

Medical Coding for Fragile X/ FMR1-Associated Conditions

For Professionals

Here we list possible procedural, service, and diagnostic codes that might be used in your practice to order Fragile X testing 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	CPT Code	Description
Carrier Screening for Fragile X	81243	Analysis of <i>FMR1</i> gene CGG repeat ranges
Diagnostic Testing for Fragile X	81244 81420	Additional analysis if applicable for methylation and AGG interruptions. Analysis for <i>FMR1</i> gene via CVS, Amniocentesis, or Cord Blood.
Next-Generation Sequencing (NGS)	81415 81416 81479	Whole Exome Sequencing. Targeted NGS Gene Panels. Unlisted NGS Testing.
Ataxia Panels (may include <i>FMR1</i> Gene)	81445 81446	Ataxia Panel Ataxia Panel (alternative)
Possible ICD-10 Diagnostic Codes	ICD-10 Code	Description
Genetic Counseling	Z15.8	
Fragile X Syndrome/Fragile X Premutation	Q99.2	
Fragile X-associated primary ovarian insufficiency (FXPOI)	E28 E28.1 N95.0	<i>Primary ovarian insufficiency</i> , which can be used for FXPOI. <i>Ovarian failure, unspecified</i> , this may be used if the exact cause of ovarian failure isn't known. <i>Menopausal and female climacteric states</i> , 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 G11.9 G25.0	<i>Other degenerative diseases of the nervous system, not elsewhere classified</i> , which can be used for cases of FXTAS. <i>Hereditary ataxia, unspecified</i> , 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. <i>Essential tremor</i> , sometimes used for tremors associated with FXTAS.