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Laurence-Moon-Bardet-Biedl Syndrome Coexisting with Chronic Liver Disease: A Rare Manifestation

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Abstract: Laurence-Moon-Bardet-Biedl Syndrome (LMBBS) is a rare autosomal recessive disorder characterized by multisystem involvement, primarily affecting vision, cognition, endocrine, renal, and skeletal systems. Here, we present a case of a 29-year-old male diagnosed with LMBBS who also exhibited features of chronic liver disease (CLD), an association not commonly reported. This case highlights the importance of a multidisciplinary approach for managing LMBBS patients and the potential implications of hepatic involvement.

Keywords: *Laurence-Moon-Bardet-Biedl Syndrome, Chronic Liver Disease, Hepatic Fibrosis, Autosomal Recessive, Multisystem Disorder.*

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I. INTRODUCTION

Laurence-Moon-Bardet-Biedl Syndrome (LMBBS) is a rare genetic disorder classified under ciliopathies, a group of diseases caused by mutations in genes affecting primary ciliary function. Cilia are crucial organelles involved in signaling pathways, development, and cellular maintenance, and their dysfunction leads to a wide spectrum of clinical manifestations.

LMBBS is characterized by six primary features: retinitis pigmentosa, obesity, polydactyly, intellectual disability, hypogonadism, and renal dysfunction. The syndrome also presents with several secondary features, including diabetes mellitus, hepatic involvement, and speech impairment. Renal complications significantly contribute to mortality in these patients. Although hepatic involvement is not a classical feature of LMBBS, some reports indicate an association with hepatic fibrosis, metabolic dysfunction, and non-alcoholic fatty liver disease (NAFLD).

In this case, we present a patient with clinically confirmed LMBBS and chronic liver disease, expanding the phenotype of this syndrome and underlining the necessity for multidisciplinary evaluation and long-term follow-up.

II. CASE PRESENTATION

A 29-year-old male, born to consanguineous parents, presented to the Gastroenterology outpatient department at KLES Dr. Prabhakar Kore Hospital & MRC, Belagavi, with progressive abdominal distension over 1.5 months and dyspnea on exertion. The patient denied symptoms of abdominal pain, vomiting, constipation, or obstipation.

➢ *Developmental and Medical History:*

- Developmental delay and intellectual impairment noted since early childhood.
- Visual impairment with night blindness since 8 years of age, later diagnosed as retinitis pigmentosa.
- Speech impairment and articulation difficulties since childhood.
- Increased appetite, obesity, and reduced mobility, requiring assistance for daily activities.

➢ *Family History:*

- Born from a consanguineous marriage (first-degree cousins).
- One healthy sibling with no known illness.

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➢ *Physical Examination:*

- Obese physique with morbid obesity (BMI >30 kg/m²).
- Low IQ with cognitive impairment.
- Vital signs: BP: 110/90 mmHg, PR: 78 bpm, RR: 16 breaths/min, GCS: 15/15.

➢ *Systemic Examination:*

- Endocrine Features: Bilateral atrophic testes, gynecomastia, and delayed secondary sexual characteristics.
- Skeletal Abnormalities: Polydactyly (right leg), brachydactyly, and mild scoliosis.
- Abdominal Examination: Distended abdomen with positive fluid thrill, suggestive of ascites.
- Other Findings: Periorbital puffiness, pedal edema, leukonychia (indicating chronic illness and protein deficiency).

➢ *Investigations*

Table 1: Comparsion of investigations of patient

Investigation	Result
Hemoglobin	10 g/dL
Platelet Count	2,00,000/µL
Serum FSH	47.64 IU/L
Serum Prolactin	27.72 ng/dL
Serum Testosterone	10.93 ng/dL
Liver Function Tests	Elevated liver enzymes, hypoalbuminemia
Abdominal Ultrasound	Ascites with features suggestive of chronic liver disease
Echocardiography	Normal cardiac function
Renal Function Tests	Mildly deranged renal function
Genetic Testing	Not performed due to financial constraints but clinically diagnostic

