



Medica Central Coverage Policy

Policy Name: Genetic Testing - Specialty Testing: Dermatology

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 tests for rare dermatologic conditions, such as congenital ichthyosis and others. Pre-test and post-test genetic counseling that facilitates informed decision-making, addresses the possibility of secondary or incidental findings, and a plan for returning results before testing occurs is strongly advised.

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.

POLICY REFERENCE TABLE

COVERAGE CRITERIA SECTIONS	EXAMPLE TESTS (LABS)	COMMON BILLING CODES	SUPPORT
Capillary Malformation-Arteriovenous Malformation Syndrome (CM-AVM)			

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<u>COVERAGE CRITERIA SECTIONS</u>	<u>EXAMPLE TESTS (LABS)</u>	<u>COMMON BILLING CODES</u>	<u>SUPPORT</u>
RASA1 and EPHB4 Sequencing and/or Deletion/Duplication Analysis or Multigene Panel	Capillary Malformation-Arteriovenous Malformation Syndrome (CM-AVM) Panel, Sequencing and Deletion/Duplication (ARUP Laboratories)	81479, Q27.3, Q27.9	Rationale/References
	Vascular Malformation NGS Panel (Greenwood Genetic Center - Molecular Diagnostic Laboratory)		
	RASA1 Full Gene Sequencing and Deletion/Duplication (Invitae Corporation)		
	EPHB4 Full Gene Sequencing and Deletion/Duplication (Invitae Corporation)		
<u>Congenital Ichthyosis</u>			
Congenital Ichthyosis Multigene Panels	Ichthyosis Panel (Blueprint Genetics)	81405, 81479, Q80	Rationale/References
	Ichthyosis NGS Panel (HNL Lab Medicine)		
	Invitae Congenital Ichthyosis Panel (Invitae Corporation)		
<u>Other Covered Dermatologic Conditions</u>			
Other Covered Dermatologic Conditions	See Below	81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408, 81479	Additional References

RELATED POLICIES

This policy document provides coverage criteria for congenital dermatologic conditions. 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).

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- **Specialty Testing: Hematology** for coverage criteria related to diagnostic tests for benign (non-cancerous) hematologic conditions including sickle cell disease, inherited anemias, and hemophilias.
- **General Approach to Laboratory Testing** for coverage criteria related to dermatology, including known familial variant testing, that is not specifically discussed in this or another non-general policy.

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COVERAGE CRITERIA

CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION SYNDROME (CM-AVM)

RASA1 and *EPHB4* Sequencing and/or Deletion/Duplication Analysis or Multigene Panel

- I. *RASA1* and *EPHB4* sequencing and/or deletion/duplication analysis or multigene panel analysis to establish a diagnosis of capillary malformation-arteriovenous malformation (CM-AVM) syndrome is considered **medically necessary** when:
 - A. The member displays one or more of the following:
 1. Capillary malformations, **OR**
 2. Arteriovenous malformations/arteriovenous fistulas, **OR**
 3. Parkes Weber syndrome phenotype, a cutaneous capillary malformation associated with underlying multiple micro-AVFs and soft-tissue and skeletal hypertrophy of the affected limb.
- II. *RASA1* and *EPHB4* sequencing and/or deletion/duplication analysis or multigene panel analysis to establish a diagnosis of capillary malformation-arteriovenous malformation (CM-AVM) syndrome is considered **investigational** for all other indications.

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CONGENITAL ICHTHYOSIS

Congenital Ichthyosis Multigene Panels

- I. Multigene panel analysis to establish or confirm a diagnosis of congenital ichthyosis is considered **medically necessary** when:
 - A. The member has scaly skin with or without a history of harlequin ichthyosis, collodion membrane, or thick, hyperkeratotic skin, **AND**
 - B. The member displays one or more of the following:
 1. Ectropion (eversion of eyelids), **OR**
 2. Eclabium (eversion of lips), **OR**

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3. Scarring alopecia, **OR**
 4. Palmar and/or plantar hyperkeratosis, **OR**
 5. Erythroderma (red skin).
- II. Multigene panel analysis to establish or confirm a diagnosis of congenital ichthyosis is considered **investigational** for all other indications.

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OTHER COVERED DERMATOLOGIC CONDITIONS

Other Covered Dermatologic Conditions

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

- I. Genetic testing to establish or confirm one of the following dermatologic conditions to guide management is considered **medically necessary** when the member demonstrates clinical features consistent with the condition (the list is not meant to be comprehensive, see II below):
 - A. [Hidrotic Ectodermal Dysplasia 2 \(Clouston Syndrome\)](#) (*GJB6*)
 - B. [Hypohidrotic Ectodermal Dysplasia](#) (*EDA, EDAR, EDARADD, WNT10A*)
 - C. [Ocular albinism, X-linked](#) (*GPR143*)
 - D. [Oculocutaneous albinism](#) (*TYR, OCA2, TYRP1, SLC45A2, SLC24A5, LRMDA, DCT, GPR143*)
 - E. [Epidermolysis Bullosa](#) (*CD151, DST, EXPH5, KLHL24, KRT5, KRT14, PLEC*).
- II. Genetic testing to establish or confirm the diagnosis of all other dermatologic conditions not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in *General Approach to Laboratory Testing* (see policy coverage criteria).

