# Genetic Carrier Screening Clinical Criteria

Policy Number: PA 32
Last Review Date:
Effective Date: 8/1/2026

**Policy**

Maryland Care, Inc., dba Maryland Physicians Care (MPC) utilizes this clinical policy when specific criteria are not available for the requested Genetic Carrier Screening

- 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 MPC covers the following options for Genetic Carrier Screening:

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

MPC considers Genetic Carrier Screening medically necessary when ALL the following criteria are met:

**I. Basic Carrier Screening coverage:**

**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. Targeted Carrier Screening**

**A. Risk Based Targeted Carrier Screening coverage:**

This can be utilized in addition to the basic carrier screening

- 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*

- Additional Risk Based testing will be <u>considered</u> when the member meets **one or more** of the following conditions:
    - 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.
    - Maternal Fetal Medicine (MFM) physician or certified genetic counselor recommendations for specific carrier screen testing.
    - 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.
        - One or both individuals have a first- or second-degree relative who is affected OR
        - One individual is known to be a carrier OR
        - 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 can be utilized in addition to the basic carrier screening.

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]

**C. 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 **<u>must meet</u>** the following ACOG consensus-determined criteria:

- have a carrier frequency of 1 in 100 or greater, have a well-defined phenotype, **<u>and</u>**
- have a detrimental effect on quality of life, **<u>or</u>**
- cause cognitive or physical impairment, **<u>or</u>**
- require surgical or medical intervention, **and**

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- 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.

**Limitations**
**None**

**Background**
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]

**Codes/Devices/Services**

| Service | Description |
|---|---|
| 81220, 81329, 83021 | Option 1: Basic Coverage: Cystic Fibrosis, Spinal Muscular Atrophy (SMA), Sickle Cell |
| 81243, 81412, various codes | Option 2: Targeted Carrier Screening: Fragile X, Risk Based Targeted Screening, Ethnic Specific Coverage |
| 81443 | Option 3: Panethnic Approach- Expanded Carrier Screening genetic testing for severe inherited conditions |

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## References

[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**

## Revision Log

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| New Policy Created | 08/01/2026 |
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