

**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

**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