



FOR PROFESSIONALS

MEDICAL CODING FOR FRAGILE X/*FMR1*-ASSOCIATED CONDITIONS

Below are possible procedural, service, and diagnostic codes that might be used in your practice to order Fragile X testing and/or provide a formal diagnosis. This is for information purposes only. Always verify these codes with the specific testing lab or a billing specialist as there may be updates or variations in coding depending on the laboratory, insurance requirements, or specific panel requested. Additionally, CPT codes can evolve to reflect the use of new technologies or genetic panels.

POSSIBLE CPT CODES:

- **Carrier Screening for Fragile X:**
 - **CPT 81243:** Analysis of *FMR1* gene CGG repeat ranges.
- **Diagnostic Testing for Fragile X:**
 - **CPT 81244:** Additional analysis if applicable for methylation and AGG interruptions.
 - **CPT 81420:** Analysis for *FMR1* gene via CVS, Amniocentesis, or Cord Blood.
- **Next-Generation Sequencing (NGS):**
 - **CPT 81415:** Whole Exome Sequencing.
 - **CPT 81416:** Targeted NGS Gene Panels.
 - **CPT 81479:** Unlisted NGS Testing.
- **Ataxia Panels (may include *FMR1* Gene):**
 - **CPT 81445:** Ataxia Panel.
 - **CPT 81446:** Ataxia Panel (alternative).

POSSIBLE DIAGNOSTIC CODES (ICD-10):

- **Z15.8 - Genetic Counseling**
- **Q99.2 - Fragile X Syndrome/Fragile X Premutation**
- **Fragile X-associated primary ovarian insufficiency (FXPOI)**
 - **E28.0** – *Primary ovarian insufficiency*, which can be used for FXPOI.
 - **E28.1** – *Ovarian failure, unspecified*, this may be used if the exact cause of ovarian failure isn't known.
 - **N95.0** – *Menopausal and female climacteric states*, this may be used for patients who are experiencing symptoms of menopause for the typical age.
- **Fragile X-associated tremor/ataxia syndrome (FXTAS) :**
 - **G31.2** – *Other degenerative diseases of the nervous system, not elsewhere classified*, which can be used for cases of FXTAS.
 - **G11.9** – *Hereditary ataxia, unspecified*, this can be used if the specific hereditary cause (like FXTAS) is not definitively determined but the clinical presentation suggests ataxia related to a genetic disorder.
 - **G25.0** – *Essential tremor*, sometimes used for tremors associated with FXTAS.

Want to know more?

• fragilex.org
• contact@fragilex.org

