

# Preparing for Your General Healthcare Visit

Every individual is unique, and the priority should be partnering with a healthcare team to address symptoms and develop a personalized treatment plan based on individual needs.

Knowing that you have an *FMR1* premutation can provide important information about potential health risks and may help you and your healthcare provider determine what concerns may need further evaluation.

Sharing your symptoms, health history, and Fragile X status with your healthcare provider can help support a focused conversation about your health and the concerns you are currently experiencing.

**To prepare for your visit, consider completing the Review of Symptoms checklist below to help you organize your top concerns.** *This checklist is a tool for discussion with a healthcare professional and is not a substitute for medical evaluation.*

## Review of Symptoms Checklist

**Directions:** Check any symptoms you are currently experiencing, have experienced recently, or would like to discuss with your healthcare provider. Add details where indicated. Please review the entire symptom inventory prior to adding “Other” conditions or symptoms.

*Please note, symptoms or conditions that you may be experiencing may or may not be related to the Fragile X premutation.*

| Constitutional / General |                         |         |
|--------------------------|-------------------------|---------|
|                          | Symptom                 | Details |
|                          | Fever or chills         |         |
|                          | Fatigue or low energy   |         |
|                          | Unexplained weight loss |         |
|                          | Unexplained weight gain |         |
|                          | Night sweats            |         |
|                          | Weakness                |         |
|                          | Changes in appetite     |         |
|                          | Trouble sleeping        |         |
|                          | Other _____             |         |

| Eyes |                        |         |
|------|------------------------|---------|
|      | Symptom                | Details |
|      | Blurred vision         |         |
|      | Double vision          |         |
|      | Eye pain               |         |
|      | Redness                |         |
|      | Dry eyes               |         |
|      | Excessive Tearing      |         |
|      | Light sensitivity      |         |
|      | Vision loss or changes |         |
|      | Other _____            |         |

| Ears, Nose, and Throat |                          |         |
|------------------------|--------------------------|---------|
|                        | Symptom                  | Details |
|                        | Earache                  |         |
|                        | Hearing loss or changes  |         |
|                        | Ringing in the ears      |         |
|                        | Nasal congestion         |         |
|                        | Runny nose               |         |
|                        | Sinus pressure or pain   |         |
|                        | Nosebleeds               |         |
|                        | Sore throat              |         |
|                        | Hoarseness               |         |
|                        | Difficulty swallowing    |         |
|                        | Dental or mouth problems |         |
|                        | Other _____              |         |

| Cardiovascular |                                     |         |
|----------------|-------------------------------------|---------|
|                | Symptom                             | Details |
|                | Chest pain or pressure              |         |
|                | Palpitations or irregular heartbeat |         |
|                | Shortness of breath with activity   |         |
|                | Shortness of breath when lying down |         |
|                | Swelling of the legs or feet        |         |
|                | Fainting or near fainting           |         |
|                | Leg pain with walking               |         |
|                | Other _____                         |         |

| Respiratory |                     |         |
|-------------|---------------------|---------|
|             | Symptom             | Details |
|             | Cough               |         |
|             | Coughing blood      |         |
|             | Wheezing            |         |
|             | Shortness of breath |         |
|             | Chest congestion    |         |
|             | Pain with breathing |         |
|             | Snoring             |         |
|             | Other _____         |         |

| Gastrointestinal |                          |         |
|------------------|--------------------------|---------|
|                  | Symptom                  | Details |
|                  | Nausea                   |         |
|                  | Vomiting                 |         |
|                  | Heartburn or acid reflux |         |

| Gastrointestinal |                        |  |
|------------------|------------------------|--|
|                  | Abdominal pain         |  |
|                  | Bloating               |  |
|                  | Diarrhea               |  |
|                  | Constipation           |  |
|                  | Blood in stool         |  |
|                  | Black or tarry stool   |  |
|                  | Change in bowel habits |  |
|                  | Loss of appetite       |  |
|                  | Other _____            |  |

