



Medica Central Coverage Policy

Policy Name: Genetic Testing - Specialty Testing: Ophthalmology (GBP)

Effective Date: 01/01/2026

Important Information – Please Read Before Using This Policy

These services may or may not be covered by all Medica Central plans. Coverage is subject to requirements in applicable federal or state laws. Please refer to the member's plan document for other specific coverage information. If there is a difference between this general information and the member's plan document, the member's plan document will be used to determine coverage. With respect to Medicare, Medicaid, and other government programs, this policy will apply unless these programs require different coverage.

Members may contact Medica Customer Service at the phone number listed on their member identification card to discuss their benefits more specifically. Providers with questions may call the Provider Service Center. Please use the Quick Reference Guide on the Provider Communications page for the appropriate phone number. <https://mo-central.medica.com/Providers/SSM-employee-health-plan-for-IL-MO-OK-providers>

Medica Central coverage policies are not medical advice. Members should consult with appropriate health care providers to obtain needed medical advice, care, and treatment.

OVERVIEW

This policy addresses the use of diagnostic tests for eye and vision disorders.

Patients may be referred from primary care to an ophthalmologist or medical geneticist for investigation and management of age-related macular degeneration. In all cases, the patient should receive counseling from a physician with expertise in inherited disease or a genetic counselor.

Whenever clinical findings suggest the presence of an inherited eye disease, the treating ophthalmologist should either discuss the potential value of genetic testing with their patient and order the appropriate tests (if any) or should offer a referral to another physician or counselor with expertise in the selection and interpretation of genetic tests. Treating physicians should also ensure that their patients receive a written copy of their genetic test results.

For additional information see the [Rationale and References](#) section.

The tests, CPT codes, and ICD codes referenced in this policy are not comprehensive, and their inclusion does not represent a guarantee of coverage or non-coverage. Please see the [Concert Platform](#) for additional registered tests.

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POLICY REFERENCE TABLE

<u>COVERAGE CRITERIA SECTIONS</u>	EXAMPLE TESTS (LABS)	COMMON BILLING CODES	SUPPORT
<u>Inherited Retinal Dystrophies</u>			
<u>Inherited Retinal Dystrophies Multigene Panel Analysis</u>	Inherited Retinal Dystrophies Panel (PreventionGenetics, part of Exact Sciences)	81404, 81406, 81408, 81434, 81479, H35.50-H35.54	Rationale/References
	Leber Congenital Amaurosis Panel (PreventionGenetics, part of Exact Sciences)		
<u>Macular Degeneration</u>			
<u>Macular Degeneration</u>	Vita Risk - 0205U (Arctic Medical Laboratories)	81404, 81405, 81408, 81479, 81599, 0205U, H35.30, H35.3110-	Rationale/References
	Macular Degeneration NGS Panel (Fulgent Genetics)	H35.3194, H35.3210-H35.3293, Z13.5	
<u>Other Covered Eye Disorders</u>			
<u>Other Covered Eye Disorders</u>	See list below	81400, 81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408	Rationale/References

RELATED POLICIES

This policy document provides coverage criteria for testing related to eye and vision disorders. Please refer to:

- **Oncology Testing: Hereditary Cancer** for coverage criteria related to genetic testing for hereditary cancer predisposition syndromes.
- **Specialty Testing: Multisystem Genetic Conditions** for coverage criteria related to diagnostic tests for genetic disorders that affect multiple organ systems (e.g., whole exome and genome sequencing, chromosomal microarray, and multigene panels for broad phenotypes).
- **General Approach to Laboratory Testing** for coverage criteria related to eye and vision disorders, including known familial variant testing, that is not specifically discussed in this or another non-general policy.

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COVERAGE CRITERIA

INHERITED RETINAL DYSTROPHIES

Inherited Retinal Dystrophies Multigene Panel Analysis

- I. Genetic testing for inherited [retinal dystrophies](#) via a multigene panel is considered **medically necessary** when:
 - A. The member has findings consistent with one of the following:
 1. Rod-cone degeneration (e.g., retinitis pigmentosa), **OR**
 2. Cone-rod degeneration (e.g., achromatopsia), **OR**
 3. Chorioretinal degeneration, **OR**
 4. Macular dystrophy, **AND**
 - B. The test includes, at a minimum, [the RPE65 gene](#).
- II. Genetic testing for inherited [retinal dystrophies](#) via a multigene panel is considered **investigational** for all other indications.

