to falling. If ataxia gets worse, a person may need a walking aid (like a cane or walker). Some people with severe ataxia may eventually need a wheelchair.

It is difficult to predict if or when someone with FXTAS may develop symptoms of cognitive decline or dementia. If you or your family members are concerned, a neurologist can test your cognitive functioning and suggest possible strategies to reduce the effects of decline.

Does FXTAS have a cure?

There currently is no cure for FXTAS. However, your doctor may be able to treat some of the symptoms of FXTAS to improve daily activities and well-being.

What can I do to slow the progression of FXTAS?

Doctors recommend a healthy lifestyle, with minimal stress. Of course this is true for everyone. But for those at risk for FXTAS, this might be more important.

For men at risk,
a lifestyle that includes
exercise, healthy diet,
and minimizing stress
is especially important.

These conversations are not easy, but it is important to have them while you are able to make your wishes known.

How can I plan for the future?

Not everyone with a premutation will develop FXTAS. And not everyone with a diagnosis of FXTAS will have the same disease progression. However, planning may be helpful for you and your family to be ready for whatever the future holds. Planning the future with FXTAS in mind may be similar to planning for old age, only sooner. Topics to think about may include health-related planning, social planning, home modifications, legal planning, and end of life issues.

Health-related planning involves putting together your health-care professional team. This includes finding a neurologist or movement disorders specialist, getting in touch with a social worker, and speaking with a geneticist or genetic counselor. See page 18 for more information.

Social planning may include discussing caregiving responsibilities and responsibilities around the house such as yard work, maintenance, paying bills, cooking, and cleaning.

Home modifications may help a person with FXTAS stay independent for longer and may prevent accidents. Occupational therapists may be able to help with suggestions of modifications. Not everyone chooses to modify their home. Instead, they may choose to move to a new house (for example, a house with one floor) or to an assisted-living community.

Legal planning involves putting together important documents known as "advance directives." These might include setting up long-term care, creating a living will, or talking about medical and legal power of attorney. Finding a lawyer who specializes in elder-care law may be helpful.

Talking about end of life issues is another part of planning for the future for everyone, not just those with FXTAS. These topics may be difficult to discuss, but it is important to have them while you are able to make your wishes known.

04 > Testing for a Fragile X Mutation and FXTAS

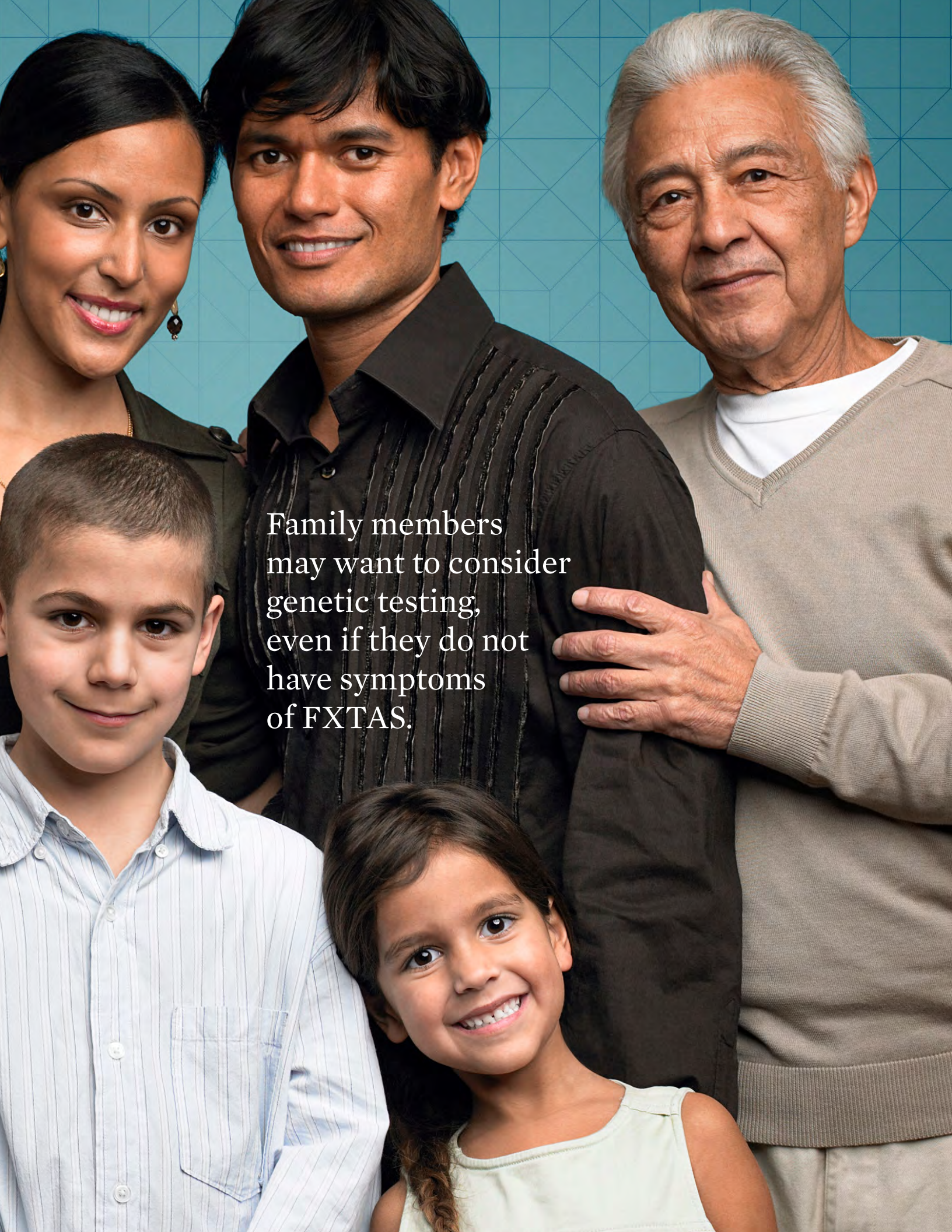
Who should be tested for a fragile X premutation and for FXTAS?

