THE KOREAN SOCIETY OF NEPHROLOGY
APCN & KSN 2024
June 13(Thu) - 16(Sun), 2024 Coex, Seoul, Korea
Promoting Sustainable Kidney Health: The Asia-Pacific and Beyond

Abstract Submission No.: A-0791

Alport Syndrome: A Case Report

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Case Study : Alport syndrome is a rare hereditary disease characterized by progressive glomerulonephritis, progressive high-tone hearing loss, and visual impairment. Early diagnosis and intervention can delay the progression of disease and improve the quality of life in these patients. We present a case of a 23 year old male, non hypertensive, non diabetic, with no family history of kidney disease coming in for proteinuria. During pre-employment check-up, patient was noted to have 4+ proteinuria on urinalysis. Creatinine was requested by company doctor with result of 1.05mg/dL (eGFR 104 ml/min/1.73m^2). Repeat urinalysis done but still with 4+ proteinuria on urinalysis. Hence, advised consult with a nephrologist due to persistence of frothy urine and proteinuria. Upon consult, work ups were done which revealed hyperuricemia, urate crystals on urinalysis, persistence of 4+ proteinuria and urine protein creatinine ratio of 2.8 mg/dL. Patient was started on ACE inhibitor, hypouricemic agent and advised kidney biopsy for further evaluation of proteinuria. The review of systems was pertinent for hearing impairment and blurring of vision. Kidney biopsy was done in which electron microscopy showed segmental podocyte foot process effacement. The glomerular basement membrane shows lamellation and alternate thickening and thinning. No definite electron-dense deposits are seen in glomerular basement membrane and mesangium. Mean glomerular basement membrane thickness is 299 nm (normal mean glomerular basement membrane thickness in adult males is 373 +/- 42 nm). He was advised consult with an ophthalmologist and otolaryngologist. Regular checkup, monitoring of renal parameters, and appropriate medications were given. In conclusion, thorough history, physical examination and characteristic findings on kidney biopsy can help in the prompt diagnosis of the disease. Multidisciplinary care and early intervention can improve the quality of life and delay the progression to kidney failure in these patients.

Summary of diagnostic test done .png

Creatinine	1.05 mg/dL (eGFR 104 ml/min/1.73m^2)
Urinalysis	Dark yellow pH 5 Specific gravity: 1.025 WBC: 0-2 RBC: 2-3 CHON: ++++ Epithelial cell: rare Bacteria: few Mucus thread: few Urates: few
Ultrasound of the Kidneys, ureter, bladder and prostate	Both kidneys are normal in size and parenchyma Right kidney 9.6 x 4.3 x 3.7 cm with cortical thickness of 1.4cm Left kidney 10.5 x 4.2 x 4.2 cm with cortical thickness of 1.7cm parenchyma of the right kidney is isoechoic in relation to the liver. Both pelvocalyces are not dilated. There are echogenic structures in the left kidney (0.2cm) The urinary bladder is distended with regular contour and outline. No lithiasis noted. Pre void volume of 487ml Post void scan shows a residual urine 8ml Impression: Normal sized right kidney with isoechoic parenchyma Normal sized kidney with lithiases Normal ultrasound of the urinary bladder with no significant urinary retention Normal sized prostate
Urine protein creatinine ratio (UPCR)	Urine Protein 223.28 Urine crea 79.64 UPCR 2.8 mg/dL
Complete Blood Count	Hemoglobin 15 Hematocrit 37.7 RBC 5 WBC 5900 Neutrophil 61 Lymphocyte 27 Monocyte 12 Platelet count 249,000
Fasting Blood Sugar	5.66 mmol/L (Normal value: 3.9 - 5.8)
Uric acid	599.07 mmol/L (Normal value: 208-428)
Lipid profile	Total cholesterol: 4.64 mmol/L (<5.2) Triglyceride: 2.02 mmol/L (< 2.26) HDL: 1.67 mmol/L (1.36 - 3.06)
Albumin	29.8 g/L (Normal value: 35-53)

Summary of diagnostic test done .png

Gross and Microscopic description	Received fresh are 3 tissue cores measuring 1.4 x 0.1 x 0.1 cm, 1.2 x 0.1 x 0.1 cm, and 0.3 x 0.1 x 0.1 cm, submitted for light, immunofluorescence, and electron microscopy
Light Microscopy	Histologic preparation (H&E, MTS, PAS, PAAg) of 1 core of cortical tissue include 11 glomeruli, 2 of which are globally sclerosed. One glomerulus show ischemic change and periglomerular fibrosis. No definite endocapillary or extracapillary proliferation is seen. Capillary loops are locally thick. There are mononuclear cells, plasma cells, and many foamy macrophages in the interstitium (20%). Tubular atrophy (5%), colloid casts, red blood cells in the tubules, tubular reabsorption droplets, focal tubular vacuolization, and interstitial fibrosis (5%) are present. Three interlobular arteries are unremarkable. There is marked arteriolar hyalinosis.
Immunofluorescence Microscopy	Anti-human IgG, IgA, fibrinogen – 12 glomeruli with diffuse segmental diffuse segmental granular mesangial staining (trace) Anti-human IgM - 12 glomeruli with diffuse granular mesangial staining (trace to 1+) Anti-human C3 - 12 glomeruli with diffuse segmental granular mesangial staining (trace); focal granular vascular staining (1+) Anti-human C1q - 12 glomeruli with negative staining
Electron Microscopy	shows segmental podocyte foot process effacement. The glomerular basement membrane shows lamellation and alternate thickening and thinning. Intracapillary leukocytes are present. No definite electron-dense deposits are seen in glomerular basement membrane and mesangium. Mean glomerular basement membrane thickness is 299 nm (normal mean glomerular basement membrane thickness in adult males is 373 +/- 42 nm
Impression	Glomerular basement membrane abnormality suspicious for alport syndrome Mild interstitial fibrosis and tubular atrophy with 9% global glomerulosclerosis (2 of 23 Glomeruli) Severe hyaline arteriolosclerosis