

Overview

Useful For

Assisting in the diagnosis of hereditary nephritis (Alport syndrome)

Special Instructions

- [Renal Biopsy Patient Information](#)
- [Renal Biopsy Preparation Instructions](#)

Method Name

Direct Immunofluorescence

NY State Available

Yes

Specimen

Specimen Type

Special

Shipping Instructions

1. Advise shipping frozen specimens (unstained slides or tissue block) in Styrofoam transportation coolers filled with dry ice to ensure specimens are received at required specimen stability temperature.
2. Attach the green "Attention Pathology" address label (T498) to the outside of the transport container before putting into the courier mailer.

Necessary Information

A pathology/diagnostic report is required.

Specimen Required

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Unstained slides (unfixed)

Source: Kidney tissue or skin tissue

Slides: 1 Slide

Collection Instructions: Submit 1 frozen tissue unstained positively charged glass slide (25- x 75- x 1-mm) per test ordered; sections should be 4-microns thick, **centered on the slide**, and shipped on dry ice.

Acceptable:

Specimen Type: Unfixed tissue block (O.C.T)

Source: Kidney tissue or skin tissue

Specimen Volume: Entire specimen

Collection Instructions:

- 1. Embed in O.C.T. compound.
- 2. Freeze specimen and ship on dry ice.

Acceptable:

Specimen Type: Wet tissue

Source: Kidney tissue or skin tissue

Supplies: Renal Biopsy Kit (T231)

Container/Tube: Renal Biopsy Kit, Zeus/Michel's

Specimen Volume: Entire specimen

Collection Instructions:

- 1. Collect specimens according to the instructions in [Renal Biopsy Preparation Instructions](#).
- 2. If standard immunoglobulin and complement immunofluorescence has already been performed, ship the residual frozen tissue (must contain glomeruli) on dry ice.

Forms

- 1. [Renal Biopsy Patient Information](#)
- 2. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:
 - [Kidney Transplant Test Request](#)
 - [Renal Diagnostics Test Request](#) (T830)

Specimen Minimum Volume

See Specimen Required

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability

Specimen Stability Information

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Special | Frozen | | |

Clinical & Interpretive

Clinical Information

Alport syndrome is a hereditary disease of basement membrane collagen type IV. Variants in collagen IV alpha genes cause characteristic abnormal immunofluorescence staining patterns within the glomerular basement membrane. Alport syndrome is characterized by hematuria, proteinuria, progressive kidney failure, and high-tone sensorineural hearing loss.

Reference Values

An interpretive report will be provided.

Interpretation

This test (when not accompanied by a pathology consultation request) will be reported as one of the following:

- 1) Normal pattern
- 2) Consistent with X-linked hereditary nephritis
- 3) Consistent with autosomal hereditary nephritis

If additional interpretation or analysis is needed, request PATHC / Pathology Consultation along with this test and send the corresponding renal pathology light microscopy and immunofluorescence (IF) slides (or IF images on a CD), electron microscopy images (prints or CD), and the pathology report.

Cautions

Approximately one-third of patients with established hereditary nephritis based on typical ultrastructural findings and family history show loss of glomerular basement membrane or epidermal basement membrane staining for the alpha 5 chain of type IV collagen. Therefore, a normal staining pattern does not exclude the diagnosis of hereditary nephritis.

Because alpha 3 and alpha 4 chains of type IV collagen are not expressed in the epidermal basement membranes, patients with autosomal hereditary nephritis have preserved staining for alpha 5 on epidermal basement membranes and, therefore, skin biopsy cannot exclude autosomal hereditary nephritis.

Clinical Reference

1. Kagawa M, Kishiro Y, Naito I, et al. Epitope-defined monoclonal antibodies against type-IV collagen for diagnosis of Alport's syndrome. *Nephrol Dial Transplant*. 1997;12(6):1238-1241
2. Hashimura Y, Nozu K, Kaito H, et al. Milder clinical aspects of X-linked Alport syndrome in men positive for the collagen IV alpha 5 chain. *Kidney Int*. 2014;85(5):1208-1213
3. Kamiyoshi N, Nozu K, Fu XJ, et al. Genetic, clinical, and pathologic backgrounds of patients with autosomal dominant Alport syndrome. *Clin J Am Soc Nephrol*. 2016;11(8):1441-1449
4. Said SM, Fidler ME, Valeri AM, et al. Negative staining for COL4A5 correlates with worse prognosis and more severe ultrastructural alterations in males with Alport syndrome. *Kidney Int Rep*. 2016;2(1):44-52

Performance**Method Description**

Direct immunofluorescence staining on sections of fresh/frozen tissue.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Test Definition: ALPRT

Alport (Collagen IV Alpha 5 and Alpha 2)
Immunofluorescent Stain, Renal Biopsy

Report Available

1 to 2 days

Specimen Retention Time

Unstained slides: 2 weeks after results are reported; Stained slides: digital images are obtained for all slides used in testing and kept indefinitely; Unfixed tissue blocks: 5 years

Performing Laboratory Location

Mayo Clinical Laboratories- Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

88346

LOINC® Information

| Test ID | Test Order Name | Order LOINC® Value |
|---------|---------------------------|--------------------|
| ALPRT | ALPORT Immunofluorescence | In Process |

| Result ID | Test Result Name | Result LOINC® Value |
|-----------|------------------------------------|---------------------|
| 71285 | Interpretation | 50595-8 |
| 71268 | Participated in the Interpretation | No LOINC Needed |
| 71272 | Material Received | 81178-6 |
| 71271 | Gross Description | 22634-0 |
| 71269 | Report electronically signed by | 19139-5 |
| 71270 | Addendum | 35265-8 |
| 71619 | Disclaimer | 62364-5 |
| 71848 | Case Number | 80398-1 |