Carrier Screening Panels, Including Targeted, Pan-Ethnic, Universal, and Expanded

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Carrier screening panels, including targeted, pan-ethnic, universal, and expanded are addressed by this guideline.

Procedures addressed

The inclusion of any procedure code in this table does not imply that the code is under management or requires prior authorization. Refer to the specific Health Plan's procedure code list for management requirements.

Procedures addressed by this guideline	Procedure codes
Carrier screening panel	81479
Genesys Carrier Panel	0400U
Genetic testing for severe inherited conditions (eg, cystic fibrosis, Ashkenazi Jewish-associated disorders, genomic sequence analysis panel, must include sequencing of at least 15 genes (eg, ACADM, ARSA, ASPA, ATP7B, BCKDHA, BCKDHB, BLM, CFTR, DHCR7, FANCC, G6PC, GAA, GALT, GBA, GBE1, HBB, HEXA, IKBKAP, MCOLN1, PAH)	81443
UNITY Carrier Screen	0449U

Criteria

Introduction

Requests for carrier screening panels are reviewed using the following criteria. For the purposes of this guideline, a test is considered a reimbursable carrier screening panel when it includes testing for at least CF, SMA, and hemoglobinopathies. For information

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on carrier screening for disorders associated with Ashkenazi Jewish ancestry (billed with 81412), please see the guideline *Ashkenazi Jewish Carrier Screening*, as that testing in not addressed here. Please see EviCore test-specific guidelines or clinical use guidelines for information on testing individual genes or conditions.

Carrier Screening Panels

Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND

At a minimum, the panel must assess carrier status for the following conditions:

- Cystic fibrosis (CF)
- Hemoglobinopathies (beta thalassemia, alpha thalassemia, sickle cell disease)
- Spinal muscular atrophy (SMA)
- Any condition for which the individual is at elevated risk (e.g. individual has family history of a condition, individual's partner is a carrier of a condition, or individual's ethnicity/country of origin increases their risk of being a carrier of a condition), AND

The panel must employ the recommended methodology to maximize the detection of carriers for all conditions in the panel (e.g., dosage analysis for SMA), AND

The individual must not have had previous testing of any genes on the panel (exceptions may be made on a case-by-case basis if CF and/or SMA were previously performed individually), AND

The individual must be of reproductive age and have potential and intention to reproduce, AND

Rendering laboratory is a qualified provider of service per the Health Plan policy.

Billing and Reimbursement

This section outlines the billing requirements for tests addressed in this guideline. These requirements will be enforced during the case review process whenever appropriate. Examples of requirements may include specific coding scenarios, limits on allowable test combinations or frequency and/or information that must be provided on a claim for automated processing. Any claims submitted without the necessary information to allow for automated processing (e.g. ICD code, place of service, etc.) will not be reimbursable as billed. Any claim may require submission of medical records for post service review.

Carrier Screening Panels

Carrier screening panels must be billed with a single procedure code (e.g., 81479, or appropriate PLA code)* to represent all genes being analyzed via all methodologies necessary to determine carrier status (e.g. sequencing, deletion/duplication, dosage).

For the purposes of this guideline, a test is considered a reimbursable carrier screening panel when it includes testing for at least CF, SMA, and hemoglobinopathies.

- If performing a panel that has been assigned a PLA code, the PLA code should be billed.
- Panels without a PLA code should be billed with 81479.

Any other procedure codes (including non-specific molecular codes) that may represent carrier screening will not be reimbursable if billed on the same date of service as a carrier screening panel code.

If individual codes representing component genes of a carrier screening panel are submitted for medical necessity review, they will be redirected to a carrier screening panel code.

If individual codes representing component genes of a carrier screening panel are submitted on a claim, this is incorrect billing of the service and will not be reimbursable as billed.

Carrier screening panel codes will not be reimbursable if any single gene components of the panel have been performed and reimbursed previously. Exceptions on a case-by-case basis for CF and SMA may be made, but CF and SMA performed separately will not be reimbursed during the same pregnancy.

Exceptions for panels that do not include the required genes may be considered on a case by case basis.

Carrier screening panels are reimbursable once per lifetime.

- If an Ashkenazi Jewish carrier screening panel was previously billed, an additional carrier screening panel will not be reimbursable.
- If a non-Ashkenazi Jewish carrier screening panel was previously billed, subsequent carrier screening of any type will not be reimbursable (e.g. individual genes, Ashkenazi Jewish carrier screening panels).

CPT 81443 is not reimbursable.

Note:

*The panel code(s) listed here may not be all-inclusive. For further discussion of what is considered an appropriate panel code, please refer to the guideline *Laboratory Billing and Reimbursement*.

For general coding requirements, please refer to the guideline *Laboratory Billing and Reimbursement.*

What are carrier screening panels?

Carrier screening panels, Including targeted, pan-ethnic, universal, and expanded carrier screening, are designed to identify carrier status or predict risk for multiple genetic diseases in a single test. It is typically offered to individuals planning a pregnancy or currently pregnant.