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MACULAR DEGENERATION

Macular Degeneration

- I. Genetic testing for macular degeneration is considered **investigational** for all indications.

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OTHER COVERED EYE DISORDERS

Other Covered Eye Disorders

The following is a list of conditions that have a known genetic association. Due to their relative rareness, it may be appropriate to approve claims for these tests to establish or confirm a diagnosis.

- I. Genetic testing to establish or confirm one of the following eye disorders to guide management is considered **medically necessary** when the member demonstrates clinical features¹ consistent with the disorder (the list is not meant to be comprehensive, see II below):
 - A. [Duane Syndrome](#)
 - B. [Familial Exudative Vitreoretinopathy](#)
 - C. [Aniridia](#)

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D. [X-linked Congenital Retinoschisis](#)

E. [Presenile Cataracts](#).

- II. Genetic testing to establish or confirm the diagnosis of all other eye disorders not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in the *General Approach to Laboratory Testing* (see policy for coverage criteria).

¹Clinical features for a specific disorder may be outlined in resources such as [GeneReviews](#), [OMIM](#), [National Library of Medicine](#), [Genetics Home Reference](#), or other scholarly sources.

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PRIOR AUTHORIZATION

Prior authorization is not required. However, services with specific coverage criteria may be reviewed retrospectively to determine if criteria are being met. Retrospective denial may result if criteria are not met.

RATIONALE AND REFERENCES

Inherited Retinal Dystrophies Multigene Panel Analysis

Food and Drug Administration (FDA)

The FDA issued an approval letter on December 19, 2017 for Luxturna stating, “Under this license, you are authorized to manufacture the product voretigene neparvovec-rzyl, which is indicated for the treatment of patients with confirmed biallelic *RPE65* mutation-associated retinal dystrophy” (p. 1).

U.S Food & Drug Administration. Approval letter for Luxturna. Approved December 19, 2027. <https://www.fda.gov/media/109487/download>

American Academy of Ophthalmology (AAO)

The American Academy of Ophthalmology Clinical Statement (2022) provides recommendations and clinical genetic assessments of patients with inherited retinal degenerations. Next generation sequencing using a retinal dystrophy panel is an efficient first step for genetic testing and should include genes for syndromic forms of retinal disease even in patients without syndromic features. Patients would also need to have genetic testing to determine eligibility for the FDA approved voretigene neparvovec or be considered for clinical trials. Genetic testing is recommended in patients with any of four major types of inherited retinal degenerations (rod-cone degenerations, cone-rod degenerations, chorioretinal degenerations and inherited macular dystrophies).

American Academy of Ophthalmology. Guidelines on clinical assessment of patients with inherited retinal degenerations. Clinical Statement. October 2022. <https://www.aao.org/education/clinical-statement/guidelines-on-clinical-assessment-of-patients-with>

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Macular Degeneration

American Society of Retina Specialists

American Society of Retina Specialists (2017) published special correspondence on the use of genetic testing in the management of patients with age-related macular degeneration, which made the following conclusions:

1. “Age-related macular degeneration (AMD) genetic testing may provide information on the progression rates from intermediate to advanced AMD. However, before ordering this testing, retina specialists should be aware of the following:
 - a. At present, there is no clinical evidence that altering the management of genetically higher risk progression patients, for example, with more frequent office visits and/or improved lifestyle changes, results in better visual outcomes for these patients compared with individuals of lower genetic susceptibility. As such, prospective studies are needed before patient care is modified.
2. Age-related macular degeneration genetic testing at present in patients with neovascular AMD does not provide clinically relevant information regarding response to anti-vascular endothelial growth factor (VEGF) treatment and is not recommended for this purpose.
3. Although genetic testing to determine the optimal nutritional supplementation may in the future prove useful, at present there is insufficient data to support the use of genetic testing in patients with AMD prior to recommendation of current Age-Related Eye Disease Study (AREDS) nutritional supplement use” (p. 75).

