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Case Report on Chronic Kidney disease with Bardet-Biedl Syndrome

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ABSTRACT

Background: Mutations in proteins present in the main basal body complex cause Bardet–Biedl syndrome, an autosomal recessive multiorgan disorder. The incidence of the syndrome is estimated to be 1 in every 160,000 people. Only about 15 cases have been reported in India.
Clinical Findings: Breathlessness, pedal edema.
Diagnostic Evaluation: Blood test: Hb- 8.2gm%, Total RBC count–2.82 millions/cu mm, HCT–24.4%, Total WBC count–8400/cu mm; KFT: Urea–88mg%, Creatinine–4.2 mg%, Potassium–5.4meq/l; LFT: ALT(SGPT)-75IU/l, ALT(SGOT)-32IU/l, Total Bilirubin-0.9mg%
Ultrasonography abdomen & pelvis: There is mild to moderate fluid collection in peritoneal cavity & laceration on right lobe of liver; 2D Echo showed Mild left ventricular hypertrophy
Therapeutic intervention: Haemodialysis, Blood transfusion, Antibiotics, Fluid & Electrolyte
Outcome: After treatment, patient show improvement. His Breathlessness and pedal edema were relieved and his Hb% increased from 8.2gm% to Total Bilirubin become normal.
Conclusion: This patient was admitted to MICU, AVBRH with a known case of CKD with Bardet Biedl syndrome and he had complaint of breathlessness and pedal edema. After getting appropriate treatment his condition was improved.

Key words: Bardet-biedl syndrome, Ciliopathy, Genome editing, Targeted therapies

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INTRODUCTION

Retinal degeneration, post-axial polydactyly, difficulties in learning, Obesity, renal failure, and hypogonadism are all symptoms of Bardet–Biedl syndrome (BBS). Many small traits that are related with BBS might help with diagnosis and are crucial in therapeutic care. The diagnosis is based on clinical signs in 80% of cases and can be verified by sequencing known disease-causing genes. BBS genes, which encode proteins found in the cilia / basal body, contribute to cilia biogenesis and function. Mutations result in cilia that are faulty, contributing to the pleiotropic effects found in BBS. We provide an analysis of BBS, clinical findings, current knowledge of cilia biology, genetic counselling, current management, and a practical approach to diagnosis [1].

Incidence

The incidence of the syndrome is estimated to be 1 in every 160,000 people only 15 BBS cases have been documented in India. Disease severity is substantially greater in some groups with a greater extent of consanguinity or those who are geographically isolated, with disease prevalence of 1 in 13,000 in isolated Island and Kuwait communities, and 1 in 17,000 live births in Kuwait [2].

Patient identification

A male patient 24years from Borgaon, Wardha admitted to MICU, AVBRH on 30 December 2020 with a known case of Chronic kidney disease with Bardet Biedl syndrome. He is 65kg and his height is 152cm.

Present medical history

A male patient 24years old was brought to AVBRH on 30 December 2020 by his parents with a chief complaint of Breathlessness and pedal edema. He was admitted to MICU. He is a known case of BBS with Chronic kidney disease. After undergoing all the investigation such as CBC, KFT, ABG analysis USG abdomen & pelvis. He is diagnosed with chronic kidney disease on maintenance haemodialysis admitted to MICU after admission Central Venous line on Right Side attached to the patient on

5/1/2021 but after 8 days due to infection he is having fever so central venous line attached on left side. Fistula formation done on left upper limb from Nagpur Deshmukh hospital on date 28/12/2020. Two units of packed red cell transfused to the patient.

Past medical history

My patient was born with extra digits on his hands and was diagnosed with Bardet Biedl syndrome at birth, as well as obesity and fat build up along the abdomen.

Past intervention and outcome

In the past history of illness, he is having breathlessness, anxiety then his family referred him to a private hospital in Wardha. He was admitted there for 5days after that doctor referred him to AVBRH for further management. Then he was admitted on date 21/12/20 with the same chief complaint. After all investigations, he was diagnosed as chronic kidney disease on maintenance haemodialysis with known case of Laurance moon Bardet Biedl Syndrome. On admission, patient was in renal failure with uremic encephalopathy with Creatinine level 13 and urea level 256. ABG analysis Showed severe Metabolic acidosis. Dialysis was done for three consecutive days and patient improved clinically. KFT monitored regularly and urea creatinine value showed decreased trend. Patient was in uremic encephalopathy during admission and patient gradually improved clinically. One unit of PCR was transferred on 22/12/20. USG abdomen pelvis done on 23/12/20 suggestive Grade 3RPD dialysis with 1point PCR transfusion done on 26/12/20. Colour doppler of left upper limb suggested fit for fistula formation. Patient and relative were advised further hospital to stay but the patient and relative were not willing for the same and wanted discharge hence the patient was discharged.

Family history

There are four members in the family. Type of marriage of the parents is non – consanguineous marriage. All other members of the family were not having complaints in their health except for my patient and his brother both suffering from Bardet Biedl syndrome.

Clinical findings

Breathlessness from 2 days and pedal edema since15 days.

