BSC_CON_2.23	Genetic Testing: Lung Disorders		
Original Policy Date:	December 1, 2023	Effective Date:	May 1, 2025
Section:	2.0 Medicine	Page:	Page 1 of 10

## **Example Test Table**

The tests and associated laboratories and CPT codes contained within this document serve only as examples to help users navigate claims and corresponding coverage criteria; as such, they are not comprehensive and are not a guarantee of coverage or non-coverage. Please see the <u>Concert Genetics</u> Platform for a comprehensive list of registered tests.

Policy Statement Locations	Example Tests, Labs	Common CPT Codes		
Alpha-1 Antitrypsin Deficiency				
SERPINAI Common Variant Anglysis or Seguensing and Jor	Alpha-1 Antitrypsin (AAT) Mutation Analysis (Quest Diagnostics)	81332		
Analysis or Sequencing and/or Deletion/Duplication Analysis	SERPINA1 Full Gene Sequencing and Deletion/Duplication (Invitae)	81479		
Donor-Derived Cell-free DNA for Lung Transplant Rejection				
Evidence-Based Donor-Derived Cell-free DNA for Lung Transplant	Prospera Lung (Natera)	81479, 0540U		
Rejection	AlloSure Lung (CareDx)	181479, 03400		
Emerging Evidence Donor-Derived Cell-free DNA for Lung Transplant Rejection	Eurofins TRAC dd-cfDNA (Transplant Genomics Inc)	0118U		
Other Covered Lung Disorders				
Other Covered Lung Disorders	See list below	81400, 81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408		

## **Policy Statement**

#### Alpha-1 Antitrypsin Deficiency

## SERPINAI Common Variant Analysis or Sequencing and/or Deletion/Duplication Analysis

- I. SERPINA1 common variant analysis (81332) or sequencing and/or deletion/duplication analysis (81479) to establish a diagnosis of alpha-1 antitrypsin (AAT) deficiency may be considered medically necessary when:
  - A. The member has **any** of the following:
    - 1. Abnormally low (less than 120 mg/dL) or borderline (90 to 140 mg/dL) alpha-1 antitrypsin levels (as measured by nephelometry)
    - 2. Early-onset emphysema (45 years of age or younger)
    - 3. Emphysema in the absence of additional risk factor (e.g., smoking, occupational dust exposure)
    - 4. Emphysema with prominent basilar hyperlucency
    - 5. Otherwise unexplained liver disease
    - 6. Necrotizing panniculitis
    - 7. C-ANCA positive vasculitis (i.e., granulomatosis with polyangiitis)
    - 8. Bronchiectasis without evident etiology
    - 9. A sibling with known AAT deficiency.

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II. SERPINA1 common variant analysis (81332) or sequencing and/or deletion/duplication analysis (81479) to establish a diagnosis of alpha-1 antitrypsin deficiency is considered investigational for all other indications.

## Donor-Derived Cell-Free DNA For Lung Transplant Rejection Evidence-Based Donor-Derived Cell-free DNA for Lung Transplant Rejection

- III. The use of peripheral blood measurement of donor-derived cell-free DNA tests (81479, 0540U) with sufficient evidence of clinical utility and validity in the management of patients after lung transplantation may be considered **medically necessary** when:
  - A. The member has undergone lung transplantation, AND
  - B. The test has not been performed in the last 12 months, AND
  - C. The member meets at least one of the following:
    - 1. The member has clinical signs of acute rejection, **OR**
    - 2. A biopsy was done and is inconclusive for rejection, OR
    - 3. The member is being monitored for adequate immunosuppression.
- IV. The use of peripheral blood measurement of donor-derived cell-free DNA tests (81479, 0540U) in the management of patients after lung transplantation is considered **investigational** for all other indications.

## Emerging Evidence Donor-Derived Cell-free DNA for Lung Transplant Rejection

V. Donor-derived cell-free DNA tests with insufficient evidence of clinical validity (0118U) in the management of patients after lung transplantation are considered **investigational**.

## Other Covered Lung Disorders

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.

