



Patient ID SA00090845	Patient Name TESTING, GRACE	Birth Date 1999-01-01	Gender F	Age 18
Order Number SA00090845	Client Order Number SA00090845	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 18 May 2017 00:00		

ALPORT Immunofluorescence

Case Number CR-17-3497

Interpretation

MCR

IMMUNOFLUORESCENCE STAINING FOR COLLAGEN IV (ALPHA 2 AND ALPHA 5 CHAINS):

Consistent with autosomal hereditary nephritis (Alport syndrome). There is global or segmental loss of alpha 5 staining of glomerular basement membranes but preserved alpha 5 staining of Bowman's capsule and distal tubular basement membranes. This pattern of loss staining is usually due to mutations in the COL4A3 or COL4A4 genes on chromosome 2.

Notes:

- 1) 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.
- 2) In patients with hereditary nephritis, preserved alpha 5 staining indicates small mutations (e.g., missense, splice site) and is generally associated with a better renal outcome, while loss of alpha 5 staining indicates larger mutations (e.g., deletion, nonsense, frame-shift) and a worse renal outcome.
- 3) 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.
- 4) Kagawa M, et al. Epitope-defined monoclonal antibodies against type-IV collagen for diagnosis of Alport's syndrome. Nephrol Dial Transplant. 1997 Jun;12(6):1238-41.

Report Electronically signed by

MCR

MATTHEW RUETER

I verify that I have examined all relevant slides/materials for the specimen(s) and rendered or confirmed the diagnosis.

Gross Description

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Immunofluorescence: Received in Zeus for immunofluorescence: 1 piece(s) of tissue measuring 0.4 x 0.04 cm. Submitted in total for immunofluorescence. (GEJ)

Material Received

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A. K17-456: Right kidney

1 - Zeus wet tissue

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292



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Disclaimer

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

Received: 19 May 2017 12:34

Reported: 22 May 2017 11:30

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Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292