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# ADMINISTRATIVE GUIDELINE – 10.01.526

# Molecular Genetic Testing: Services Reviewed by Carelon Medical Benefits Management

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| Effective Date: | Aug. 1, 2024 |
| Last Revised: | July 8, 2024 |

REPLACES MEDICAL POLICIES:

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| 2.04.68 | Laboratory and Genetic Testing for Use of 5-Fluorouracil in Patients with Cancer |
| 4.01.21 | Noninvasive Prenatal Screening for Fetal Aneuploidies and Microdeletions using Cell-Free Fetal DNA |
| 12.04.13 | Genetic Testing for Alzheimer Disease |
| 12.04.28 | Genetic Testing for Predisposition to Inherited Hypertrophic Cardiomyopathy |
| 12.04.36 | Assays of Genetic Expression in Tumor Tissue as a Technique to Determine Prognosis in Patients with Breast Cancer |
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## Introduction

Carelon Medical Benefits Management conducts reviews for molecular genetic testing. Guidelines for genetic testing, including for pharmacogenomics, prenatal diagnosis, cardiac disease, cancer susceptibility, tumors and malignancies as well as single-gene genetic testing, and whole exome and whole genome sequencing will be used to conduct clinical reviews. The link to Carelon's Clinical Appropriateness Guidelines for Genetic Testing can be found below. To submit a request for services, providers will need to use the Carelon Medical Benefits Management provider portal, which is also provided below.

## Administrative Guideline

| Service | Link |
|---|---|
| **Molecular genetic testing** | **Carelon Clinical Appropriateness Guidelines for Genetic Testing encompass:**<br>• Carrier screening in the prenatal setting and preimplantation genetic testing<br>• Cell-free DNA testing for the management of cancer<br>• Chromosomal microarray analysis<br>• Genetic testing for inherited conditions<br>• Hereditary cancer testing<br>• Pharmacogenetic testing<br>• Poylgenic risk scores<br>• Prenatal testing using cell-free DNA<br>• Somatic tumor testing<br>• Whole exome sequencing and whole genome sequencing<br><br>**View** Carelon Medical Benefits Guidelines |
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## Related Information

N/A

## Rationale

Pre-test genetic counseling provides individuals seeking genetic testing the opportunity to make informed decisions about their genetic testing and subsequent medical management options. Genetic counseling combines expertise in obtaining and interpreting family history information, the ability to identify the most beneficial individual in a family to initiate testing, identification of most appropriate testing options, and proficiency in genetic variant interpretation to maximize the genetic testing experience for patients and their healthcare providers. Patients who receive genetic counseling report increased knowledge, understanding, and satisfaction regarding their genetic testing experience.[7-8] The genetic counseling informed consent process also educates and empowers patients to consider the psychological, financial, employment, disability, and insurance implications of genetic testing results.[4]

The advent of multi-gene panels and genome-scale sequencing have increased the complexity of the genetic testing landscape. Misuse of genetic testing increases the risk for adverse events and patient harm including missed opportunities for diagnosis and disease prevention.[9-11] Genetic information requires expert interpretation and ongoing re-evaluation to ensure the most accurate interpretation is used for informing medical management decision making. The multitude of genetic testing options as well as the complex information revealed by genetic testing can make choosing the most appropriate test and interpretation of results difficult for non-genetics healthcare providers.[12] Involvement of a clinical genetics provider has been shown to ensure the correct test is ordered, limits result misinterpretation and allows patients to make informed, evidence-based medical decisions with their healthcare providers.[13]

Genetic counseling not only improves patient outcomes but also reduces unnecessary healthcare spending. Pre-test genetic counseling has been shown to reduce inappropriate test ordering and prevent unnecessary medical procedures and interventions from inaccurate result interpretation.[14] While genetic testing is now available to providers in almost all clinical specialties, correct use and interpretation is necessary to prevent adverse outcomes. While genetic counseling may benefit any patient considering or undergoing genetic testing, tests that offer predictive information or have a higher chance of identifying uncertain variants often carry

stronger recommendations in the form of consensus guidelines and professional statements for genetic counseling by trained genetics professionals.

## Genetic Counseling for WES

There is consensus that involvement of trained genetics professionals in consulting with patients is essential prior to and after ordering such tests and can identify the appropriate patients for large multi-gene panels or WES.[15]

Obtaining informed consent and providing pre-test genetic counseling by a trained genetics professional is an essential component of WES. The American College of Medical Genetics (ACMG) published specific recommendations[6]:

- Pre-test counseling should be done by a medical geneticist or an affiliated genetic counselor and should include a formal consent process.
- Prior to initiating whole genome sequencing (WGS)/WES, participants should be counseled regarding the expected outcomes of testing, the likelihood and type of incidental results that could be generated, and what results will or will not be disclosed.
- As part of the pre-test counseling, a clear distinction should be made between clinical and research-based testing. In many cases, findings will include variants of unknown significance that might be the subject for research; in such instances a protocol approved by an institutional review board must be in place and appropriate prior informed consent obtained from the participant.