- VI. Genetic testing to establish or confirm one of the following genetic lung disorders to guide management may be considered **medically necessary** when the member demonstrates clinical features\* consistent with the disorder (the list is not meant to be comprehensive, see VII below):
  - A. Familial Pulmonary Fibrosis
  - B. Primary Ciliary Dyskinesia
  - C. Pulmonary lymphangioleiomyomatosis (LAM)
  - D. Pulmonary alveolar proteinosis (PAP)
- VII. Genetic testing to establish or confirm the diagnosis of all other lung disorders not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in *General Approach to Genetic and Molecular Testing* (see policy for coverage criteria).

\*Clinical features for a specific disorder may be outlined in resources such as <u>GeneReviews</u>, <u>OMIM</u>, <u>National Library of Medicine</u>, <u>Genetics Home Reference</u>, or other scholarly source.

NOTE: Refer to Appendix A to see the policy statement changes (if any) from the previous version.

## **Policy Guidelines**

#### Coding

See the **Codes table** for details.

## Description

One of the most common inherited lung disorders is alpha-1 antitrypsin deficiency (AATD). AATD is an autosomal recessive genetic disorder that results in decreased production of the alpha-1 antitrypsin (AAT) protein, or production of abnormal types of the protein that are functionally deficient. Individuals with AATD have an increased risk to develop lung and liver disease. Genetic testing to diagnose AATD aids in directing proper treatment and identifying at-risk family members. With the use of donor-derived cell-free DNA (dd-cfDNA), biomarker tests have been developed as an alternative to more invasive procedures for post-lung transplant care to optimize graft longevity while avoiding side effects and toxicity of immunosuppressive therapies.

## **Related Policies**

This policy document provides coverage criteria for Genetic Testing for Lung Disorders. Please refer to:

- Genetic Testing: Multisystem Inherited Disorders, Intellectual Disability, and
   Developmental Delay for coverage criteria related to diagnostic testing for cystic fibrosis
   and other multisystem inherited disorders.
- Genetic Testing: General Approach to Genetic and Molecular Testing for coverage criteria related to genetic testing for lung disorders and disease that are not specifically discussed in this or another non-general policy, including known familial variant testing.

## **Benefit Application**

Benefit determinations should be based in all cases on the applicable contract language. To the extent there are any conflicts between these guidelines and the contract language, the contract language will control. Please refer to the member's contract benefits in effect at the time of service to determine coverage or non-coverage of these services as it applies to an individual member.

Some state or federal mandates (e.g., Federal Employee Program [FEP]) prohibits plans from denying Food and Drug Administration (FDA)-approved technologies as investigational. In these instances, plans may have to consider the coverage eligibility of FDA-approved technologies on the basis of medical necessity alone.

## **Regulatory Status**

#### State:

SB 496 requires health plans licensed under the Knox-Keene Act ("Plans"), Medi-Cal managed care plans ("MCPS"), and health insurers ("Insurers") to cover biomarker testing for the diagnosis, treatment, appropriate management, or ongoing monitoring of an enrollee's disease or condition to guide treatment decisions, as prescribed. The bill does not require coverage of biomarker testing for screening purposes. Restricted or denied use of biomarker testing for these purposes is subject to state and federal grievance and appeal processes. Where biomarker testing is deemed medically necessary, Plans and Insurers must ensure that the testing is provided in a way that limits disruptions in care.

## Rationale

#### ALPHA-1 ANTITRYPSIN DEFICIENCY

SERPINAI Common Variant Analysis or Sequencing and/or Deletion/Duplication Analysis

American Thoracic Society and European Respiratory Society

The American Thoracic Society and European Respiratory Society published a joint statement on the diagnosis and management of individuals with alpha-1 antitrypsin deficiency (2003) which provided recommendations for diagnostic testing.

