

Image Quiz 6

CLINICAL HISTORY:

A 29 year old male presented with intermittent pedal edema and feeling of generalized weakness since last six months.

Investigations: 24 hours Urine protein 1.6 gms, Serum Creatinine 1.7 mg% (150.28 micro mol/L)

A renal biopsy was performed. Provided below are images of a glomerulus in unstained cryostat sections, viewed under polarized light, and in sections stained with Masson's Trichrome and Silver Methenamine.

MICROSCOPY:

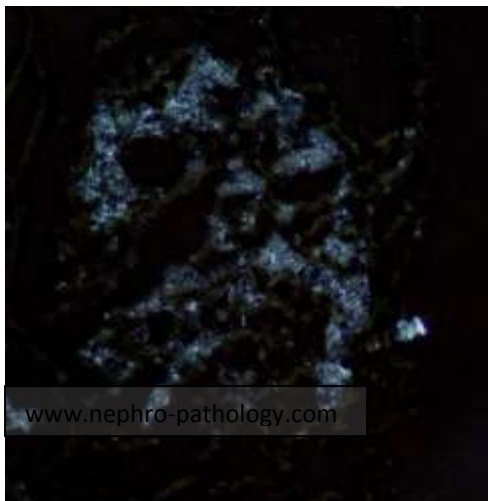


Fig 1

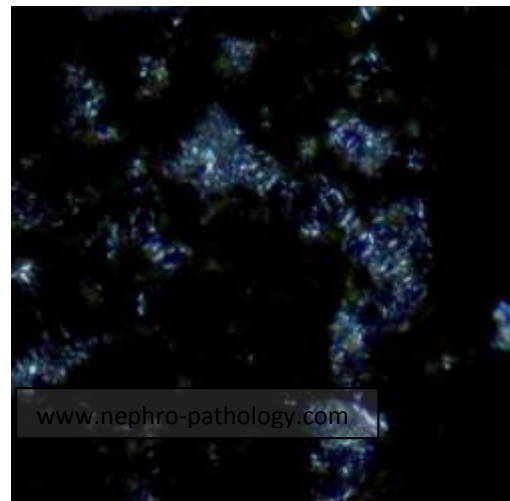


Fig 2

Unstained cryostat sections viewed under polarized light showed refractile/ birefringent material in the glomeruli (mesangial areas, and podocytes)

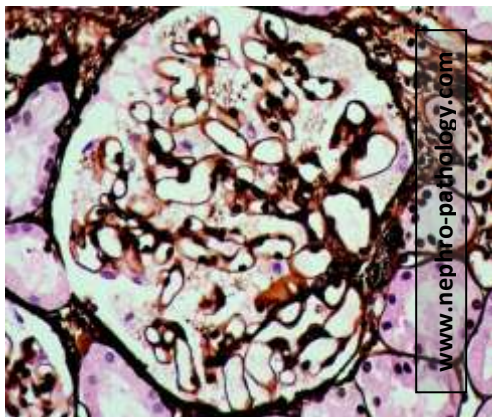


Fig 3

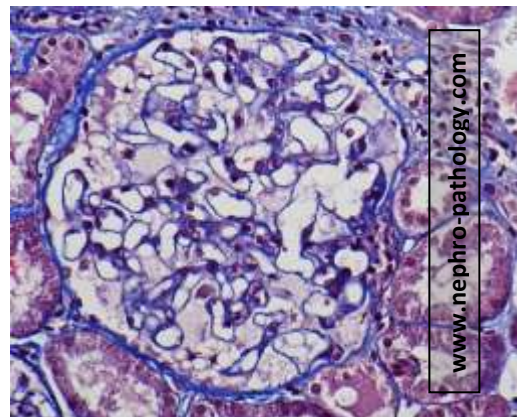


Fig 4

The silver methenamine and Massons Trichrome stained slides revealed prominent vacuolization in visceral epithelial cell cytoplasm, corresponding to the birefringent material noted in the unstained cross sections.

Further Evaluation:

A lysosomal storage disorder was suspected and enzymatic assays revealed nearly undetectable levels of alpha galactosidase A. Ultrastructural studies revealed lamellated osmiophilic structures in podocyte cytoplasm and focally in the tubular epithelial cells.

DIAGNOSIS:

Fabry Disease (with renal involvement)

NOTE : Please see the discussion of this entity in the case of the week section (case 7) on www.nephro-pathology.com