| Genitourinary <sup>1</sup> |                                |         |
|----------------------------|--------------------------------|---------|
|                            | Symptom                        | Details |
|                            | Pain or burning with urination |         |
|                            | Increased urinary frequency    |         |
|                            | Urgent need to urinate         |         |
|                            | Blood in urine                 |         |
|                            | Incontinence                   |         |
|                            | Increased thirst or urination  |         |
|                            | Pelvic pain                    |         |
|                            | Other _____                    |         |

| Musculoskeletal |                |         |
|-----------------|----------------|---------|
|                 | Symptom        | Details |
|                 | Joint pain     |         |
|                 | Joint swelling |         |

<sup>1</sup> The genitourinary system (also called the urogenital system) combines the organs of the urinary system and the reproductive system because they sit close together, share a common embryological origin, and overlap in physical pathways

| Musculoskeletal |                  |  |
|-----------------|------------------|--|
|                 | Muscle pain      |  |
|                 | Muscle cramps    |  |
|                 | Back pain        |  |
|                 | Neck pain        |  |
|                 | Stiffness        |  |
|                 | Limited movement |  |
|                 | Recent injury    |  |
|                 | Other _____      |  |

| Skin |                      |         |
|------|----------------------|---------|
|      | Symptom              | Details |
|      | Rash                 |         |
|      | Itching              |         |
|      | Hives                |         |
|      | New or changing mole |         |
|      | Skin discoloration   |         |
|      | Excessive dryness    |         |
|      | Easy bruising        |         |
|      | Hair loss            |         |
|      | Non-healing wound    |         |
|      | Other _____          |         |

| Neurological |                      |         |
|--------------|----------------------|---------|
|              | Symptom              | Details |
|              | Headache             |         |
|              | Dizziness or vertigo |         |

| Neurological |                                  |  |
|--------------|----------------------------------|--|
|              | Numbness or tingling             |  |
|              | Weakness in arm or leg           |  |
|              | Tremor                           |  |
|              | Seizure                          |  |
|              | Memory changes                   |  |
|              | Difficulty concentrating         |  |
|              | Balance or coordination problems |  |
|              | Speech changes                   |  |
|              | Loss of consciousness            |  |
|              | Other _____                      |  |

| Mental Health |                                         |         |
|---------------|-----------------------------------------|---------|
|               | Symptom                                 | Details |
|               | Anxiety                                 |         |
|               | Depression or low mood                  |         |
|               | Irritability                            |         |
|               | Mood changes                            |         |
|               | Panic attacks                           |         |
|               | Excessive worry                         |         |
|               | Hallucinations                          |         |
|               | Difficulty concentrating                |         |
|               | Thoughts of self-harm or suicide        |         |
|               | Difficulty coping with daily activities |         |
|               | Other _____                             |         |

| Endocrine |                               |         |
|-----------|-------------------------------|---------|
|           | Symptom                       | Details |
|           | Increased sensitivity to heat |         |
|           | Increased sensitivity to cold |         |
|           | Excessive sweating            |         |
|           | Increased thirst              |         |
|           | Increased urination           |         |
|           | Changes in appetite           |         |
|           | Changes in menstrual cycle    |         |
|           | Intolerance to heat or cold   |         |
|           | Other _____                   |         |

| Hematologic / Lymphatic |                                |         |
|-------------------------|--------------------------------|---------|
|                         | Symptom                        | Details |
|                         | Easy bruising                  |         |
|                         | Excessive bleeding             |         |
|                         | Anemia                         |         |
|                         | Enlarged or tender lymph nodes |         |
|                         | History of blood clots         |         |
|                         | Other _____                    |         |

| Allergy / Immune |                      |         |
|------------------|----------------------|---------|
|                  | Symptom              | Details |
|                  | Seasonal allergies   |         |
|                  | Food allergies       |         |
|                  | Medication allergies |         |
|                  | Frequent infections  |         |

