Bardet Biedl syndrome- A rare case

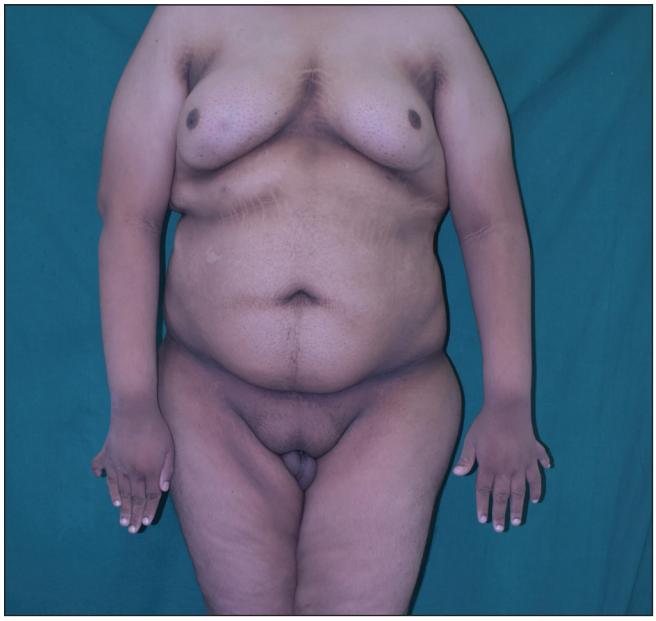

Figure 1: Features of BBS- central obesity, polydactyly, micropenis, gynaecomastia, and acanthosis nigricans.

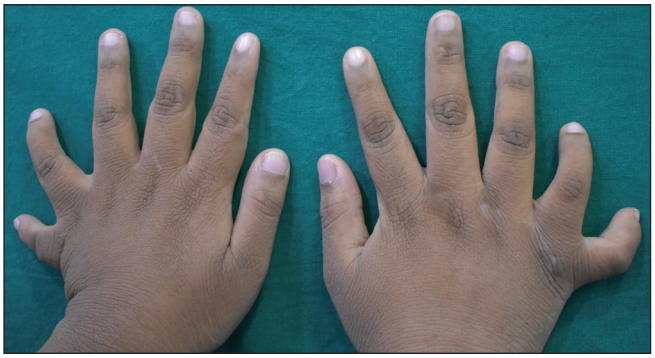

Figure 2: Post axial polydactyly of hands.

A 20-year-old obese male presented with a darkening of the skin over the neck, axillae, cheeks, and trunk. There was a history of second-degree consanguinity in parents. He was diagnosed with bilateral atypical retinitis pigmentosa and had a history of delayed milestones. On examination, he had acanthosis nigricans, deep-set eyes, hypertelorism, high-arched palate, post-axial polydactyly of all the limbs, gynaecomastia, and micropenis [Figures 1 and 2].

Based on the above clinical findings, Bardet Biedl syndrome (BBS) was diagnosed. The parents were counseled and advised to have regular follow-ups to look for any renal abnormalities.

BBS is a pleiotropic non-motile ciliopathy. It is related to multiple defective genes called Bbsomes like BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, BBS7, etc. BBS- protein, a gene product of these genes, plays a role in intra-flagellar transport. Diagnosis is mainly clinical; genetic confirmation is required when the clinical presentation is atypical.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent.
Financial support and sponsorship: Nil.
Conflicts of interest: There are no conflicts of interest.
Use of artificial intelligence (AI)-assisted technology for manuscript preparation: The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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How to cite this article: Shende A, Rambhia K, Kapadia F. Bardet Biedl syndrome- A rare case. Indian J Dermatol Venereol Leprol. 2024;90:681. doi: 10.25259/IJDVL_668_2023

Received: June, 2023 **Accepted:** August, 2023 **EPub Ahead of Print:** October, 2023 **Published:** August, 2024

DOI: 10.25259/IJDVL_668_2023 **PMID:** 38031692