Ovarian Torsion in a patient with Laurence Moon Bardet - Biedl Syndrome

Jais Kumar*, Moiz Naeem and Sundas Naeem

Department of Nephrology, Akbar Niazi Teaching Hopsital and Islamabad Medical College, Pakistan

Received Date: March 01, 2022; Published Date: May 24, 2022

***Corresponding author:** Jais Kumar, Department of Nephrology, Pakistan Institute of Medical Sciences, Pakistan. E-mail: drkarmani@gmail.com

Citation: Jais Kumar, Moiz Naeem and Sundas Naeem. Ovarian Torsion in a patient with Laurence Moon Bardet Biedl Syndrome. J Clin Case Rep On. 2022;2(1):1016.

Abstract

Laurence-Moon-Bardet-Biedl condition (LMBBS), an uncommon autosomal recessive defect, generally happens in youngsters conceived from consanguineous relationships [1]. The significant highlights of this condition are cone-rod dystrophy, polydactyly, obesity, learning incapacities, hypogonadism in males, renal diseases, nystagmus, discourse issues, developmental delays, polyuria/polydipsia, ataxia, and poor coordination/ungainliness [2]. In this report, we present a case of a 12 year old female who presented with lower abdominal pain and burning micturition. She presented with central obesity, right sided scoliosis, polydactyly on hands, low intelligence quotient and retinal thinning on fundoscopy. A diagnosis of LMBBS was made.

Introduction

Laurence Moon Bardet - Biedl Syndrome (LMBBS) is a rare autosomal recessive disorder, caused by mutations in the PNPLA6 gene, seen most commonly in children born in consanguineous marriages [3]. These patients generally show symptoms in the first 10 years of life, poor night vision being the most common early manifestation [4]. The presence of four primary features on their own or three primary and two secondary features form the basis for making a clinical diagnosis [5].

Primary Features Include:

- Cone rod dystrophy/ Retinitis pigmentosa
- Polydactyly
- Obesity
- Learning disabilities
- Hypogonadism in males
- Renal anomalies

Secondary Features Comprise of:

- Speech disorders
- Brachydactyly
- Developmental delay
- Polyuria/ Polydipsia
- Ataxia
- Poor coordination/ clumsiness
- Diabetes mellitus
- Left ventricular hypertrophy
- Hepatic fibrosis
- Hearing loss

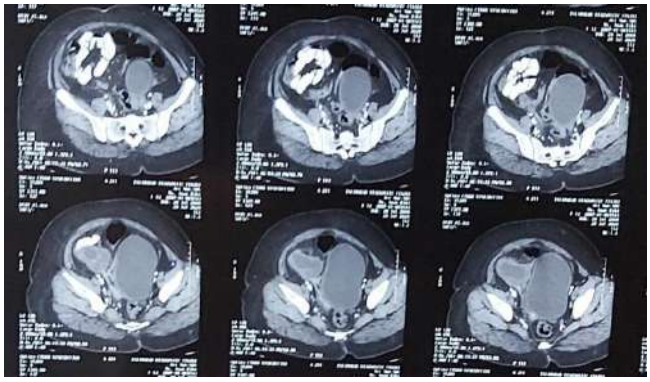
Case Presentation

A 12 year old female presented to Nephrology outpatient department with complains of lower abdominal pain which radiated to the back, burning micturition and polyuria. She was in her usual state of health 4 days earlier. Upon further inquiry, her back pain began initially 2 years to 3 years ago, which was usually relieved by taking painkillers but increased in severity in the past 3 days and was not affected by painkillers. Her burning micturition began 1 day before along with the polyuria during the night. Her attendant explained that she passed urine every 1 hour to 2 hours. She had a past surgical history of appendectomy 4 years prior which was uncomplicated.

She was admitted in the nephrology department, and examination showed short stature, morbid obesity, right sided kyphoscoliosis and polydactyly on both hands. She possessed low intellectual capabilityand had not yet reached menarche yet.She wore glasses as she was a myope from a young age. Further supporting the diagnosis of Laurence Moon Bardet Biedl syndrome, was the fact that two of her siblings, one younger brother and one older sister, possessed the same features, which shows strong family history.

An ultrasound abdomen and pelvis which revealed enlarged liver with increased parenchymal echogenicity, bilateral dilation of pelvicalcyeal system in kidneys and a prepubertal uterus. Her right iliac fossa was tender and ultrasound revealed a thick walled cystic structure with thin septations and right sided ovary could not be visualized along with fluid in the right half of pelvis. Diagnosis of ovarian torsion was made and further CT scan of abdomen pelvis with contrast was advised.

The CT scan revealed right ovary was enlarged, measuring 6.5 x 5.1 cm with thickened walls and strand changes in its surroundings further strengthening diagnosis of ovarian torsion.



Her full blood count was done which showed WBC count of 11380/$mm^{3,}$ Hemoglobin of 11.5 g/dl, with neutrophils and lymphocytes within normal range. Her urine routine examination was done due to burning micturition, which revealed leucocyte esterase +++, WBCs 35 to 40, and epithelial cells 8 to 10, which showed the presence of lower urinary tract infection.

Investigation into the affected siblings revealed a 16 year old female and 10 year old male, both morbidly obese with poor vision and low intelligence quotient. Eye examination showed nystagmus in both eyes and subsequent fundoscopy of both showed retinal thinning with cone rod dystrophy, tilted discs and tessellated fundi. PTA showed conductive hearing loss. The sister's ultrasound showed grade 1 fatty liver, splenomegaly with a well-defined lesion at the hilum, bilateral dilation of pelvicalyceal system of kidneys and thickened urinary bladder. The brother's ultrasound showed fatty liver, thickened urinary bladder, and bilaterally dilated pelvicalyceal system of kidneys.

Discussion

This report primarily focuses on the rarity that is ovarian torsion in a patient of Laurence Moon Biedl Bardet Syndrome. As far as we know, the only incidence of ovarian related anomalies in patients with LMBBS is a case of ovarian teratoma in a 20 year old female in Romania [6]. It is rare since mostly findings related to reproductive system in LMBBS are in males who commonly present with hypogonadism [7]. This point to the scarcity of information we possess currently about this condition, especially in a country like Pakistan where consanguineous marriages are common [8,9].

Conclusion

The treatment of LMBBS is directed towards managing the manifestations of the disease. The renal abnormalities, which are a cardinal feature of LMBBS, progressed to end stage renal disease eventually, especially in the third and fourth decade of life. Due to this significant risk, good blood pressure control and routine testing should be emphasized. A strict diet consisting of nutritious and well balanced meals along with regular exercise is recommended. Regular ophthalmologist visits and glasses are recommended for the poor vision. Hormone replacement therapy can be done to manage the poor functional capacity of the anterior pituitary. Because of its multisystem involvement, it can often easily be missed, and a thorough management plan would allow those affected to mix into society better and earlier.

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