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Lawrence Moon Bardet Biedl syndrome with Chronic kidney Disease

Shravani Rama
Vijay Kollipara

Abstract:
Laurence-Moon-Bardet-Biedl syndrome is a rare ciliopathic autosomal recessive disorder mostly seen in children of consanguineous marriages. It is identified by its characteristic features namely retinitis pigmentosa, obesity, mental retardation and polydactyly.
Herein, we report a case of a 24 year old female presenting with features of renal dysfunction like anemia, decreased urine output, vit D3 deficiency and further examination was found to show features of LMBBS i.e retinitis pigmentosa, learning disabilities, polydactyly and obesity. It is important for the physicians to be aware of this syndrome for early identification. Multidisciplinary approach and periodic follow up can decrease the morbidity and mortality of the patient.

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I. Background

Laurence-Moon-Bardet-Biedl syndrome (LMBBS) is a rare ciliopathic autosomal recessive disorder mostly seen in children born out of consanguineous marriage which is identified by its characteristic features namely, retinitis pigmentosa, obesity, mental retardation and polydactyly(1). However, it can manifest in many different ways in different individuals and also in different individuals belonging to the same family(2).

Laurence moon syndrome and bardet biedl syndrome are two different entities which are considered as one as it is difficult to accurately differentiate them. LMS predominantly presents with loss of vision, mental retardation and features of hypogonadism whereas, BBS mostly presents with obesity and polydactyly and congenital cardiac anomalies(3).

It has an incidence of 1/100,000 cases with slight female predominance. The most common cause of mortality in them being renal dysfunction(1,3).

II. Case Report

A 24 year old female patient presented with chief complaints of generalized edema with periorbital puffiness and shortness of breath for 10 days and decreased urine output for 3 days.There was a history of progressive loss of vision since she was 12 years of age which started as night blindness and progressed to the present stage which is only light perception which was confirmed as retinitis pigmentosa by an opthamologist 4 years ago. She was born out of a consanguineous marriage at term and showed developmental delay. She is immunized up to date. She attained menarche at 13 years of age and has regular cycles. She has a sibling of age 19 years who does not have any similar complaints and his developmental milestones were up to mark.

On general examination, the patient was conscious, coherent, co-operative with a short stature, she is moderately built and has central obesity. She has moon facies and widely spaced eyes along with polydactyly on both the lower limbs and her fingers showed brachydactyly. There is absence of axillary hair along with non-pruritic hyperpigmentation of hands. No underdevelopment of gential organs was seen. Abdomen was distended and showed hyperpigmentation with no scars. There was no tenderness on palpation. She had learning disabilities and diminished vision, due to which she discontinued school after 5th grade

She had a history of repeated hospital admissions since childhood. Recently, she was admitted with severe cramps in the lower limbs and lower back since 1 month ago for which she was treated with vit D3 supplementation. She had a repeated history of loose stools and puffiness of face. Due to severe anemia and frequent cramps, along with abnormal RFT, she was diagnosed with chronic kidney disease associated with metabolic acidosis 2 months ago. Known case of hypothyroidism for 12 years.

Her vitals were normal and upon examination, pallor was noticed. On CNS examination, higher mental functions were intact. Normal bulk on all the limbs. There was 4/5 power in both lower limbs. Reflexes were slightly diminished, +1 in all the limbs. There was a sluggish pupillary reflex.

Investigations revealed Hb% to be 3.6g%, reticulocyte count 5%, RBC- anisopoikilocytosis, normocytic, hypochromic cells, blood urea 317 mg/dl, serum creatinine 14.3 mg/dl, sodium 139 mEq/l, potassium 6.2 mEq/l, chloride 116 mEq/l, pco2 24.2 mmol/l, total serum proteins 7.94 gm%, serum albumin 2.40 gm%, alkaline phosphatase 106 KAU/100ml

Gene mapping was not done due to the economical constraints of the patient's family

She was given 7 blood transfusions owing to her severe anemia, and was put on vitamin D3 and calcium supplementations and was on hemodialysis treatment for thrice a week.

Discussion

The patient presented to us is a typical case of LMBBS syndrome presented with low urine output. The genetic techniques available now have led to early identification of this disease which is associated with 19 different genes and most of them are proteins coded for clilia(4) Consanguinity being a common practice in India, is one of the most influencing factors for the growing number of cases.

The first prime feature is retinal dystrophy, found in the first decade of life while obesity begins in childhood and progresses with age.

The primary features of LMBBS are Cone-Rod Dystrophy, Polydactyly, Obesity, Learning Disabilities, Hypogonadism in males & Renal Anomalies.

The secondary features are Speech disorders, Brachydactyly, Developmental disorders, Polyuria/Polydipsia, Ataxia, Diabetes, LVH, Hepatic fibrosis, Spasticity, Hearing loss, Short stature, Crowding of teeth, Hypermobile joints, and Early osteoarthritis.The presence of 4 primary features or 3 primary coupled with 2 secondary features are clinical grounds for making the diagnosis (5). However, the full spectrum of disease is seen only in 40-50% of the patients.

The average age for diagnosis of the disease is 9 years with the longest being 50 years. Our patient was 20 years old when diagnosed.

Although their childhood passes by smoothly, puberty can be the most stressful period for the patients and hence, the family must be given guidance by an experienced counselor. The additional digits are non-functional and can be excised for cosmetic purposes. Obesity is a major concern as it can lead to various metabolic disorders. Most of the risk factors of obesity such as insulin resistance, increased coagulability, increased oxidative stress, and impaired RAAS activity are a main cause of early onset CKD(6). The patient and their family are advised beforehand regarding healthy dietary and lifestyle practices that involve less carbohydrates, less fat, and frequent exercises which will delay the disease progression(7).

LMBBS patients are usually friendly people. They require frequent attention of medical personnel especially, nephrologists, opthamologist, cardiologists to have a regular update of their medical condition which could help improve their quality of life. Yearly renal function tests, lipid profile, ophthalmological assessment, and frequent BP checks are necessary to predict complications(8).

Chronic kidney disease is managed by periodic dialysis and the option of renal transplantation should be put forward. Early screening of the patient for renal impairment, hypertension, and diabetes is needed. All the modalities of renal replacement therapy, i.e. chronic peritoneal dialysis, renal transplantation and hemodialysis are offered to these patients(9).

III. Conclusion

LMBBS is a condition that primarily presents with a pentad of symptoms like retinitis pigmentosa, obesity, polydactyly, hypogonadism and intellectual impairment. It is really difficult to diagnose the disease at an early stage due to the presence of seemingly unrelated symptoms. The disease progression leads to renal impairment which is the most common cause of mortality in these patients. As it is an autosomal disorder, children born out of a consanguineous marriage are at more risk of developing the disease. Hence, genetic counseling to the parents of consanguineous marriage might prevent conceiving of such babies and is of utmost importance.

Despite the presence of risk factors i.e. consanguineous marriages, there are not many cases reported in the hospitals. This is due to the lack of awareness about the disease and the presenting features. This has to be addressed along with the other different presentations of the syndrome that can replenish the medical literature.

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