A normal range of plasma alpha-1 antitrypsin (measured via nephelometry) is 83/120 - 200/220 mg/dL. Individuals with borderline normal levels of plasma alpha-1 antitrypsin (90-140 mg/dL) or with abnormally low levels (below 120 mg/dL) should be evaluated for alpha-1 antitrypsin deficiency. (p. 826 and 827)

"The following features should prompt suspicion by physicians that their patient may be more likely to have AAT deficiency:

- Early-onset emphysema (age of 45 years or less)
- Emphysema in the absence of a recognized risk factor (smoking, occupational dust exposure, etc.)
- Emphysema with prominent basilar hyperlucency
- Otherwise unexplained liver disease
- Necrotizing panniculitis
- Anti-proteinase 3-positive vasculitis (C-ANCA [anti-neutrophil cytoplasmic antibody]-positive vasculitis)
- Family history of any of the following: emphysema, bronchiectasis, liver disease, or panniculitis
- Bronchiectasis without evident etiology..." (p. 820)

The statement also recommended that individuals with a sibling with AAT deficiency should also be offered genetic testing. (p. 827)

# DONOR-DERIVED CELL-FREE DNA FOR LUNG TRANSPLANT REJECTION Evidence-Based Donor-Derived Cell-free DNA for Lung Transplant Rejection

Centers for Medicare and Medicaid Services

The CMS local coverage determination (LCD) entitled "MolDX: Molecular Testing for Solid Organ Allograft Rejection" states the following regarding donor-derived cell-free DNA tests in individuals who have had solid organ transplantation:

"This Medicare contractor will provide limited coverage for molecular diagnostic tests used in the evaluation and management of patients who have undergone solid organ transplantation. These tests can inform decision making along with standard clinical assessments in their evaluation of organ injury for active rejection (AR).

These tests may be ordered by qualified physicians considering the diagnosis of AR affiliated with a transplant center, helping to rule in or out this condition when assessing the need for or results of a diagnostic biopsy. They should be considered along with other clinical evaluations and results and may be particularly useful in patients with significant contraindications to invasive procedures. The intended use of the test must be:

- To assist in the evaluation of adequacy of immunosuppression, wherein a non-invasive or minimally invasive test can be used in lieu of a tissue biopsy in a patient for whom information from a tissue biopsy would be used to make a management decision regarding immunosuppression, OR
- As a rule-out test for AR in validated populations of patients with clinical suspicion of rejection with a non-invasive or minimally invasive test to make a clinical decision regarding obtaining a biopsy, OR

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- For further evaluation of allograft status for the probability of allograft rejection after a physician-assessed pretest, OR
- To assess rejection status in patients that have received a biopsy, but the biopsy results are inconclusive or limited by insufficient material."

#### Concert Note

For monitoring patients post lung transplantation, absent clear, specific and evidence-based guideline recommendations for a particular regimen of screening, a default frequency of once every 12 months will be adopted.

## Emerging Evidence Donor-Derived Cell-free DNA for Lung Transplant Rejection

Tests that have limited established clinical utility or validity as defined in the Concert policy for General Approach to Genetic and Molecular testing do not meet the threshold for coverage. Evidence for validity may include a Technology Assessment conducted by an independent third party (e.g.MolDx Tech, ECRI, Optum Genomic) and/or evidence-based guidelines published by professional societies. Such evidence was not identified for the tests referenced by this policy.

## References

- American Thoracic Society; European Respiratory Society. American Thoracic Society/European Respiratory Society statement: standards for the diagnosis and management of individuals with alpha-1 antitrypsin deficiency. Am J Respir Crit Care Med. 2003;168(7):818-900. doi:10.1164/rccm.168.7.818
- 2. Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from: <a href="https://www.ncbi.nlm.nih.gov/books/NBK1116/">https://www.ncbi.nlm.nih.gov/books/NBK1116/</a>
- Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD). World Wide Web URL: https://omim.org/
- 4. MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US). Available from: https://medlineplus.gov/genetics/.
- Centers for Medicare & Medicaid Services. Medicare Coverage Database: Local Coverage Determination. MolDX: Molecular Testing for Solid Organ Allograft Rejection (L38582).
   Available at: <a href="https://www.cms.gov/medicare-coverage-database/view/lcd.aspx?lcdid=38582">https://www.cms.gov/medicare-coverage-database/view/lcd.aspx?lcdid=38582</a>

## Documentation for Clinical Review

#### Please provide the following documentation:

- Name of the test being requested or the Concert Genetics GTU identifier. The Concert Genetics GTU can be found at <a href="https://app.concertgenetics.com">https://app.concertgenetics.com</a>
- CPT codes to be billed for the particular genetic test (GTU required for unlisted codes)
- History and physical and/or consultation notes including:
  - o Clinical findings:
    - > Signs/symptoms leading to a suspicion of genetic condition
    - Family history if applicable
  - o Prior evaluation/treatment:
    - Previous test results (i.e., imagining, lab work, etc.) related to reason for genetic testing
    - > Family member's genetic test result, if applicable
  - Rationale
    - Reason for performing test
    - How test result will impact clinical decision making

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Post Service (in addition to the above, please include the following):

Results/reports of tests performed

## Coding

This Policy relates only to the services or supplies described herein. Benefits may vary according to product design; therefore, contract language should be reviewed before applying the terms of the Policy.