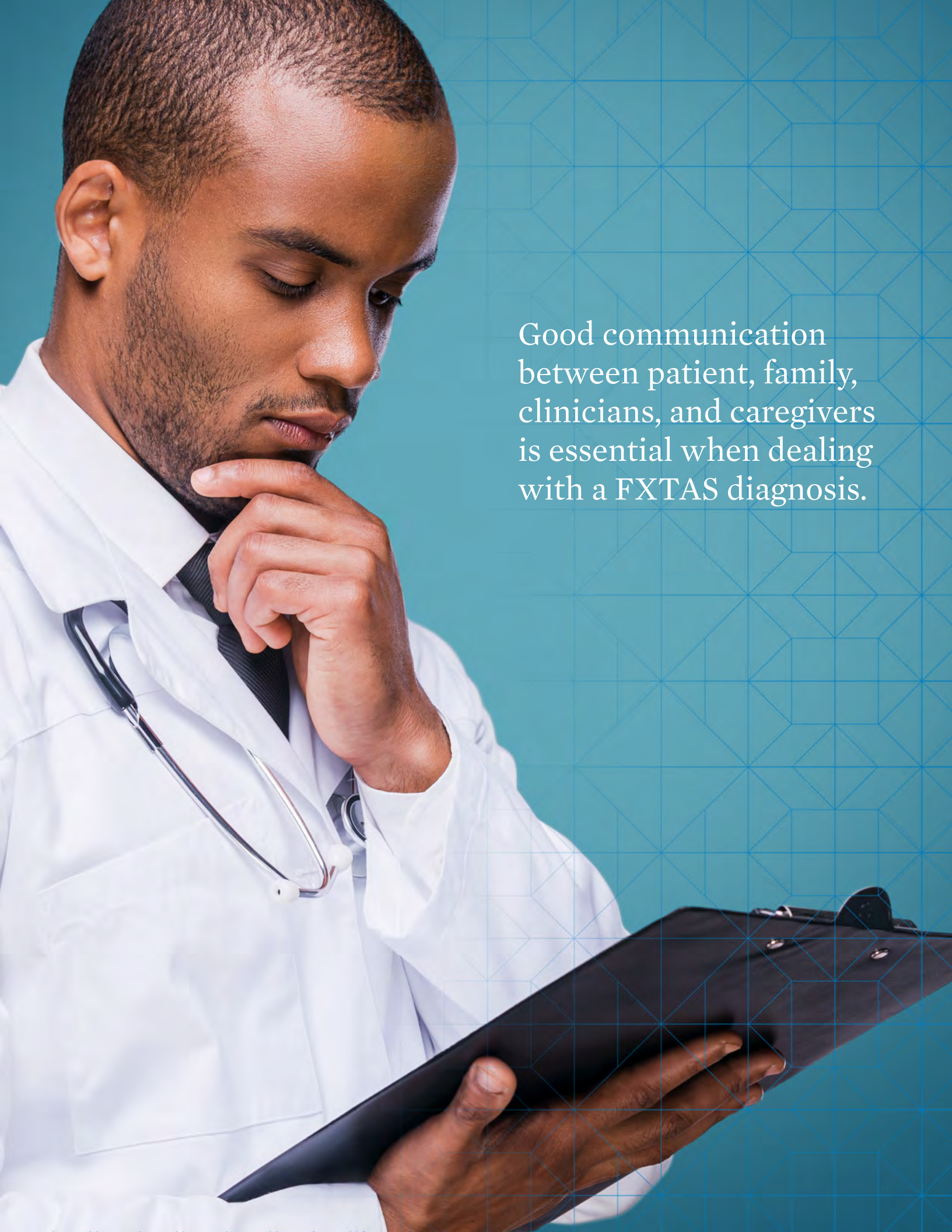
Anyone who has a family member diagnosed with a fragile X-associated disorder or a fragile X mutation may want to be tested. The three well-established fragile X-associated disorders include: fragile X syndrome (due to a full mutation), FXTAS (due to a premutation), and specifically for women, fragile X-associated primary ovarian insufficiency (FXPOI) (due to a premutation). A fragile X mutation can be identified using a simple genetic test. See page 8 for further details.

