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Clinical Spectrum of Bardet-Biedl Syndrome: A Case Series

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Abstract

Background: Bardet-Biedl Syndrome (BBS) is a rare autosomal recessive ciliopathy characterized by a wide spectrum of clinical manifestations including retinal dystrophy, obesity, post-axial polydactyly, learning disabilities, hypogonadism, and renal abnormalities. Early recognition is often challenging due to its gradual progression and overlapping features with other conditions, particularly in resource-limited settings where genetic testing is not readily available. We report a series of four Bangladeshi cases with variable phenotypic expressions of BBS. All patients presented with progressive visual impairment, obesity, and polydactyly, accompanied by additional features such as diabetes mellitus, dyslipidemia, hypogonadism, renal impairment, developmental delay, and behavioral disturbances. Diagnosis was established based on Beales' clinical criteria, with patients meeting multiple primary and secondary features. Renal involvement was observed in two cases, including diabetic nephropathy and focal segmental glomerulosclerosis, underscoring its role as a major determinant of morbidity and mortality. Management required a multidisciplinary approach involving endocrinology, ophthalmology, nephrology, and mental health services, alongside lifestyle modification, pharmacotherapy, and family counseling. This case series highlights the clinical heterogeneity of BBS, the importance of heightened clinical suspicion for early detection, and the need for comprehensive multidisciplinary care to improve long-term outcomes in affected individuals. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S49]*

Keywords: Bardet-Biedl Syndrome, Autosomal recessive, Retinal dystrophy, Obesity, Post-axial polydactyly

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