Fig 1. ECG on admission shows RBBB

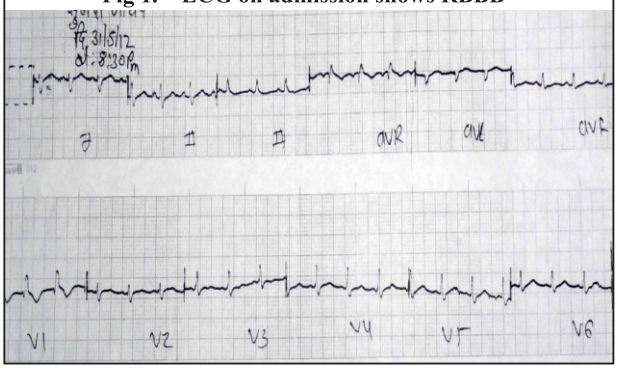


Fig 2:ECG after 1/2 an hr shows Ventricularectopics & Ventricular Bigeminy

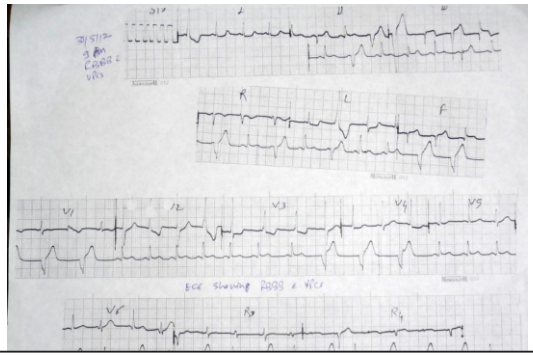


Fig 3: Subsequent ECG on day 3rd shows ST coving & T wave inversion in Lead V1-V3

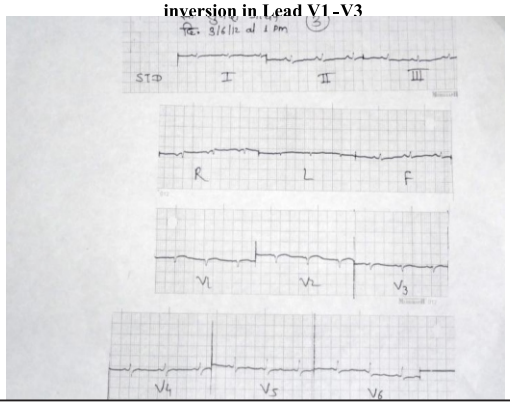
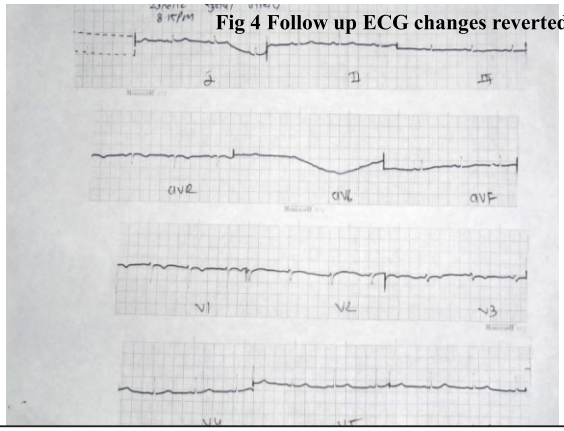


Fig 4 Follow up ECG changes reverted



Alport Syndrome

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Abstract:

Alport Syndrome is an inherited disorder characterized by hematuria and several associated features. Four forms of disease are now recognized: Although Alport syndrome is a rare condition, but proper history with thorough clinical examination (slit lamp & pure tone Audiometry), and relevant investigations helped to clinch the diagnosis.We here4 report 2 cases of Alport syndrome

Introduction:

Alport Syndrome is an inherited disorder characterized by hematuria and several associated features. Four forms of disease are now recognized:

(1) Classic Alport syndrome, which is inherited as an X-linked disorder with hematuria, sensorineural deafness, and conical deformation of the anterior surface of the lens (lenticonus),(2) A subtype of the X-linked form associated with diffuse leiomyomatosis,(3) An autosomal recessive form,(4) An autosomal dominant form.[1] we here report two cases of Alport syndrome.

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Case Report

Case 1:

A 23 years old unmarried female diagnosed case of chronic renal failure came to OPD with complaint of breathlessness. History comprised of episodes of frank hematuria 10years ago, decreased in hearing since last 4 years, diminution of vision since 2 years and 2 cycles of hemodialysis done in outside hospital. She had family history of similar complaints in her elder brother and her father died because of renal failure and also had similar

complaints.

