International Journal of Advances in Medicine
Kaluvai SI et al. Int J Adv Med. 2023 Jan;10(1):76-78
http://www.ijmedicine.com
pISSN 2349-3925 | eISSN 2349-3933
DOI: https://dx.doi.org/10.18203/2349-3933.ijam20223302

Case Report

A case of Laurence Moon Bardet Biedl syndrome

Sai Indraneel Kaluvai, Rajalakshmi K. V.*, Anantha Kumar P. K., Bubhaneshwar N.

Department of General medicine, Saveetha Medical College, Chennai, Tamil Nadu, India

Received: 25 November 2022
Accepted: 10 December 2022

***Correspondence:**
Dr. Rajalakshmi K. V.,
E-mail: drrajeesakthi@gmail.com

ABSTRACT

Laurence Moon Bardet Biedl syndrome (LMBBS) is a rare autosomal recessive disorder characterized by retinitis pigmentosa, obesity, hypogonadism, mental retardation and polydactyly. We present a case of 20-year-old male with history of intellectual delay, speech impairment and progressive vision loss. Presented with chief complaints of Breathlessness, oliguria, abdominal distension. Upon examination he is morbidly obese, short stature, pedal edema, facial puffiness, polydactyly of feet, micro penis and retinitis pigmentosa. He presented with primary pentad features of LMBBS with CKD stage 5 for which he was started on hemodialysis. He also had secondary features of speech delay, developmental delay, dental crowding, high arched palate and ventricular hypertrophy. Management of LMBBS is supportive. genetic counselling of affected families raises awareness about need to get the other family members assessed for renal and cardiovascular problems.

Keywords: LMBBS, Polydactyly, Retinitis pigmentosa, Mental retardation

INTRODUCTION

Laurence Moon Bardet Biedl syndrome (LMBBS) is a rare autosomal recessive disorder characterized by retinitis pigmentosa, obesity, hypogonadism, mental retardation and polydactyly.[1,2] It is commonly found in families with inter family marriages. The prevalence of LMBBS is estimated to be 1: 1,60,000.[3] Only a few cases were reported in India, so we are presenting a case case of classical LMBBS with chronic kidney disease.

CASE REPORT

A 20-year-old male, who is morbidly obese [BMI=41] with history of intellectual delay, speech impairment and progressive vision loss. Presented with chief complaints of Breathlessness, oliguria, abdominal distension since 15 days. Upon examination he is morbidly obese, lethargic, lying down on bed and he is short statured, crowded teeth present, pedal edema present, facial puffiness present, with polydactyly of feet and micro penis. Opthalmological examination revealed retinitis pigmentosa. He is mentally retarded. Family history revealed that mother and father had consanguineous marriage.

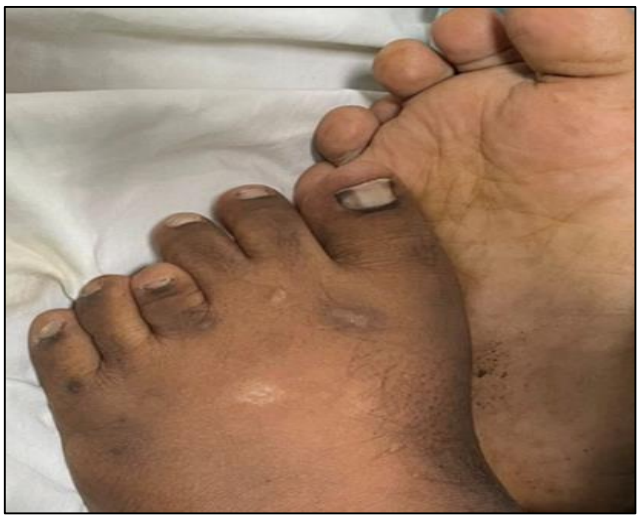

Figure 1: Polydactyly of right and left foot.

