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Bardet-Biedl Syndrome (BBS) and Renal Failure: A Report of Two Cases

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Abstract **Case Report**

Bardet-Biedl syndrome (BBS) is an autosomal recessive genetic disorder characterized by intellectual disability and multi-organ involvement, particularly renal impairment, which is a major determinant of prognosis. We report two cases of BBS managed in our department due to renal insufficiency. Case 1 involves a 17-year-old female with facio-truncal obesity, retinitis pigmentosa, and learning difficulties, who developed end-stage renal failure over two years and is currently undergoing pre-transplantation evaluation. Case 2 describes a 7-year-old male with obesity, hexadactyly, cryptorchidism, and retinitis pigmentosa, diagnosed with BBS based on five major and two minor criteria, showing moderate renal insufficiency. Diagnosis of BBS is based on the presence of major and minor criteria, and it is genetically heterogeneous, with mutations in 12 genes responsible for the function of primary cilia. Renal involvement is the primary determinant of prognosis, and early detection through molecular diagnosis and genetic counseling is crucial. Consanguinity is a key risk factor for BBS, and renal function should be closely monitored in affected individuals.

Keywords: Bardet-Biedl syndrome, autosomal recessive, molecular diagnosis, renal failure, intellectual disability, facio-truncal obesity, retinitis pigmentosa.

INTRODUCTION

Bardet-Biedl syndrome (BBS) is an autosomal recessive genetic disorder characterized by intellectual disability and multi-organ involvement. Renal impairment can occur at variable ages and is typically a major determinant of prognosis. We report two cases of this syndrome managed in our department due to renal insufficiency.

Observation No. 1

The patient is a 17-year-old female (O.B.), from a first-degree consanguineous marriage, followed since the age of 8 for BBS based on the presence of three major criteria: facio-truncal obesity (BMI: 31 kg/m²), retinitis pigmentosa associated with night blindness, and learning difficulties, alongside two minor criteria: psychomotor developmental delay with delayed speech acquisition and phonetic disorders. Laboratory tests revealed dyslipidemia and moderate renal insufficiency (creatinine clearance = 50 ml/min). Renal ultrasound showed two kidneys of normal size but moderately differentiated. Over a two-year period, her condition progressed to end-stage renal failure requiring permanent hemodialysis. The patient is currently admitted for pre-transplantation assessment.

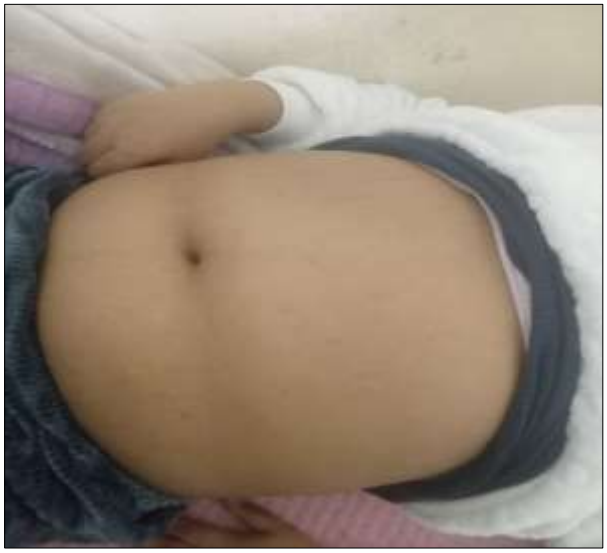

Fig.1

Observation No. 2

The patient is a 7-year-old male (A.B.), from a first-degree consanguineous marriage, who has presented since birth with obesity, hexadactyly, cryptorchidism with micropenis, learning difficulties,

delayed speech acquisition, and psychomotor developmental delay. Ophthalmological examination revealed retinitis pigmentosa (both clinically and electrophysiologically). The clinical diagnosis of Bardet-Biedl syndrome was established in this patient based on the presence of five major criteria and two minor criteria. Laboratory investigations revealed dyslipidemia and renal insufficiency (creatinine clearance is 72 ml/min/m^3). Renal ultrasound demonstrated two kidneys of normal size.

Fig.2

DISCUSSION

The diagnosis of Bardet-Biedl syndrome (BBS) is made based on the criteria described by Beales *et al*., [1]. The syndrome is diagnosed with the presence of four major criteria or three major criteria and two minor criteria. The major criteria include obesity, retinitis pigmentosa, polydactyly, genital organ developmental anomalies, learning difficulties, and renal abnormalities. Minor criteria include delayed speech acquisition and phonetic disorders, brachydactyly and/or syndactyly, psychomotor developmental delay, excessive thirst and urination (diabetes insipidus), poor coordination of movements, the presence of diabetes mellitus, and cardiovascular abnormalities [2, 3].

BBS is genetically heterogeneous, with autosomal recessive inheritance. Twelve genes (BBS1 to BBS12) involved in the function of primary cilia have been identified [4]. These genes encode proteins involved in the development and function of primary cilia. Dysfunction of these proteins leads to ciliopathies affecting organs such as the kidney and eye. Molecular diagnosis is possible, providing the opportunity for genetic counseling and potential prenatal diagnosis [5].

CONCLUSION

Consanguinity is a major risk factor for the occurrence of Bardet-Biedl syndrome (BBS). The prognosis is primarily dependent on renal involvement, which should be systematically screened for.

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