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Case report

Physiotherapy management in a rare *triallelic bardet- beidl* syndrome

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ABSTRACT

The Bardet–Biedl syndrome (BBS) is a rare ciliopathic human autosomal-recessive disorder, which is characterized principally by cardinal symptoms of marked central obesity, retinal dystrophy, polydactyly, mental retardation and hypogonadism and renal dysfunction.We report a case of 24 year old boy with Bardet-Biedl Syndrome, having classical symptoms like central obesity, hypogonadism, brachydactyly and strabismus. Laboratory investigations show reduced hemoglobin (6.1) and reduced WBC (700). Kidney function test (KFT) revealed an increased Urea level (130 mg/dl) and creatine (6.6mg/dl). Liver function test revealed (LFT) near normal bilirubin levels (1.4 mg/dl). CXR revealed subpleural fibrotic bands. Physiotherapy intervention included positioning, breathing re-training, chest PNF techniques, limb mobility exercises and HEP. The evidence from this study suggest that inpatient physiotherapy intervention plays a vital role in managing a patient with a rare case of Bardet-beidl syndrome. Early physiotherapy intervention has proven to be effective on this rare ciliopathic human autosomal-recessive disorder in ICU setup.

Keywords: Bardet-beidl syndrome, Rare autosomal recessive disorder, Inpatient rehabilitation.

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INTRODUCTION

The Bardet–Biedl syndrome (BBS) is a rare ciliopathic human autosomal-recessive disorder, which is characterized principally by cardinal symptoms of marked central obesity, retinal dystrophy, polydactyl, mental retardation and hypogonadism and renal dysfunction. Being a ciliary disorder, dysfunction and dysgenesis of cilia occurs [1]. Ciliated cells in mammals are colossally present in vertebral bodies. These cilia are primarily responsible for the mobility of cells and transfer of proteins and fluids across the epithelial cells. The frequency of the syndrome is estimated to be 1:1, 60,000 [2]. Twenty different genes (BBS1-BBS20) has been depicted. Mutation in these variable genes contributes to a wide variety of BBS presentation. The phenotype of BBS evolves very slowly in the first decade of life [3]. Less than 15 cases have been reported from India. Bardet–Biedl syndrome and Laurence Moon syndrome, both being rare conditions are often interchanged but they have now being proven to be two different integers with few similarities. Being an autosomal-recessive disorder, for a person to have the syndrome the gene which is responsible in each cell must have a mutation in both of its copies. At least 3 mutations within the gene is required for a person to exhibit the features of condition, which is also termed as triallelic inheritance [4]. Whereas the carriers of this recessive condition usually are unaffected and do not show any symptoms. When two carriers have children, 25% chances of them being affected are present. The complete spectrum of clinical features occurs in 40 to 45% of patients with the condition. Schachat and Maumenee have suggested that four of the five cardinal features have to be present to make the diagnosis. There is strong evidence from the literature that renal disease is also present [5]. Even though there is no cure for Bardet–Biedl syndrome, the usual approach of treatment is based on the signs and symptoms of the affected individual.

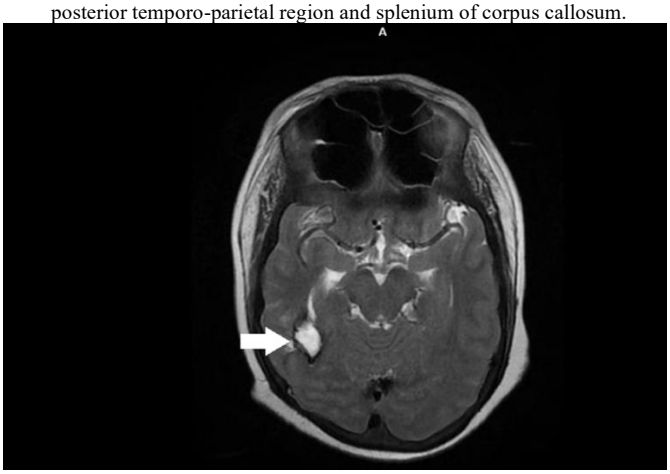
Patient information

A known case of Bardet Biedl Syndrome, 24-year-old male came to hospital for Dialysis as he was on maintenance hemodialysis (MHD) for chronic kidney disease. Presenting as a case of Bardet Biedl syndrome, central obesity, hypogonadism, brachydactyly and strabismus was positive. Patient was taken for routine MHD, where he experienced restlessness, pain in abdomen and giddiness so the MHD was stopped. The vital signs revealed tachycardia (125 bpm). The patient was shifted to ICU and in view of breathlessness he was taken

on 2 L of O2 via face mask. The patient complained of tingling and numbness in bilateral upper and lower limb along with pain in abdomen (epigastric region). Above mentioned complaints were treated with medical management. After the symptoms resolved patient was again taken for dialysis where he again underwent hypotension which resulted in cessation of dialysis. In view of tachycardia, tachypnea and desaturation at room air (SpO2-90%) which the patient presented on next day, he was taken on BiPap. The patient was then referred for in-patient rehabilitation.