Csaky KG, Schachat AP, Kaiser PK, Small KW, Heier JS; American Society of Retina Specialists Genetics Task Force. The use of genetic testing in the management of patients with age-related macular degeneration: American Society of Retina Specialists Genetics Task Force Special Report. *J VitreoRetinal Dis.* 2017;1(1):75-78. doi:10.1177/2474126416680671

American Academy of Ophthalmology (AAO)

A Preferred Practice Pattern published in 2020 concluded that there is no evidence to support the need for genotyping to guide recommendations for use of supplements containing antioxidants and zinc in AMD (age-related macular degeneration) (p. P15). In addition they state that routine use of genetic testing is not supported by existing literature and is not recommended at this time (p. P16).

Flaxel CJ, Adelman RA, Bailey ST, et al. Age-related macular degeneration preferred practice pattern *Ophthalmology.* 2020;127(1):P1-P65. doi:10.1016/j.ophtha.2019.09.024 Erratum in: *Ophthalmology.* 2020 Sep;127(9):1279.

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Other Covered Eye Disorders

American Academy of Ophthalmology (AAO)

The American Academy of Ophthalmology (AAO) Task Force on Genetic Testing published the following recommendations for genetic testing of inherited eye diseases (2012, revised 2014):

1. “Offer genetic testing to patients with clinical findings suggestive of a Mendelian disorder whose causative gene(s) have been identified. If unfamiliar with such testing, refer the

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patient to a physician or counselor who is. In all cases, ensure that the patient receives counseling from a physician with expertise in inherited disease or a certified genetic counselor.

2. Use Clinical Laboratories Improvement Amendments– approved laboratories for all clinical testing. When possible, use laboratories that include in their reports estimates of the pathogenicity of observed genetic variants that are based on a review of the medical literature and databases of disease-causing and non–disease-causing variants.
3. Provide a copy of each genetic test report to the patient so that she or he will be able independently to seek mechanism-specific information, such as the availability of gene-specific clinical trials, should the patient wish to do so.
4. Avoid direct-to-consumer genetic testing and discourage patients from obtaining such tests themselves. Encourage the involvement of a trained physician, genetic counselor, or both for all genetic tests so that appropriate interpretation and counseling can be provided.
5. Avoid unnecessary parallel testing— order the most specific test(s) available given the patient’s clinical findings. Restrict massively parallel strategies like whole-exome sequencing and whole-genome sequencing to research studies conducted at tertiary care facilities.
6. Avoid routine genetic testing for genetically complex disorders like age-related macular degeneration and late-onset primary open-angle glaucoma until specific treatment or surveillance strategies have been shown in 1 or more published prospective clinical trials to be of benefit to individuals with specific disease-associated genotypes. In the meantime, confine the genotyping of such patients to research studies.
7. Avoid testing asymptomatic minors for untreatable disorders except in extraordinary circumstances. For the few cases in which such testing is believed to be warranted, the following steps should be taken before the test is performed: (1) the parents and child should undergo formal genetic counseling, (2) the certified counselor or physician performing the counseling should state his or her opinion in writing that the test is in the family’s best interest, and (3) all parents with custodial responsibility for the child should agree in writing with the decision to perform the test” (p. 4 and 5).

Stone EM, Aldave AJ, Drack AV, et al. Recommendations for genetic testing of inherited eye diseases: report of the American Academy of Ophthalmology task force on genetic testing. *Ophthalmology*. 2012;119(11):2408-2410. doi:10.1016/j.ophtha.2012.05.047

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DEFINITIONS

1. **Retinal dystrophies** are degenerative diseases of the retina, which have marked clinical and genetic heterogeneity. Vision impairment may vary from poor peripheral or night vision to complete blindness, and severity usually increases with age.
2. **The RPE65 gene** encodes the RPE54 protein, which is an all trans-11-cis-retinyl isomerase, a key enzyme expressed in the retinal pigment epithelium (RPE) that is responsible for regeneration of 11-cis-retinol in the visual cycle.

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ADDITIONAL REFERENCES

1. MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US). Available from: <https://medlineplus.gov/genetics/>
2. Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK11116/>
3. Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD). World Wide Web URL: <https://omim.org/>

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Note: The Health Plan uses the genetic testing clinical criteria developed by Concert Genetics, an industry-leader in genetic testing technology assessment and policy development.

	Committee/Source	Date(s)
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