| Allergy / Immune |                                                  |  |
|------------------|--------------------------------------------------|--|
|                  | Swelling of the face, lips, or throat            |  |
|                  | Difficulty breathing due to an allergic reaction |  |
|                  | Other _____                                      |  |

| Reproductive / Sexual Health |                             |         |
|------------------------------|-----------------------------|---------|
|                              | Symptom                     | Details |
|                              | Menstrual changes           |         |
|                              | Abnormal vaginal bleeding   |         |
|                              | Vaginal discharge           |         |
|                              | Pelvic pain                 |         |
|                              | Erectile difficulties       |         |
|                              | Testicular pain or swelling |         |
|                              | Sexual pain                 |         |
|                              | Concerns about fertility    |         |
|                              | Other _____                 |         |

| Current Medications or Supplements |  |
|------------------------------------|--|
| 1                                  |  |
| 2                                  |  |
| 3                                  |  |
| 4                                  |  |
| 5                                  |  |
| 6                                  |  |
| 7                                  |  |
| 8                                  |  |
| 9                                  |  |
| 10                                 |  |

| Medications or Environmental Allergies |  |
|----------------------------------------|--|
| 1                                      |  |
| 2                                      |  |
| 3                                      |  |
| 4                                      |  |
| 5                                      |  |

| Additional Symptoms or Concerns |  |
|---------------------------------|--|
| 1                               |  |
| 2                               |  |
| 3                               |  |
| 4                               |  |
| 5                               |  |

This checklist is a tool for discussion with a healthcare professional and is not a substitute for medical evaluation. NFXF disclaimer.

Using the information captured above, complete the following worksheet to help organize your top priorities. Consider bringing this worksheet to your appointment with your provider to help them understand the Fragile X premutation and communicate your top concerns.

# My General Healthcare Visit Worksheet

|                                                         |  |
|---------------------------------------------------------|--|
| <b>Patient Name:</b>                                    |  |
| <b>Date of Birth</b>                                    |  |
| <b>Primary Concern / Reason for Visit</b>               |  |
| <b>Fragile X CGG repeat number / status<sup>2</sup></b> |  |

|                                           |         |
|-------------------------------------------|---------|
| <b>Why am I seeing my provider today?</b> | Answer: |
|                                           |         |

|                                                              |  |
|--------------------------------------------------------------|--|
| <b>What are the most important things I want to discuss?</b> |  |
| 1                                                            |  |
| 2                                                            |  |
| 3                                                            |  |
| 4                                                            |  |
| 5                                                            |  |

|                                                  |         |
|--------------------------------------------------|---------|
| <b>What changed or prompted me to seek care?</b> | Answer: |
|                                                  |         |

<sup>2</sup> Fragile X status can be a helpful risk identifier. The individual sharing this information with you wants you to understand this in addition to the symptoms they are experiencing. This individual also understands that they may have symptoms or conditions that are unique to them and are not associated with their Fragile X status.

| What would I like help with today? |                                                               |
|------------------------------------|---------------------------------------------------------------|
|                                    | Understanding what may be causing my symptoms                 |
|                                    | Evaluating a new or changing symptom                          |
|                                    | Discussing treatment or management options                    |
|                                    | Discussing whether additional testing or evaluation is needed |
|                                    | Discussing whether I should see a specialist                  |
|                                    | Other:                                                        |

## My Top Concerns

You may have several symptoms identified as concerns. Narrow your list to the three concerns that are most important for you to discuss with your provider at this visit. You may only have one or two concerns.

