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Case Report

Bardet-Biedl Syndrome: A Case Report

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Abstract:

Bardet Biedl syndrome is an autosomal recessive ciliopathy. The primary clinical features of BBS include rod-cone dystrophy, polydactyly or dystrophic extremities (brachydactyly and syndactyly), obesity, reduced intelligence, renal dysfunction, and male hypogonadism that manifests in the first decade of life with polydactyly as a congenital feature. The other commonly associated secondary features include hepatic fibrosis, endocrinological disturbances such as diabetes mellitus, hypercholesterolemia, and reproductive abnormalities, short stature, speech defects, and developmental delay. We report a 29-year-old male patient presenting with classical features of BBS with significant similar history in siblings. we confirm the diagnosis on basis of clinical criteria. Genetic testing could not be done due to limited resources. There is no definite treatment. Early diagnosis and symptomatic, supportive and rehabilitative measures can reduce the disability. This includes dietary modification, oral hypoglycaemic drugs, testosterone supplement etc. Relatives of the patient should be screened for renal abnormality.

Keywords: Bardet-Biedl Syndrome, retinitis pigmentosa, hypogonadism

Introduction

Bardet-Biedl Syndrome (BBS) is a rare, autosomal recessive inherited disorder categorized as a model of non-motile ciliopathy. It exhibits significant clinical variability both within and between families. BBS is closely related to Laurence-Moon Syndrome, sharing features such as retinitis pigmentosa, intellectual disability, hypogonadism and spastic paresis (1). The core manifestations of BBS include retinitis pigmentosa, obesity and postaxial polydactyly. This case highlights a patient presenting with early-onset blindness, key ocular features, and most of the general clinical manifestations of BBS along with a suggestive family history (2).

Patient Presentation:

A 29-year-old male, the first child in a non-consanguineous marriage, presented to the medicine outpatient department with complaints of lower limb weakness and fever for the past four days. The patient reported a history of chronic cough with mucoid expectoration since childhood but had no known history of diabetes, hypertension, tuberculosis, or renal dysfunction. There was a family history of similar presentations in siblings, while both parents were unaffected.

On examination, the patient was afebrile and vitally stable. He exhibited:

- Short stature, Childhood-onset blindness
- Obesity (BMI: 40 kg/m², WHR >0.9)
- Postaxial polydactyly of the right hand
- Abdominal striae
- Hypogonadism
- Normal mental status
- Ataxic gait
- Spastic paraparesis with distal weakness
- Respiratory findings: crepitations in the right middle and lower lung lobes

Ophthalmologic examination revealed severe vision loss with bilateral pale optic discs, attenuated vessels, and bony spicules, confirming retinitis pigmentosa.

Based on the clinical presentation, the patient fulfilled the diagnostic criteria for BBS, having three primary features as shown in images and two secondary features including Diabetes mellitus and Mild spasticity.

Genetic testing was not feasible due to resource limitations.

Investigations:

Haematology:

HB:11.1g/dl, TLC-3.3k/ul , platlets-1,54,000/ml

MCV: 89 fl, MCH: 25 pg, MCHC: 29.3 g/dL

Peripheral smear: normocytic normochromic RBCs, reduced WBCs and nothing significant on peripheral smear.

Liver Function Tests:

- Total bilirubin: 1.1 mg/dL (Direct: 0.90 mg/dL)
- SGOT: 164 IU/L, SGPT: 57 IU/L
- Renal Function Tests:
- Urine Routine/Microscopy: Albumin ++
- Serum creatinine: 1.1 mg/dL, Blood urea: 23 mg/dL
- Serum Electrolytes in normal range.

Metabolic Panel:

- Blood sugar: 83 mg/dL, HbA1c: 6.3%
- Lipid profile in normal range.

Serology:

- Hepatitis B and C: Negative

ECG: Sinus rhythm

Ultrasound Abdomen:

- Bilateral kidneys: Normal size
- Mild splenomegaly, normal liver size

Fundoscopy:

- Bilateral pale optic discs, attenuated vessels, bony spicules
- Suggestive of bilateral retinitis pigmentosa

CT Thorax:

- Traction bronchiectasis in the right upper and middle lobes
- Cystic bronchiectasis in the right lower lobe.

Lumbar Puncture: Normal cellularity and protein levels

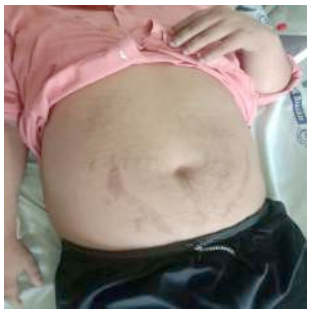
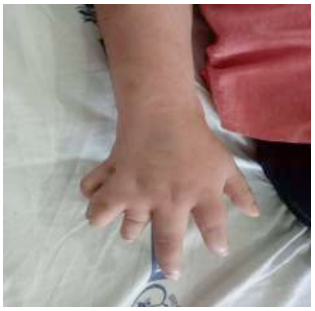


Figure: 1a.Truncal Obesity



1b. Polydactyly

1c. Hypogonadism

1d. Moon shaped Face

Primary clinical features in Bardet- Biedl Syndrome 1a) Truncal obesity 1b) Polydactyly in hands 1c) Hypogonadism. 1d) Moon shaped face

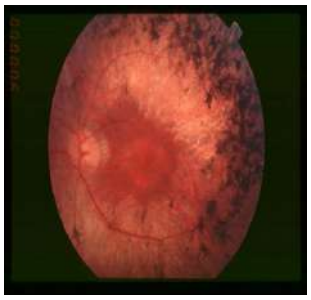
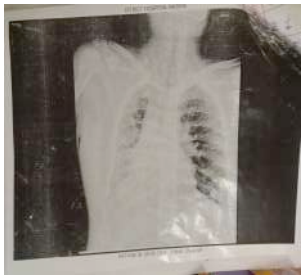


Figure : 2a. Retinitis Pigmentosa



2b. Chest X-RAY, PA view

Other primary clinical features in Bardet- Biedl Syndrome: 2a) Retinitis Pigmentosa 2b) Chest X-Ray PA view showing tractional bronchiectasis.

3a 3b 3c 3d

Figure: Sibling photographs showing same presentation in the family 3a-b Female sibling 3a. Polydactyly 3b. Moon shaped face 3c-d Male sibling 3c. Polydactyly 3d. Moon shaped face

Discussion:

Bardet-Biedl Syndrome was first described in 1922 as an autosomal recessive disorder affecting multiple organ systems due to defects in ciliary structure and function (2). The cardinal features include polydactyly, pigmentary retinopathy, obesity, intellectual disability, and hypogonadism (3). Genetic mutations in BBS1 to BBS18

account for approximately 70%-80% of cases. Dysfunction in these BBS genes leads to defects in ciliary-related proteins, classifying BBS as a ciliopathy disorder (3).

Conclusion:

Currently, there is no definitive treatment for BBS-associated retinal degeneration, and vision loss is progressive. However, research by Simons et al. has demonstrated the potential of gene therapy using adeno-associated virus-mediated BBS4 delivery in a BBS4-null mouse model to correct rhodopsin mislocalization (4). Given the genetic basis of BBS, genetic counselling plays a crucial role in patient management, providing affected families with essential information on inheritance patterns and potential therapeutic interventions (5).

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