The following codes are included below for informational purposes. Inclusion or exclusion of a code(s) does not constitute or imply member coverage or provider reimbursement policy. Policy Statements are intended to provide member coverage information and may include the use of some codes for clarity. The Policy Guidelines section may also provide additional information for how to interpret the Policy Statements and to provide coding guidance in some cases.

Туре	Code	Description
	0118U	Transplantation medicine, quantification of donor-derived cell-free DNA using whole genome next-generation sequencing, plasma, reported as percentage of donor-derived cell-free DNA in the total cell-free DNA
	0493U	Transplantation medicine, quantification of donor-derived cell-free DNA (cfDNA) using next-generation sequencing, plasma, reported as percentage of donor-derived cell-free DNA (Code effective 10/1/2024)
	0540U	Transplantation medicine, quantification of donor-derived cell-free DNA using next-generation sequencing analysis of plasma, reported as percentage of donor-derived cell-free DNA to determine probability of rejection (Code effective 4/1/2025)
CPT®	81332	SERPINA1 (serpin peptidase inhibitor, clade A, alpha-1 antiproteinase, antitrypsin, member 1) (e.g., alpha-1-antitrypsin deficiency), gene analysis, common variants (e.g., *S and *Z)
	81400	Molecular Pathology Procedure Level 1
	81401	Molecular Pathology Procedure Level 2
	81402	Molecular Pathology Procedure Level 3
	81403	Molecular Pathology Procedure Level 4
	81404	Molecular Pathology Procedure Level 5
	81405	Molecular Pathology Procedure Level 6
	81406	Molecular Pathology Procedure Level 7
	81407	Molecular Pathology Procedure Level 8
	81408	Molecular Pathology Procedure Level 9
	81479	Unlisted molecular pathology procedure
HCPCS	None	

## **Policy History**

This section provides a chronological history of the activities, updates and changes that have occurred with this Medical Policy.

Effective Date	Action
12/01/2023	New policy.
07/01/2024	Annual review. Policy statement, guidelines and literature updated.
01/01/2025	Annual review. Policy statement and literature updated.

Effective Date	Action
05/01/2025	Coding update.

## **Definitions of Decision Determinations**

Medically Necessary: Services that are Medically Necessary include only those which have been established as safe and effective, are furnished under generally accepted professional standards to treat illness, injury or medical condition, and which, as determined by Blue Shield, are: (a) consistent with Blue Shield medical policy; (b) consistent with the symptoms or diagnosis; (c) not furnished primarily for the convenience of the patient, the attending Physician or other provider; (d) furnished at the most appropriate level which can be provided safely and effectively to the patient; and (e) not more costly than an alternative service or sequence of services at least as likely to produce equivalent therapeutic or diagnostic results as to the diagnosis or treatment of the Member's illness, injury, or disease.

**Investigational/Experimental:** A treatment, procedure, or drug is investigational when it has not been recognized as safe and effective for use in treating the particular condition in accordance with generally accepted professional medical standards. This includes services where approval by the federal or state governmental is required prior to use, but has not yet been granted.

**Split Evaluation:** Blue Shield of California/Blue Shield of California Life & Health Insurance Company (Blue Shield) policy review can result in a split evaluation, where a treatment, procedure, or drug will be considered to be investigational for certain indications or conditions, but will be deemed safe and effective for other indications or conditions, and therefore potentially medically necessary in those instances.

## Prior Authorization Requirements and Feedback (as applicable to your plan)

Within five days before the actual date of service, the provider must confirm with Blue Shield that the member's health plan coverage is still in effect. Blue Shield reserves the right to revoke an authorization prior to services being rendered based on cancellation of the member's eligibility. Final determination of benefits will be made after review of the claim for limitations or exclusions.