Prevalence

The genetic diseases that are tested for range in severity from lethal in infancy to so mild an affected individual may never develop symptoms. Some conditions are quite common, especially in certain ethnic groups, while others are rare.

It is generally believed that all people carry several recessive gene mutations. An estimated 1 in 580 births has an autosomal recessive condition and 1 in 2000 has an X-linked condition.[1]

Inheritance

Carrier screening panels may include autosomal recessive and X-linked conditions.

Autosomal recessive inheritance

In autosomal recessive inheritance, individuals have 2 copies of the gene and an individual typically inherits a gene mutation from both parents. Usually only siblings are at risk for also being affected. Males and females are equally affected. Individuals who inherit only one mutation are called carriers. Carriers do not typically show symptoms of the disease, but have a 50% chance, with each pregnancy, of passing on the mutation to their children. If both parents are carriers of a mutation, the risk for each pregnancy to be affected is 1 in 4, or 25%.

X-Linked Inheritance

In X-linked inheritance, the mutation is carried on the X chromosome. Females have two X chromosomes, and males have one. Males typically have more severe symptoms than females. A female with a mutation has a 50% chance to pass that mutation to her children. A male with a mutation cannot pass the mutation to any sons, but will pass it to all daughters. A process called X-inactivation in females results in random inactivation of expression of one X-chromosome in each cell of the body. For females with one mutation, the percentage and distribution of cells with expression of the X chromosome carrying the mutation can influence the degree of severity.

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Common uses

Carrier screening is most commonly done for reproductive planning, to identify couples at risk for having a child with a recessive inherited disorder. In most cases, couples who have a child with a recessive inherited disorder have no family history of that disorder or any other risk factors.

Carrier screening for a specific disorder may be indicated when there is a positive family history, when a reproductive partner is a carrier of or is affected with a recessive disorder, or when there is a known increased risk based on ethnicity or other factors.

Test information

Carrier screening panels determine carrier status for numerous genetic conditions simultaneously for the purposes of reproductive planning.

Carrier screening panels

Several carrier screening panels are available. Each test has a unique set of diseases included in novel and proprietary genetic testing platforms. The number of mutations tested varies considerably by condition, ranging from a single mutation for rare conditions to over 100 mutations for cystic fibrosis. Many panels consist of full gene sequencing. Complete testing information, including a list of all conditions screened and the technology used, can be found at a laboratory's website.

Guidelines and evidence

American College of Obstetrics and Gynecology

The American College of Obstetrics and Gynecology (ACOG, 2017; Reaffirmed 2023) published a committee opinion that stated the following regarding Carrier Screening Panels:[2]

- "Ethnic-specific, panethnic, and expanded carrier screening are acceptable strategies for prepregnancy and prenatal carrier screening. 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. After counseling, a patient may decline any or all carrier screening."

  "Given the multitude of conditions that can be included in expanded carrier screening panels, the disorders selected for inclusion should meet several of the following consensus-determined criteria: have a carrier frequency of 1 in 100 or greater, have a well-defined phenotype, have a detrimental effect on quality of life, cause cognitive or physical impairment, require surgical or medical intervention, or have an onset early

in life. 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."

- "Carrier screening panels should not include conditions primarily associated with a disease of adult onset."

ACOG released a practice advisory (2023) that served to update the practice bulletin on hemoglobinopathies in pregnancy that stated:[3]

- "ACOG recommends offering universal hemoglobinopathy testing to persons planning pregnancy or at the initial prenatal visit if no prior testing results are available for interpretation. This helps ensure that at-risk individuals receive counseling about genetic risks; learn their reproductive options, which include preimplantation genetic testing and prenatal diagnosis; and make informed decisions. Hemoglobinopathy testing may be performed using hemoglobin electrophoresis or molecular genetic testing (eg, expanded carrier screening that includes sickle cell disease [SCD] and other hemoglobinopathies)."

American College of Medical Genetics and Genomics

The American College of Medical Genetics and Genomics (ACMG, 2021) released an educational practice resource on carrier screening.[4] This consensus statement asserted that general population carrier screening should be ethnicity and family history agnostic. To accomplish this, screening all individuals in the prenatal/preconception period for autosomal recessive and X-linked conditions with a carrier frequency of >1/200 was suggested. ACMG generated a list of 113 genes meeting these criteria.

References

1. Thompson MW, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 5th ed. Philadelphia: Saunders; 1991.
2. ACOG Committee Opinion. Number 690, March 2017. Reaffirmed 2023. Carrier screening in the age of genomic medicine. *Obstet Gynecol*. 2017;129(3):595-596.
3. ACOG Practice Advisory. Hemoglobinopathies in Pregnancy. August 2022. Reaffirmed September 2023. Available at: **https://www.acog.org/clinical/clinical-guidance/practice-advisory/articles/2022/08/hemoglobinopathies-in-pregnancy**
4. Gregg AR, Aarabi M, Klugman S, et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2021;23(10):1793-1806. doi: 10.1038/s41436-021-01203-z