Sometimes a person may have possible symptoms of FXTAS, but no one in their family has been tested for a fragile X-associated disorder. In this case, their doctor needs to consider all the possible causes for their patient's symptoms, one of which could be FXTAS. A set of guidelines are provided here to help healthcare providers decide whether their patient should be tested for a fragile X premutation. Although there are no specific treatments for FXTAS, it is important for doctors to identify which of their patient's symptoms are caused by a genetic condition. This can help their patient understand the reason for their symptoms and inform their family members. It is also important to connect their patient to specialists and other families who have experience with FXTAS.

Clinicians should test for a fragile X mutation if their patient has any of the following:

CEREBELLAR ATAXIA	If symptoms start at age 50 or older and the cause is unknown.
ACTION TREMOR	If symptoms start at age 50 or older, the cause is unknown **and** if they occur along with cerebellar ataxia, parkinsonism, or cognitive decline.
DEMENTIA	If symptoms start at age 50 or older, the cause is unknown **and** if symptoms also include cerebellar ataxia, parkinsonism, or action tremor.
MULTIPLE SYSTEM ATROPHY CEREBELLAR SUBTYPE	Especially if a prolonged course.
SOME SYMPTOMS PLUS FAMILY HISTORY	Symptoms of FXTAS **and** a family history of ovarian insufficiency (including infertility) **or** intellectual disability **or** autism spectrum disorder **or** family history fragile X syndrome, FXPOI, or FXTAS.


Family members
may want to consider
genetic testing,
even if they do not
have symptoms
of FXTAS.



Good communication between patient, family, clinicians, and caregivers is essential when dealing with a FXTAS diagnosis.

Communicating genetic test results

It is important to share genetic test results with your family members. A geneticist or genetic counselor can help you identify family members who may also carry a fragile X mutation. By sharing this information, you can allow your family members to decide if they would like to get tested themselves.

Whenever you share genetic information, consider four things:

When: Make sure there is enough time to discuss this important information and help answer questions.

Where: If possible, choose a private place with few distractions, so people can pay full attention.

How: This is up to you. If possible, it is best to talk in person. If this doesn't work for you, you could talk by telephone or send a letter that describes the information.

What: Genetic information is hard for most people. It can cause distress. Try to plan what information to share with family members ahead of time. This will make you more confident. It will also make sure you cover all the points you think are important.

Communicating with clinicians

Not all doctors will be familiar with the fragile X-associated disorders and their inheritance. Sharing this booklet with your healthcare providers can help them learn about these topics. It may also be helpful for you to connect your clinician with a genetic counselor, geneticist, or movement disorder specialist familiar with FXTAS. See Resources on page 28.

Caregivers of individuals with FXTAS

It is important for caregivers to also get the support they need. Finding support groups, speaking with friends and family, or talking to other caregivers can be helpful for family members. For caregivers of someone with severe FXTAS, joining dementia or other caregiver support groups can be helpful.

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National Fragile X Foundation

The National Fragile X Foundation is an organization created to support families with fragile X-associated disorders. Their website includes information about the associated disorders, clinical care, and ongoing research. They will help you find a local family organization or a way to connect with other premutation carriers.

HTTPS://FRAGILEX.ORG

Fragile X-Associated Tremor/ Ataxia Syndrome (FXTAS)

This document provides summary information about FXTAS and clinical care. This was provided by the FXTAS Task Force.

HTTPS://FRAGILEX.ORG/FXTAS-CONSENSUS-DOCUMENT

FRAXA Research Foundation

FRAXA Research Foundation supports research to help reduce the effects of the fragile X mutation, with a focus on fragile X syndrome. This website provides information of on-going research and clinical trials.

HTTPS://WWW.FRAXA.ORG

National Society of Genetic Counselors

The National Society of Genetic Counselors website is a great resource to find a genetic counselor in your area.

HTTPS://WWW.NSGC.ORG

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