On examination, Patient was conscious cooperative and well oriented to time place and person. Patient was on Bipap – IPAP: 10cm H20, EPAP: 5cm H2O. Vital signs were pulse rate was 112 bpm with regular rhythm and normal volume, respiratory rate was 25 breaths /min. Examination revealed chest excursion bilaterally decreased at all three levels, dull note over bilateral lower zones on percussion and reduced air entry bilaterally in lower zones on auscultation. Chest Xray – Subpleural fibrotic band in posterior basal segments. Laboratory investigations show reduced hemoglobin (6.1), reduced WBC (700). Kidney function test (KFT) relvealed an increased Urea level (130 mg/dl) and creatine (6.6mg/dl). Liver function test revealed (LFT) near normal bilirubin levels (1.4 mg/dl). Medications- Inj. Augmentin, Inj. Meropenem, Tab. Shelcal, Sobisis, Sevelamer, Neurobion Forte, Febuxostat, Inj. Neomol(if temp>101).

Figure 1: MRI brain showing two small intraparenchymal bleed in right posterior temporo-parietal region and splenium of corpus callosum.



ICU Management

The goal of physiotherapy in ICU was focused on improving the general condition and reduce the level of dependency in order to carry out ADL with minimum discomfort. Considering this, our goal was to wean of the patient from Bipap, relieve dyspnea, improve ventilation, and improve lung expansion and physical activity level. Patient and his relatives were educated about his present condition, explained about the advantages of physiotherapeutic intervention for better compliance and the extent of recovery and the expected time to the achieve the goal.

Positioning was given to prevent bed sores, deformities and improve ventilation in basal segments. Semi fowler's position was given thrice a day. Limb elevation for both upper and lower limb was given to prevent pooling of fluid in the extremities. Active limb mobility exercises were taught to maintain the overall integrity of the joints and functioning of muscles. Stretching of all major muscles was also given to maintain the flexibility. Chest physiotherapy included breathing exercises along with chest PNF technique. Diaphragmatic breathing and thoracic expansion exercises with upper limb coordinated movements were performed for breathing control along with improved chest expansion. Chest PNF was given to facilitate primary respiratory muscles by providing a proprioceptive stimulus. This leads to an overall improvement in their functioning and increase in the chest wall mobility. Relaxation of the chest wall muscles and an increase in diaphragm and abdominal muscle activity was also seen with PNF. Jacobson's relaxation technique was taught to induce relaxation.

The patient was weaned off from BiPAP after 3 days. The breathing exercises were continued and dyspnea relieving positions were taught. In view of the CXR findings, incentive spirometer was advised to perform every 2 hourly. He was progressed from in bed mobility to edge of the bed sitting position through active transfer techniques.

The patient and his relatives were counselled for home exercise program to maintain and progress the achieved functional recovery of respiratory and musculoskeletal system.

DISCUSSION

Kumar et al (2011) reported a case of classical phenotype of BBS where they reported that despite being a rare case, it was diagnosed because of the presence of hypogonadism and the absence of secondary sexual characters. It has been reported that BBS also presents with associated complains of heart disease, bowel disease, dental problems and behavioral disorders [6]. However there is no cure for this rare disease, obesity can be managed through diet, exercise and education, children reaching puberty can be screened for hormonal levels, early intervention, speech therapy and special treatment via speech therapy for intellectual disability [7]. The diagnosis of BBS is done on the basis of presenting complains of the patient like deep set eyes, flat nasal bridge, small mouth, malar hypoplasia etc. It has also been suggested that BBS is not diagnosed until visual disturbances are seen [8]. Molecular diagnosis is confirmed via direct sequencing of BBS genes. Billingsley et al recommended an outlook to mutational screening where sequencing is done with the frequency of pathogenic mutations in and between BBS genes. This was proven to be cost efficient.

The present reported case was admitted in ICU for routine MHD with all the major clinical features of Bardet-Beidl syndrome. In

the course of MHD, he underwent some complications for which he was admitted and referred to inpatient physiotherapy management. Despite of having a long list of comorbidities, physiotherapeutic intervention was proved to be beneficial in preventing the patient to be subjected to nosocomial infections.

The central obesity, a major clinical feature was not be targeted in our intervention due to unstable general condition of the patient. Further management for the same in the follow-up should be considered.

CONCLUSION

The ability to do ADLs in bed side sitting without assistance contributed to his compliance to exercise and provided an encouragement to the caregivers. Early physiotherapy intervention has proven to be effective on this rare ciliopathic human autosomal-recessive disorder in ICU setup. The present case report describes the need of ardency explore the field of physiotherapy and its effectiveness in early life-span of such atypical condition.

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