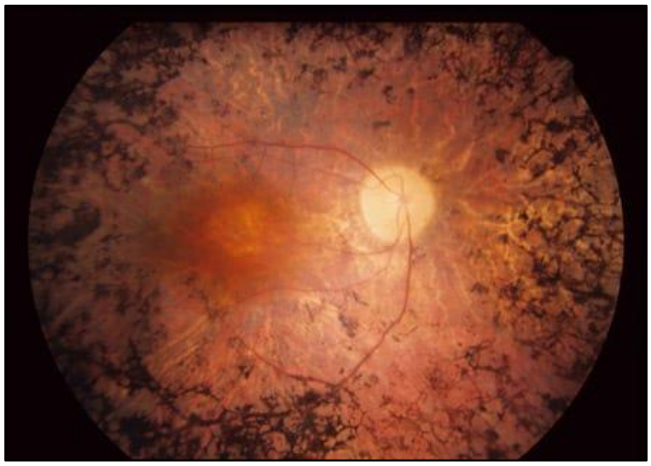

Fig 1: Retinitis Pigmentosa

Fig 2: Moon Like Face

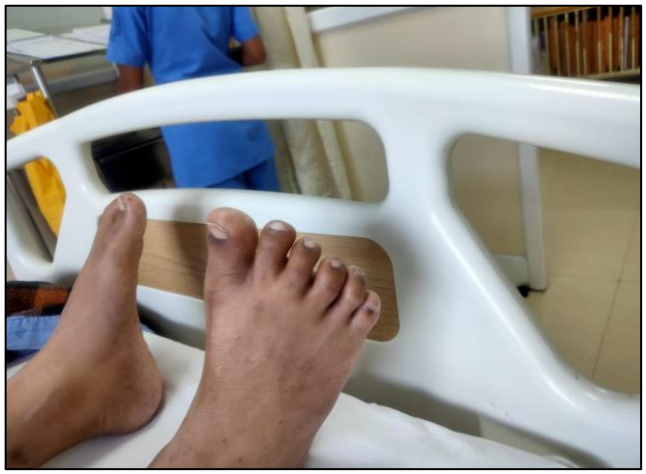

Fig 3: Polydactyly

III. MANAGEMENT

➢ *Multidisciplinary Care:*

- Ophthalmology: Monitoring retinitis pigmentosa, low-vision aids.
- Endocrinology: Hormonal therapy for hypogonadism if required.
- Nephrology: Renal function monitoring, early intervention for renal impairment.
- Gastroenterology: Ascites management with diuretics (spironolactone, furosemide) and sodium restriction.
- Nutritional & Metabolic Management: Dietary modifications to prevent obesity and metabolic complications.

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➢ *Genetic Counseling:*

- Educating affected families about recurrence risks and genetic inheritance.
- Early genetic screening for at-risk siblings.

➢ *Long-Term Monitoring:*

- Routine liver, renal, and endocrine function tests.
- Regular imaging to assess liver fibrosis progression.

IV. DISCUSSION

LMBBS is a genetically heterogeneous disorder associated with ciliary dysfunction, which is crucial for cell signaling and development. The diagnosis is based on primary and secondary criteria, and our patient met all six primary features and multiple secondary features, including hepatic involvement.

Although hepatic fibrosis and NAFLD are not widely recognized components of LMBBS, growing evidence suggests their presence in patients with metabolic dysfunction. The pathophysiology involves:

- Ciliary Dysfunction: Mutations in genes encoding ciliary proteins disrupt hepatic architecture, promoting fibrosis and liver dysfunction (Zaghloul et al., 2009).
- Metabolic Dysregulation: Obesity, insulin resistance, and dyslipidemia contribute to NAFLD, which can progress to cirrhosis (Forsythe & Beales, 2013).
- Renal-Hepatic Crosstalk: Shared molecular pathways in renal and hepatic fibrosis suggest a systemic involvement in ciliopathies (Beales et al., 1999).

Given its autosomal recessive inheritance, consanguinity is a significant risk factor for LMBBS, reinforcing the importance of genetic counseling.

V. CONCLUSION

This case highlights a rare association of LMBBS with chronic liver disease, emphasizing the need for early recognition and comprehensive management. Multidisciplinary coordination is crucial for improving the quality of life in these patients. Genetic counseling should be prioritized in high-risk families to prevent disease recurrence.

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