Etiology

CKD can be caused by a variety of factors. Polycystic kidney disease, for example, is a genetic cause of CKD. Lupus can cause glomerulonephritis. It can also arise because of a streptococcal infection, or because of a genetic disease [3].

BBS is caused by mutations in at least 14 distinct genes (often called BBS genes). These genes are known or hypothesised to perform significant functions in cell structures called cilia. Cilia are little projections that protrude from the surface of several different types of cells. They are involved in cell mobility and several chemical signalling pathways. Cilia are also required for the perception of sensory input (such as sight, hearing, and smell). The proteins generated by the BBS genes sustain and function Cilia.

When BBS genes are altered, it affects the physiology of Cilia. Defects in these cellular components are expected to alter important chemical signalling pathways throughout development, leading to sensory perception problems [4].

Physical examination

In the head-to-toe examination found that patient is having fat deposition along the abdomen extra digitalis present on upper extremities no abnormalities present other than these.

Diagnostic assessment

Blood test: Hb–8.2gm%, Total RBC count–2.82 millions/cumm, RDW–15.9%, HCT–24.4%, Total WBC count–8400/cumm, Granulocytes–68%, Lymphocytes–26%, AST(SGOT)–112 U/L. Peripheral Smear: RBCs-Predominantly normocytic mildly hypochromic with few microcytic RBCs seen. Platelets-Adequate on smear no Haemoparasite seen.

Kidney function test

Urea 88mg%, Creatinine- 4.2mg%, potassium- 5.4meq/l.

Liver function test

Alkaline Phosphate- 596IU/l, ALT- 75IU/L, Total Protein-5.2mg%, Albumin- 2.3g/dl, Total Bilirubin- 0.9mg%, Bilirubin Conjugated- 0.3g/dl, Bilirubin Unconjugated–0.3mg/dl.

Coagulation profile

Prothrombin Time-15.0sec, Magnesium-2.5mg/dl, Phosphorus-2.0mg/dl.

USG abdomen and pelvis

There is mild to moderate fluid collection in the peritoneal cavity,2D Echo- Mild left Ventricular hypertrophy present.

ECG

V1, V2, V3, V5, V6 abnormal 1 wave ST depression possible mild anteroseptal myocardial ischemia no completeness right bundle block longitudinal left axis deviation.

ABG analysis

Shows metabolic acidosis.

Therapeutic Intervention

Haemodialysis, Blood transfusion, AV fistula formation done on 28/12/2020 in left upper limb , Central venous line attached on right side on 15/1/2021 for administration of medications and fluid, Inj Meropenem 500mg x TDS, Inj Linezolid 600mg X BD, Tab Neurobion Forte X OD, Tab Nicardia X BD , Inj Augmentin 100mg X BD, Tab Sobosis 500mg X TDS.

DISCUSSION

A male patient of 24years old from Borgaon, Wardha was admitted to MICU, AVBRH on 30 December 2020 with a complaint of Breathlessness and Pedal edema and Hb%, Total Protein and Phosphorus less than normal limit Urea, Creatinine and Total Bilirubin in increased than normal limit . He is a patient of CKD on MHD with known case of Bardet Biedl Syndrome as soon as he was admitted to hospital investigations were done and appropriate treatment were started. After getting treatment, his condition improved, and the treatment was still going on till my last date of care.

Bardet and Biedl first described the many symptoms of BBS in the 1920s. Renal failure was only recently recognised as a component of the BBS clinical picture. Despite a high occurrence of post-mortem anatomical defects, renal problems in BBS had been rare. In one study, 26 out of 57 people (46%) had anatomical abnormalities in their kidneys. Pal and Bhattacharyya described an 18-year-old woman with hypogonadism, polydactyly, obesity, and mental disability, but no renal involvement. Cysts were discovered in a 30-year-old woman's left kidney, although her kidney function was normal. Gupta described a 20-year-old woman who had renal dysfunction and multiple fractures, possibly related to renal osteodystrophy. USG revealed bilateral hypoplastic kidneys and a blood creatinine level of 3.0 mg/dL. The BBS describes renal dysfunction at various rates. A series of questions review revealed anatomic malformations, renal failure, and ESRD, with anomalies such as renal cysts, foetal lobulation, dysplasia, unilateral agenesis, ectopia, calyceal clubbing or vesico-ureteric reflux blunting among the results Between 5% and 25% of BBS patients have renal dysfunction, with 4% to 10% of those going on to have (ESRD). Renal failure is the leading cause of death in BBS [5]. Future genotyping investigations could show genotype-phenotype associations, enabling for earlier detection and a better understanding of the pathophysiology of related kidney problems [6]. Several related cases and studies on chronic kidney diseases were reported [7-11].

INFORMED CONSENT

Before undertaking this case, the patients and their relatives were informed, and informed consent was acquired from both the patient and the relative.

CONCLUSION

Bardet–Biedl syndrome is a disease that requires early identification and treatment. To avoid further difficulties, cardiovascular and renal risk factors must be assessed and, if present, addressed early.

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