Guidelines for Medical Necessity Determination for Fragile X Carrier Screening

These guidelines for medical necessity determination (guidelines) identify the clinical information that MassHealth needs to determine medical necessity for screening for maternal Fragile X carrier status.

Fragile X syndrome is transmitted as an X-linked disorder. However, the molecular genetics of the syndrome are complex. These guidelines are based on generally accepted standards of practice, review of the medical literature, and federal and state policies and laws applicable to Medicaid programs.

Providers should consult MassHealth regulations at 130 CMR 401.000: *Independent Clinical Laboratory Services*, 130 CMR 433.000: *Physician Services*, and 130 CMR 450.000: *Administrative and Billing Regulations*; Subchapter 6 of the Independent Clinical Laboratory Manual; and Subchapter 6 of the Physician Manual for information about coverage, limitations, service conditions, and other prior authorization (PA) requirements.

Providers serving members enrolled in a MassHealth-contracted accountable care partnership plan (ACPP), managed care organization (MCO), One Care Organization, Senior Care Organization (SCO), or Program of All-inclusive Care for the Elderly (PACE) should refer to the ACPP's, MCO's, One Care Organization's, SCO's, or PACE's medical policies, respectively, for covered services.

MassHealth requires PA for Fragile X carrier screening. MassHealth reviews requests for PA on the basis of medical necessity. If MassHealth approves the request, payment is still subject to all general conditions of MassHealth, including member eligibility, other insurance, and program restrictions.

SECTION I. GENERAL INFORMATION

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Carrier screening is a term used to describe genetic testing that is performed on an individual who does not have any overt phenotype for a genetic disorder but may have one variant allele within a gene(s) associated with a diagnosis. Carrier screening for a particular condition generally should be performed only once in a person's lifetime, and the results should be documented in the patient's health record. Because of the rapid evolution of genetic testing, additional mutations may be included in newer screening panels. The decision to rescreen a patient should be undertaken only with the guidance of a genetics professional who can best assess the incremental benefit of repeat testing for additional mutations.

Fragile X syndrome is the most common inherited form of intellectual disability. The syndrome occurs in approximately 1 in 3,600 males and 1 in 4,000–6,000 females, from a variety of ethnic backgrounds. Intellectual disability or impairment ranges from borderline, including learning disabilities, to severe, presenting with cognitive and behavioral disabilities, including autism with intellectual disability; attention deficit-hyperactivity disorder; or both. Most affected males have significant intellectual disability. Fragile X syndrome is a commonly known cause of autism or autism spectrum disorder behaviors with intellectual disability, with the diagnosis of Fragile X occurring in approximately 25% of individuals affected by these conditions.

The American College of Obstetricians & Gynecologists recommends Fragile X premutation carrier screening for pregnant people with a family history of Fragile X-related disorders or intellectual disability suggestive of Fragile X syndrome and who are considering pregnancy or are currently pregnant. If a person has unexplained ovarian insufficiency or failure or an elevated follicle-stimulating hormone level before the age of 40, Fragile X carrier screening is recommended to determine whether they have an FMR1 premutation. All identified individuals with intermediate results and carriers of a Fragile X premutation or full mutation should be provided follow-up genetic counseling to discuss the risk to their offspring of inheriting an expanded full-mutation Fragile X allele and to discuss Fragile X-associated disorders (premature ovarian insufficiency and Fragile X tremor/ataxia syndrome).

Prenatal diagnostic testing for Fragile X syndrome should be offered to known carriers of the Fragile X premutation or full mutation. DNA-based molecular analysis (e.g., Southern blot analysis and polymerase chain reaction) is the preferred method of diagnosis of Fragile X syndrome and of determining FMR1 triplet repeat number (e.g., premutations).

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SECTION II. CLINICAL GUIDELINES

A. CLINICAL COVERAGE

MassHealth bases its determination of medical necessity for maternal Fragile X carrier screening on clinical data including, but not limited to, indicators that would affect the relative risks and benefits of the test. These criteria, which are adapted from ACOG Committee Opinion Number 691, March 2017, include, but are not limited to, the test being used as a screening for carrier status of a person who is pregnant or planning to become pregnant and who is at an increased risk of being a carrier of the Fragile X premutation as evidenced by meeting any of the following indications:

1. A personal or family history of Fragile X-related disorders or intellectual disability suggestive of Fragile X syndrome; or
2. Unexplained ovarian insufficiency or failure or an elevated follicle-stimulating hormone level before age 40.

B. NONCOVERAGE

MassHealth does not consider screening for Fragile X carrier status to be medically necessary under certain circumstances. Examples of such circumstances include, but are not limited to, the following.

1. Fragile X carrier status screening in people who are not at increased risk of carrying the Fragile X premutation
2. Fragile X carrier status screening in egg donor pregnancies
3. Fragile X carrier status screening performed at laboratories that are not contracted with MassHealth

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SECTION III. SUBMITTING CLINICAL DOCUMENTATION

Requests for PA for Fragile X carrier screening must be accompanied by clinical documentation that supports the medical necessity for this procedure.

A. Documentation of medical necessity must include all of the following.

   1. A copy of the completed test requisition, which has been signed and dated by the ordering physician;
   2. Primary diagnosis name and ICD-10 code pertinent to clinical scenario; and
   3. Documentation of the clinical history and/or results of prior testing that meet clinical coverage criteria (see Section II.A "Clinical Coverage").

B. Clinical information must be submitted by the MassHealth-enrolled clinical laboratory performing the genetic testing.

Providers are strongly encouraged to submit requests electronically. Providers must submit the request for PA and all supporting documentation using the Provider Online Service Center (POSC), or by completing a MassHealth *Prior Authorization Request* (PA-1 form), found at www.mass.gov/masshealth, and attaching all supporting documentation. Questions about POSC access should be directed to the MassHealth Customer Service Center at (800) 841-2900, TDD/TTY: 711.

SELECT REFERENCES

1. ACOG Committee on Genetics Committee Opinion No. 691: Carrier screening for genetic conditions. *Obstet Gynecol*. 2017;129:e41-55.

These Guidelines are based on review of the medical literature and current practice in Fragile X carrier screening. MassHealth reserves the right to review and update the contents of these Guidelines and cited references as new clinical evidence and medical technology emerge.

This document was prepared for medical professionals to assist them in submitting documentation supporting the medical necessity of the proposed treatment, products, or services. Some language used in this communication may be unfamiliar to other readers; in this case, such readers should contact their health care provider for guidance or explanation.

Policy Effective Date: June 14, 2023

Approved by: 

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