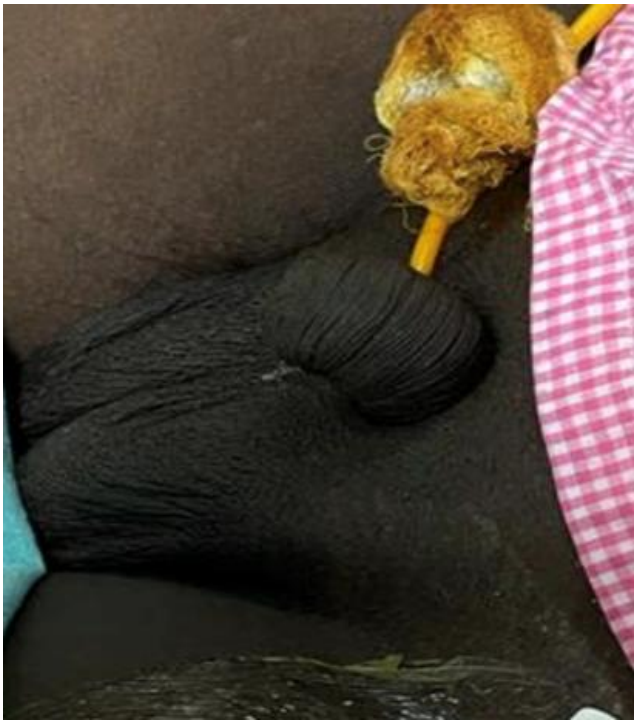

Figure 2: Micro-penis

His mother had no history of consumption of any drugs during pregnancy. He was born by normal vaginal delivery and he was born by full term. According to the patient mother he had delayed mental and physical growth. He started walking at the age of 5 years and speech was started at the age of 6 years. Laboratory investigations revealed hemoglobin of 6.9%, serum creatinine is 12 mg/dl, serum urea is 217 mg/dl, potassium of 7.7 meq/L, serum bicarbonate of 8.3 meq/L. 2D Echo was which showed eccentric left ventricular hypertrophy. CT abdomen revealed hepatomegaly, right contracted kidney with renal pelvic lipomatosis, left enlarged kidney with hydroureteronephrosis grade 4.

DISCUSSION

In 1886 Laurence and Moon first described a case with mental retardation, hypogenitalia, polydactyly, obesity and rod-cone dystrophy. Then in 1920 Bardet described a case with polydactyly, obesity and rod-cone dystrophy.[4,5] Later Biedl explained about two cases with clinical findings of mental retardation, skull abnormalities and gastrointestinal problems. Later years, termed as LMBBS.

LMBBS has pentad of features retinitis pigmentosa, obesity, hypogonadism, mental retardation and polydactyly.[6] Secondary features include renal impairment, speech delay, congenital heart diseases, eye abnormalities and diabetes mellitus. Our patient had pentad of primary features with secondary features like speech delay, developmental, chronic kidney disease stage 4 and concentric left ventricular hypertrophy.

He had a seizure episode during the course of hospital and for which antiepileptics was started. He had derangement of renal function tests and decreased urine output which prompted the need for nephrologist referral and started on hemodialysis. Genetic evaluation was not done as they are not financially affordable and due to unavailability of genetic testing in our centre.

Management of LMBBS is supportive. Management of chronic kidney disease by haemodialysis or renal replacement.[7] Hormonal replacement with testosterone with low hormones. Low calorie diet for treatment of obesity. Regular screening for hypertension, diabetes, cardiac and renal involvement.[8] Rehabilitation for mentally retarded and vision loss patients. Consanguineous marriages are associated with LMBBS, so awareness of consequences of consanguineous marriages should be done.

CONCLUSION

LMBBS is a disorder with identified pentad of symptoms which are obesity, hypogonadism, polydactyly, intellectual impairment and retinitis pigmentosa. Renal function loss is most common cause of mortality in these patients. Because of these seemingly unrelated symptoms this disorder remains largely under diagnosed. Most the reported cases in LMBBS are associated with consanguineous marriages, so the affected families should undergo genetic counselling. And also, genetic counselling of affected families raises awareness about need to get the other family members assessed for renal and cardiovascular problems.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Green JS, Parfrey PS, Harnett JD. The cardinal manifestations of Bardet-Biedl syndrome, a form of Laurence-Moon-Biedl syndrome. N Engl J Med. 1989;321:1002-9.
2. Azizul MH, Sharmin LS, Tarikul QI, ARM Safiuddin E. Bardet Biedl syndrome-A Case Report. J Teachers Association RMC Rajashahi. 2007;20(1):56-9.
3. Sahu JK, Jain V. Laurence-Moon-Bardet-Biedl Syndrome. J Nepal Med Assoc. 2008;47(172):235-7.
4. Khan PA, Nishaat J, Noor S, Fatima N. Laurence Moon Bardet Biedl Syndrome: a rare case report in a tertiary care teaching hospital, Hyderabad, Telangana, India. Int J Med Sci Public Health. 2017;7:68-71.
5. Rajasekhar P, Parmi MK, Aalekhya PS. Laurence Moon Bardet Biedl Syndrome. Int J Sci Res. 2013;6(14):2319.
6. Forsyth RL, Gunay-Aygun M. GeneReviews®. Seattle: University of Washington; Bardet-Biedl syndrome overview. 2003.

7. Bryan W, David J, John W. Chronic renal failure; an important feature of the Laurence-Moon-Biedl syndrome. Postgraduate Med J. 1988;65:462-4.
8. Khan OA, Majeed R, Saad M, Khan A, Ghassan A. Rarity of Laurence Moon Bardet Biedl Syndrome and its poor management in the Pakistani population. Cureus. 2019;11:0.

Cite this article as: Kaluvai SI, Rajalakshmi KV, Kumar APK, Bubhaneshwar N. A case of Laurence Moon Bardet Biedl syndrome. Int J Adv Med 2023;10:76-8.