Images in Clinical Practice

Bardet-Biedl Syndrome

An eleven year old obese boy weighing 41 Kg presented with repeated episodes of urinary retention *(Fig.* 2). He also had severe visual disability and night blindness from the very early childhood. General physical examination revealed mental retardation, polydactyly (Fig. 2) and hypogenitalism. On ocular examination, best corrected vision was 1/60 inboth eyes. He had nystagmus and fundus findings included waxy pallor of disc, attenuation of retinal vessels, atrophy of retinal pigment epithelium and bone-spicule pigmentation in the equatorial retina *(Fig.* 3). He was diagnosed to have a neurogenic bladder for which treatment with pharmacotherapy and clean intermittent catheterization was instituted.

Although the term Laurence-Moon-Biedl syndrome has been universally employed to describe the constellation of features described above, this is actually a misnomer. Now two distinct entities with a few overlapping features are recognized; namely the Laurence-Moon and the Bardet-Beidl syndrome. Common to both are retinal degeneration, mental retardation and hypogenitalism. Spastic paraplegia is characteristic of Laurence-Moon syndrome whereas polydactyly and obesity are prominent in the Bardet-Biedl syndrome. Although the patients with Bardet-Biedl syndrome are known to have renal involvement, association of neurogenic bladder as seen in our case has not been reported earlier.

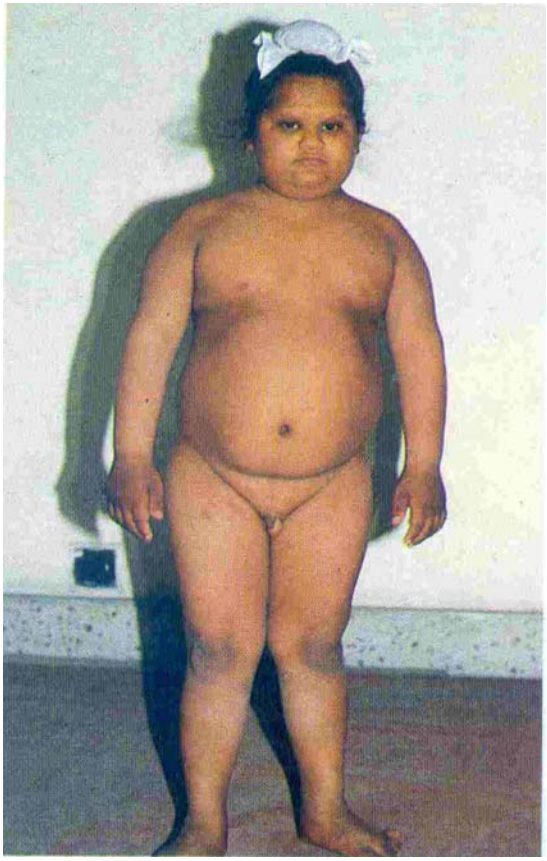

Fig. 1. Note short stature, fat moon shaped face and truncal obesity

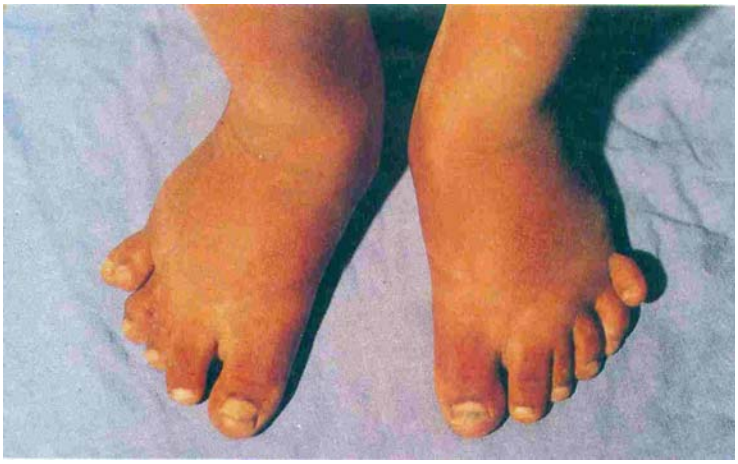

Fig. 2. *Close up photograph showing polydactyly in both feet*

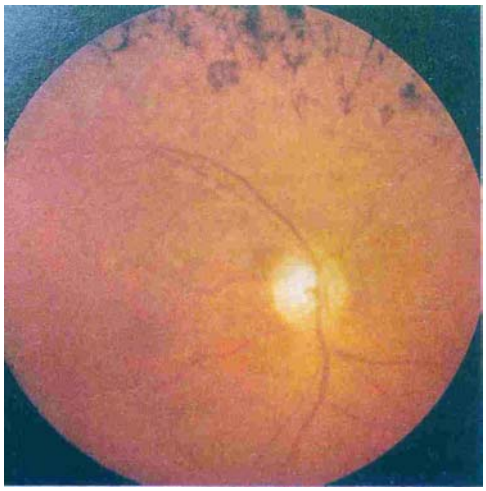

Fig. 3. *Fundus photograph showing pale disc and bone-spicule pigmentary degeneration.*

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