Slit lamp examination was done. It was suggestive of Left eye cataract with anterior Lenticonus, and changes of grade II hypertensive retinopathy. Pure tone audiometry was suggestive of bilateral sensorineural hearing loss.on investigations, Blood urea was 258.3mg/dl & Serum Creatinine:was 11.2mg/dl.USG for KUB revealed chonic kidney disease. A renal biopsy showed diffuse mesangial proliferation with focal thickening. Patient was advised CRRT in the form of regular maintenance hemodialysis.

Case 2:

32years old unmarried male elder brother of case 1 was called for work up.His history comprised of episodes frank of hematuria 4years ago, decreased in hearing since last 2 years and diminution of vision since 1year.Family history was same as 1st case.

Slit lamp examination was suggestive of right eye anterior Lenticonus. Pure Tone audiometry was suggestive of bilateral sensorineural hearing loss.His renal function tests revealed blood Urea: 48mg/dl & serum Creatinine: 4mg/dl. USG for KUB was suggestive of bilateral renal parenchymal disease with loss of corticomedullary differentiation and reduced size suggestive of chronic kidney disease. He was started on medical line of treatment and subsequent need for CRRT in future. Patient is in regular follow up for same.

Discussion

Our first case had six and the second cases had five, out of ten, diagnostic criteria, and were diagnosed as Alport syndrome. In above both cases, patients had history of hematuria, sensorineural deafness, lenticonus, renal failure, and significant family history. Although Alport syndrome is a rare condition, but proper history with thorough clinical examination (slit lamp & pure tone Audiometry), and relevant investigations helped to clinch the diagnosis.

Alport syndrome. was first identified in a British family by Dr. Cecil A. Alport(in 1927)[2,3] Incidence of Alport syndrome is about 1 in 10,000 births in general population. About 80% of Alport syndrome patients have the classical X-linked variant. Most patients have mutations in four of the six genes for the chains of type IV collagen (COL 4A3, COL 4A4, COL 4A5, COL 4A6). COL 4A1 & COL 4A2 genes are head to head on chromosome 13q34, the COL 4A3 & COL 4A4 genes are on chromosome 2q35-37, and the COL 4A5 & COL 4A6 genes are on chromosome Xq22.[1]

Criteria for the clinical diagnosis:

Gregory et al., 1996, gave the following 10 criteria for the diagnosis of Alport syndrome[4] (four of the 10 criteria must be met):

1. Family history of nephritis of unexplained hematuria in a first degree relative of the index case or in a male relative linked through any numbers of females.
2. Persistent hematuria without evidence of another possibly inherited nephropathy such as thin GBM disease, polycystic kidney disease or IgA nephropathy.
3. Bilateral sensorineural hearing loss in the 2000 to 8000 Hz range. The hearing loss develops gradually, is not present in early infancy and commonly presents before the age of 30 years.
4. A mutation in COL4An (where n = 3, 4 or 5).
5. Immunohistochemical evidence of complete or partial lack of the Alport epitope in glomerular, or epidermal basement membranes, or both.
6. Widespread GBM ultrastructural abnormalities, in particular thickening, thinning and splitting.
7. Ocular lesions including anterior Lenticonus, posterior subcapsular cataract, posterior polymorphous dystrophy and retinal flecks.
8. Gradual progression to ESRD in the index case of at least two family members.
9. Macrothrombocytopenia or granulocytic inclusions, similar to the May-Hegglin anomaly.
10. Diffuse leiomyomatosis of esophagus or female genitalia, or both.

Hematologic disorders

Several reports describe families with hereditary nephritis associated with deafness, megathrombocytopenia (giant platelets), and, in some families, granulocyte abnormalities. Clinical features include bleeding tendency, Macrothrombocytopenia, abnormalities of platelet aggregation, and, occasionally, neutrophil inclusions that resemble Dohle bodies (i.e, May-Hegglin anomaly, Fechtner syndrome).

Our both patient didn't had any hematological abnormality except for normocytic normochromic anemia secondary to CRF in case 1.

No specific treatment exists for patients with Alport syndrome, but those who develop ESRD are offered renal transplantation and usually have excellent allograft survival rates. The cause of anti-GBM nephritis is unclear, but about 3-5% of males with Alport syndrome who undergo renal transplantation develop this disorder. These individuals usually have early onset Alport syndrome with clinically significant hearing loss and ESRD by about age