Think about:

- Which concern is affecting me the most?
- Which concern has changed recently?
- Which concern do I most want help understanding?
- Which concern do I want my provider to evaluate or address?

|                                                         |                                                                                                                                               |
|---------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Concern #1:</b>                                      |                                                                                                                                               |
| <b>When did I first notice this?</b>                    | Answer:                                                                                                                                       |
| <b>How often does it happen?</b>                        | <input type="checkbox"/> Occasionally <input type="checkbox"/> Weekly <input type="checkbox"/> Daily <input type="checkbox"/> Other:          |
| <b>Has it changed over time?</b>                        | <input type="checkbox"/> Improving <input type="checkbox"/> Worsening <input type="checkbox"/> About the same <input type="checkbox"/> Unsure |
| <b>What does this concern look or feel like for me?</b> | Answer:                                                                                                                                       |
| <b>How does it affect my daily life or activities?</b>  | Answer:                                                                                                                                       |

|                                                         |                                                                                                                                               |
|---------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Concern #2:</b>                                      |                                                                                                                                               |
| <b>When did I first notice this?</b>                    | Answer:                                                                                                                                       |
| <b>How often does it happen?</b>                        | <input type="checkbox"/> Occasionally <input type="checkbox"/> Weekly <input type="checkbox"/> Daily <input type="checkbox"/> Other:          |
| <b>Has it changed over time?</b>                        | <input type="checkbox"/> Improving <input type="checkbox"/> Worsening <input type="checkbox"/> About the same <input type="checkbox"/> Unsure |
| <b>What does this concern look or feel like for me?</b> | Answer:                                                                                                                                       |
| <b>How does it affect my daily life or activities?</b>  | Answer:                                                                                                                                       |

|                                                         |                                                                                                                                               |
|---------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Concern #3:</b>                                      |                                                                                                                                               |
| <b>When did I first notice this?</b>                    | Answer:                                                                                                                                       |
| <b>How often does it happen?</b>                        | <input type="checkbox"/> Occasionally <input type="checkbox"/> Weekly <input type="checkbox"/> Daily <input type="checkbox"/> Other:          |
| <b>Has it changed over time?</b>                        | <input type="checkbox"/> Improving <input type="checkbox"/> Worsening <input type="checkbox"/> About the same <input type="checkbox"/> Unsure |
| <b>What does this concern look or feel like for me?</b> | Answer:                                                                                                                                       |
| <b>How does it affect my daily life or activities?</b>  | Answer:                                                                                                                                       |

# My Current Health Information

Current medications, over-the-counter products, and supplements. Include everything you currently take or use, even if it does not require a prescription.

This may include vitamins, minerals, herbal products, natural or “wellness” products, CBD products, or other products you take or use for your health.

| Medication or product | How much / how often? | Why do I take or use it? |
|-----------------------|-----------------------|--------------------------|
|                       |                       |                          |
|                       |                       |                          |
|                       |                       |                          |
|                       |                       |                          |
|                       |                       |                          |
|                       |                       |                          |
|                       |                       |                          |
|                       |                       |                          |
|                       |                       |                          |

**Tip:** If you are unsure whether something is a medication or supplement, include it anyway and bring the product or a photo of the label to your appointment

|                                               |         |
|-----------------------------------------------|---------|
| Medication, environmental, or other allergies | Answer: |
| Other diagnosed health conditions             | Answer: |
| Other diagnosed health conditions             | Answer: |

# Questions for My Provider

|                                                           |         |
|-----------------------------------------------------------|---------|
| <b>The most important question I want answered today:</b> | Answer: |
|-----------------------------------------------------------|---------|

|                                       |  |
|---------------------------------------|--|
| <b>Other questions I want to ask:</b> |  |
| 1                                     |  |
| 2                                     |  |
| 3                                     |  |
| 4                                     |  |
| 5                                     |  |

|                                                                              |         |
|------------------------------------------------------------------------------|---------|
| <b>Is there anything I want to make sure I discuss before my visit ends?</b> | Answer: |
|------------------------------------------------------------------------------|---------|

# Before I Leave My Appointment

|                                                         |         |
|---------------------------------------------------------|---------|
| <b>Additional or recommended testing or evaluation:</b> | Answer: |
| <b>Treatment or management recommendations:</b>         | Answer: |
| <b>Referrals or specialist evaluation</b>               | Answer: |
| <b>Follow-up recommendations:</b>                       | Answer: |
| <b>Other next steps:</b>                                | Answer: |