## Genetic Counseling for Hereditary Cardiac Conditions

Both the joint consortium of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines and the Heart Rhythm Society (AHA/ACC/HRS) as well as the ACMG have issued strong recommendations for genetic counseling for individuals undergoing evaluation for inherited cardiac disease.

In their Task Force publication from 2017[4], the AHA/ACC/HRS provided this recommendation:

> The decision to proceed with genetic testing requires discussion regarding the clinical use of genetic information to be obtained for both the proband and family members, as well as consideration of the important psychological, financial, employment, disability, and life

insurance implications of positive genotyping. Balancing privacy of health care information for the proband with the "right to know" for family members, and the ability to provide appropriate communication of information to all potentially affected family members can be challenging on many levels, including family dynamics, geographic proximity, and access to healthcare. For these reasons, genetic counseling generally occurs before proceeding with genetic testing, and, from a patient's perspective, is optimally provided by genetic counselors, if available, in collaboration with physicians. A combined approach of genetic counseling with medical guidance may appropriately balance the decision as to whether genetic testing would be beneficial on an individual basis.

In the recent joint statement put forth by the ACMG and Heart Failure Society[16], genetic counseling performed by a board-certified or board-eligible genetic specialist or specialized physician in the absence of a genetics professional is recommended as a key component of the evaluation of individuals with suspected familial cardiomyopathies with a level of evidence of A, their strongest recommendation. In addition, this recommendation includes a specific structure for genetic counseling that notes genetics professionals are specially trained to provide, including review of medical records essential for phenotyping, obtaining a pedigree, patient and family education, evaluating genetic testing options, obtaining consent for genetic testing, facilitating family communication, and ordering and interpreting genetic test results while addressing psychosocial issues.

## Genetic Counseling for Hereditary Cancer Conditions

Finally, the field of genetics that has included the broadest support for genetic counseling is hereditary cancer susceptibility. Many consensus organizations including the American Society of Clinical Oncology (ASCO)[17], the National Comprehensive Cancer Network (NCCN)[1,2], the American College of Obstetricians and Gynecologists (ACOG)[18] and the U.S. Preventive Services Task Force (USPSTF)[19] recommend genetic counseling as an integral part of the evaluation of individuals at risk for hereditary cancer susceptibility syndromes. Additionally, the Affordable Care Act [20] has established that counseling prior to mutation testing is an established essential health benefit appropriate for individuals with breast cancer.

Per the NCCN, cancer risk assessment and genetic counseling is highly recommended when genetic testing is offered (i.e., pre-test counseling) and after results are disclosed (i.e., post-test counseling), with assurance that the pre-test counseling includes collection of a comprehensive family history, evaluation of risk, full genetic differential review and education for the patient on the outcomes of testing, as well as full informed consent.

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## History

| Date | Comments |
|---|---|
| 10/10/18 | New Administrative Guideline, approved October 9, 2018. |
| 12/14/18 | Interim Review, approved December 13, 2018. Clarified genetic counseling requirement rationale. |
| 11/01/19 | Annual Review, approved October 4, 2019. References updated. Administrative guideline unchanged. |
| 09/01/20 | Annual Review, approved August 20, 2020. Administrative guideline unchanged. |
| 08/01/21 | Annual Review, approved July 22, 2021. References updated; administrative guideline remains unchanged. |
| 08/01/22 | Annual Review, approved July 25, 2022. Guideline reviewed; references updated. No content changes were made. |
| 03/01/23 | Annual Review, approved February 20, 2023. Policy updated to reflect AIM Specialty Health name change to Carelon Medical Benefits Management. URL updated from AIMspecialtyhealth.com to carelon.com with no content changes made to the policy. This name change is effective March 1, 2023. |
| 08/01/24 | Annual Review, approved July 8, 2024. Carelon clinical appropriateness guidelines for genetic testing have been updated with the following title changes: Carrier screening in the prenatal setting and preimplantation genetic testing, Cell-free DNA testing for the management of cancer, Chromosomal microarray analysis, Genetic testing for inherited conditions, Hereditary cancer testing, Poylgenic risk scores, Prenatal testing using cell-free DNA, and Somatic tumor testing. |

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**Scope**: Medical policies are systematically developed guidelines that serve as a resource for Company staff when determining coverage for specific medical procedures, drugs or devices. Coverage for medical services is subject to the limits and conditions of the member benefit plan. Members and their providers should consult the member benefit booklet or contact a customer service representative to determine whether there are any benefit limitations applicable to this service or supply. This medical policy does not apply to Medicare Advantage.