Questions regarding the applicability of this policy should be directed to the Prior Authorization Department at (800) 541-6652, or the Transplant Case Management Department at (800) 637-2066 ext. 3507708 or visit the provider portal at <a href="https://www.blueshieldca.com/provider">www.blueshieldca.com/provider</a>.

We are interested in receiving feedback relative to developing, adopting, and reviewing criteria for medical policy. Any licensed practitioner who is contracted with Blue Shield of California or Blue Shield of California Promise Health Plan is welcome to provide comments, suggestions, or concerns. Our internal policy committees will receive and take your comments into consideration.

For utilization and medical policy feedback, please send comments to: MedPolicy@blueshieldca.com

Disclaimer: This medical policy is a guide in evaluating the medical necessity of a particular service or treatment. Blue Shield of California may consider published peer-reviewed scientific literature, national guidelines, and local standards of practice in developing its medical policy. Federal and state law, as well as contract language, including definitions and specific contract provisions/exclusions, take precedence over medical policy and must be considered first in determining covered services. Member contracts may differ in their benefits. Blue Shield reserves the right to review and update policies as appropriate.

## Appendix A

POLICY ST	TATEMENT	
BEFORE	AFTER	
Red font: Verbiage removed	Blue font: Verbiage Changes/Additions	
Genetic Testing: Lung Disorders BSC_CON_2.23	Genetic Testing: Lung Disorders BSC_CON_2.23	
Policy Statement:	Policy Statement:	
Alpha-1 Antitrypsin Deficiency	Alpha-1 Antitrypsin Deficiency	
SERPINA1 Common Variant Analysis or Sequencing and/or Deletion/Duplication Analysis	SERPINAI Common Variant Analysis or Sequencing and/or	
I. SERPINAI common variant analysis (81332) or sequencing and/or	Deletion/Duplication Analysis  I. SERPINAI common variant analysis (81332) or sequencing and/or	
deletion/duplication analysis (81479) to establish a diagnosis of	deletion/duplication analysis (81479) to establish a diagnosis of	
alpha-1 antitrypsin (AAT) deficiency may be considered <b>medically</b>	alpha-1 antitrypsin (AAT) deficiency may be considered <b>medically</b>	
necessary when:	necessary when:	
A. The member has <b>any</b> of the following:	A. The member has <b>any</b> of the following:	
<ol> <li>Abnormally low (less than 120 mg/dL) or borderline (90 to 140 mg/dL) alpha-1 antitrypsin levels (as measured by</li> </ol>	<ol> <li>Abnormally low (less than 120 mg/dL) or borderline (90 to 140 mg/dL) alpha-1 antitrypsin levels (as measured by</li> </ol>	
nephelometry)	nephelometry)	
2. Early-onset emphysema (45 years of age or younger)	2. Early-onset emphysema (45 years of age or younger)	
<ol> <li>Emphysema in the absence of additional risk factor (e.g., smoking, occupational dust exposure)</li> </ol>	<ol> <li>Emphysema in the absence of additional risk factor (e.g., smoking, occupational dust exposure)</li> </ol>	
4. Emphysema with prominent basilar hyperlucency	4. Emphysema with prominent basilar hyperlucency	
5. Otherwise unexplained liver disease	5. Otherwise unexplained liver disease	
6. Necrotizing panniculitis	6. Necrotizing panniculitis	
7. C-ANCA positive vasculitis (i.e., granulomatosis with polyangiitis)	7. C-ANCA positive vasculitis (i.e., granulomatosis with polyangiitis)	
8. Bronchiectasis without evident etiology	8. Bronchiectasis without evident etiology	
9. A sibling with known AAT deficiency.	9. A sibling with known AAT deficiency.	
II. SERPINA1 common variant analysis (81332) or sequencing and/or deletion/duplication analysis (81479) to establish a diagnosis of alpha-1 antitrypsin deficiency is considered investigational for all other indications.	II. SERPINA1 common variant analysis (81332) or sequencing and/or deletion/duplication analysis (81479) to establish a diagnosis of alpha-1 antitrypsin deficiency is considered investigational for all other indications.	
Donor-Derived Cell-Free DNA For Lung Transplant Rejection Evidence-Based Donor-Derived Cell-free DNA for Lung Transplant Rejection	Donor-Derived Cell-Free DNA For Lung Transplant Rejection Evidence-Based Donor-Derived Cell-free DNA for Lung Transplant Rejection	