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

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RATIONALE AND REFERENCES

***RASA1* and *EPHB4* Sequencing and/or Deletion/Duplication Analysis or Multigene Panel**

GeneReviews: Capillary Malformation-Arteriovenous Malformation Syndrome

GeneReviews is an expert-authored review of current literature on genetic disease, and goes through a rigorous editing and peer review process before being published online.

The recommended diagnostic testing for CM-AVM is as follows:

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"CM-AVM syndrome should be suspected in individuals who have any of the following:

- Capillary malformations (CMs), the hallmark of CM-AVM syndrome. CMs are generally:
 - Multifocal, atypical pink-to-reddish brown, multiple, small (1-2 cm in diameter), round-to-oval lesions sometimes with a white halo;
 - Composed of dilated capillaries in the papillary dermis
 - Mostly localized on the face and limbs;
 - Seen in combination with arteriovenous malformations (AVMs) or arteriovenous fistulas (AVF), but may be the only finding.
- AVMs/AVFs in soft tissue, bone, and brain that may be associated with overgrowth
- Parkes Weber syndrome phenotype, a cutaneous capillary malformation associated with underlying multiple micro-AVFs and soft-tissue and skeletal hypertrophy of the affected limb"

"The diagnosis of CM-AVM syndrome is established in a proband with suggestive clinical findings and a heterozygous pathogenic variant in *EPHB4* or *RASA1* identified by molecular genetic testing."

"When the phenotypic and laboratory findings suggest the diagnosis of CM-AVM syndrome, molecular genetic testing approaches can include use of a multigene panel. A multigene panel that includes *EPHB4*, *RASA1*, and other genes of interest is most likely to identify the genetic cause of the condition at the most reasonable cost while limiting identification of variants of uncertain significance and pathogenic variants in genes that do not explain the underlying phenotype."

Bayrak-Toydemir P, Stevenson DA. Capillary Malformation-Arteriovenous Malformation Syndrome. 2011 Feb 22 [Updated 2019 Sep 12]. In: 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/NBK52764/>

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Congenital Ichthyosis Multigene Panels

GeneReviews: Autosomal Recessive Congenital Ichthyosis

GeneReviews is an expert-authored review of current literature on a genetic disease, and goes through a rigorous editing and peer review process before being published online.

The recommended diagnostic testing for nonsyndromic congenital ichthyosis is as follows:

"Autosomal recessive congenital ichthyosis (ARCI) encompasses several forms of nonsyndromic ichthyosis. Although most neonates with ARCI are collodion babies, the clinical presentation and severity of ARCI may vary significantly, ranging from harlequin ichthyosis, the most severe and often fatal form, to lamellar ichthyosis (LI) and (nonbullous) congenital ichthyosiform erythroderma (CIE). These phenotypes are now recognized to fall on a continuum; however, the phenotypic descriptions are clinically useful for clarification of prognosis and management."

- The diagnosis of ARCI is established in a proband (typically an infant):
 - With scaly skin with or without a history of harlequin ichthyosis, collodion membrane, or thick, hyperkeratotic skin AND the later development of ONE of the following:
 - Classic lamellar ichthyosis (LI). Brown, plate-like scale over the entire body, associated with ectropion (eversion of eyelids), eclabium (eversion of lips), scarring alopecia, and palmar and plantar hyperkeratosis
 - (Nonbullous) congenital ichthyosiform erythroderma (CIE). Erythroderma (red skin) with fine, white scale and often with palmoplantar hyperkeratosis

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- Intermediate forms with some features of both LI and CIE, or nonLI/nonCIE form with mild hyperkeratosis;

AND/OR

- By identification of biallelic pathogenic variants in one of the genes listed below.
 - "The twelve genes known to be associated with ARCI are *ABCA12*, *ALOX12B*, *ALOXE3*, *CASP14*, *CERS3*, *CYP4F22*, *LIPN*, *NIPAL4*, *PNPLA1*, *SDR9C7*, *SLC27A4*, *SULT2B1*, and *TGM1*. A multigene panel that includes these genes is the diagnostic test of choice. If such testing is not available, single-gene testing can be considered starting with *ABCA12* in individuals with harlequin ichthyosis, *TGM1* in individuals with ARCI without harlequin presentation at birth and *SLC27A4* in those presenting with ichthyosis-prematurity syndrome."

Richard G. Autosomal Recessive Congenital Ichthyosis. 2001 Jan 10 [Updated 2023 April 20]. In: 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/NBK1420/>

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ADDITIONAL REFERENCES

1. 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/NBK1116/>
2. Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD). World Wide Web URL: <https://omim.org/>
3. MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US). Available from: <https://medlineplus.gov/genetics/>

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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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