Maryland
DEPARTMENT OF HEALTH

Genetic Carrier Screening - Testing Options & Clinical Criteria

Carrier screening, whether targeted or expanded, allows individuals to consider their range of reproductive options. Ultimately, the goal of genetic screening is to provide individuals with meaningful information that they can use to guide pregnancy planning based on their personal values. Ethnic-specific, panethnic, and expanded carrier screening are acceptable strategies for pre pregnancy and prenatal carrier screening. Because all of these are acceptable strategies, each obstetrician–gynecologist or other health care provider or practice should establish a standard approach that is consistently offered to and discussed with each patient, ideally before pregnancy.[1,2]

- 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.
- Prenatal carrier screening does not replace newborn screening, nor does newborn screening replace the potential value of prenatal carrier screening.
- The cost of carrier screening for an individual condition may be higher than the cost of testing through commercially available expanded carrier screening panels. When selecting a carrier screening approach, the cost of each option to the patient and the health care system should be considered.

Maryland Medicaid covers the following options for Carrier Screening.

I. Basic Carrier Screening
II. Targeted Screening
    a. Risk Based Targeted Screening
    b. Ethnic- Specific Screening
III. Expanded Carrier Screening

I. <u>Basic Carrier Screening Coverage</u>

All patients considering pregnancy, or who are already pregnant, regardless of screening strategy and ethnicity, should be offered the following carrier screening tests:

- Cystic Fibrosis
- Spinal Muscular Atrophy
- Complete Blood Count and screening for Thalassemias and Hemoglobinopathies (Sickle Cell Disease).

II. <u>Targeted Carrier Screening</u>

A. Risk Based Targeted Carrier Screening

This testing option would allow for specific risk based screening and can be utilized in addition to the Basic Carrier Screening Coverage.

i. Fragile X Premutation Carrier Screening. *This is recommended for women with a family history of fragile X-related disorders or intellectual disability suggestive of fragile X syndrome, or patient with a personal history of ovarian insufficiency. NO - PRE AUTHORIZATION REQUIRED*

ii. Additional Risk Based testing will be considered when the member meets one or more of the following conditions:

ii.1. Consanguinity - Couples with consanguinity should be offered genetic counseling to discuss the increased risk of recessive conditions being expressed in their offspring and the limitations and benefits of carrier screening.

ii.2. Maternal Fetal Medicine (MFM) physician or certified genetic counselor recommendations for specific carrier screen testing.

ii.3. Previous reproductive or family history. Individuals with a family history of a genetic disorder may benefit from the identification of the specific familial mutation or mutations. rather than carrier screening.

ii.3.1. One or both individuals have a first- or second-degree relative who is affected OR

ii.3.2. One individual is known to be a carrier OR

ii.3.3. One or both individuals are members of a population known to have a carrier rate that exceeds a threshold considered appropriate for testing for a particular condition.

B. Ethnicity- Specific Carrier Screening

This option provides targeted ethnicity specific screening and can be utilized in addition to the Basic Carrier Screening Coverage.

When patients have a known specific ethnicity, screening based on obtained family history.* Examples: *Ashkenazi Descent: Tay Sachs, Canavan Disease, Familial Dystonia, Bloom Syndrome, Familial Hyperinsulinism Fanconi Anemia, Gaucher Disease, Joubert Syndrome, Maple Syrup Urine Disease, Mucolipidosis Type !V, Niemann Pick Disease & Usher Syndrome.* [1&2]

III. Expanded Carrier Screening

Expanded Carrier Screening can be offered to all individuals regardless of ethnicity rather than traditional ethnicity-specific screening. Expanded carrier screening should cover the tests included in basic carrier screening.

Given the multitude of conditions that can be included in expanded carrier screening panels, the disorders selected for inclusion **must meet** the following ACOG consensus-determined criteria:

- have a carrier frequency of 1 in 100 or greater, have a well-defined phenotype, **and**
- have a detrimental effect on quality of life, **or**
- cause cognitive or physical impairment, **or**
- require surgical or medical intervention, **and**
- have an onset early in life. (Carrier screening panels should not include conditions primarily associated with a disease of adult onset)

Additionally, screened conditions should be able to be diagnosed prenatally and may afford opportunities for antenatal intervention to improve perinatal outcomes, changes to delivery management to optimize newborn and infant outcomes, and education of the parents about special care needs after birth.

IV. Genetic counseling

Genetic Counseling should be considered for those patients with a positive screen, when recommended by the patient's treating provider.

81220 81329 83021	**Option 1: Basic Coverage** Cystic Fibrosis Spinal Muscular Atrophy (SMA) Sickle Cell
81243 Various Codes 81412	**Option 2: Targeted Carrier Screening** **Fragile X** **Risk Based Targeted Screening** **Ethnic Specific Coverage**
81443	**Option 3: Panethnic Approach - Expanded Carrier Screening** Genetic test severe inherited cond. Genomic sequence analysis panel for severe inherited conditions with sequencing "genetic testing for severe inherited conditions, genomic sequence analysis panel of at least 15 genes."

[1]**American College of Obstetrics & Gynecology (ACOG) - Committee Opinion No. 690,** ***Carrier Screening in the age of genomic medicine., and*** **ACOG - Committee Opinion No. 691,** ***Carrier Screening for Genetic Conditions*****. Reaffirmed 2025**

[2] **American College of Obstetrics & Gynecology (ACOG) - Committee Opinion No. 691,** ***Carrier Screening for Genetic Conditions.*** **Reaffirmed 2025**

Prior authorization of benefits is not the practice of medicine nor the substitute for the independent medical judgment of a treating medical provider. The materials provided are a component used to assist in making coverage decisions and administering benefits. Prior authorization does not constitute a contract or guarantee regarding member eligibility or payment. Prior authorization criteria are established based on a collaborative effort using input from the current medical literature and based on evidence available at the time.

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