#### **POLICY STATEMENT BEFORE AFTER** Red font: Verbiage removed **Blue font: Verbiage Changes/Additions** III. The use of peripheral blood measurement of donor-derived cell-free The use of peripheral blood measurement of donor-derived cell-free DNA tests (81479) with sufficient evidence of clinical utility and DNA tests (81479, 0540U) with sufficient evidence of clinical utility validity in the management of patients after lung transplantation and validity in the management of patients after lung may be considered **medically necessary** when: transplantation may be considered **medically necessary** when: A. The member has undergone lung transplantation, AND A. The member has undergone lung transplantation, AND B. The test has not been performed in the last 12 months, AND B. The test has not been performed in the last 12 months, AND C. The member meets at least one of the following: C. The member meets **at least one** of the following: 1. The member has clinical signs of acute rejection, **OR** 1. The member has clinical signs of acute rejection, **OR** 2. A biopsy was done and is inconclusive for rejection, **OR** 2. A biopsy was done and is inconclusive for rejection, **OR** 3. The member is being monitored for adequate 3. The member is being monitored for adequate immunosuppression. immunosuppression. IV. The use of peripheral blood measurement of donor-derived cell-free IV. The use of peripheral blood measurement of donor-derived cell-free DNA tests (81479) in the management of patients after lung DNA tests (81479, 0540U) in the management of patients after lung transplantation is considered investigational for all other transplantation is considered investigational for all other indications. indications. Emerging Evidence Donor-Derived Cell-free DNA for Lung Transplant Emerging Evidence Donor-Derived Cell-free DNA for Lung Transplant Rejection Rejection V. Donor-derived cell-free DNA tests with insufficient evidence of V. Donor-derived cell-free DNA tests with insufficient evidence of clinical validity (0118U) in the management of patients after lung clinical validity (0118U) in the management of patients after lung transplantation are considered investigational. transplantation are considered investigational. Other Covered Lung Disorders Other Covered Lung Disorders The following is a list of conditions that have a known genetic association. 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 Due to their relative rareness, it may be appropriate to cover these genetic tests to establish or confirm a diagnosis. tests to establish or confirm a diagnosis. VI. Genetic testing to establish or confirm one of the following genetic VI. Genetic testing to establish or confirm one of the following genetic lung disorders to guide management is considered **medically** lung disorders to guide management may be considered **medically necessary** when the member demonstrates clinical features\* necessary when the member demonstrates clinical features\* consistent with the disorder (the list is not meant to be consistent with the disorder (the list is not meant to be comprehensive, see VII comprehensive, see VII below A. Familial Pulmonary Fibrosis A. Familial Pulmonary Fibrosis B. Primary Ciliary Dyskinesia B. Primary Ciliary Dyskinesia C. Pulmonary lymphangioleiomyomatosis (LAM) C. Pulmonary lymphangioleiomyomatosis (LAM)

POLICY STATEMENT		
BEFORE	AFTER	
Red font: Verbiage removed	Blue font: Verbiage Changes/Additions	
D. Pulmonary alveolar proteinosis (PAP)	D. Pulmonary alveolar proteinosis (PAP)	
VII. Genetic testing to establish or confirm the diagnosis of all other lung disorders not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in <i>General Approach to Genetic and Molecular Testing</i> (see policy for coverage criteria).	VII. Genetic testing to establish or confirm the diagnosis of all other lung disorders not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in <i>General Approach to Genetic and Molecular Testing</i> (see policy for coverage criteria).	
*Clinical features for a specific disorder may be outlined in resources such	*Clinical features for a specific disorder may be outlined in resources such	
as GeneReviews, OMIM, National Library of Medicine, Genetics Home	as <u>GeneReviews</u> , <u>OMIM</u> , <u>National Library of Medicine</u> , <u>Genetics Home</u>	
Reference, or other scholarly source.	Reference, or other scholarly source.	