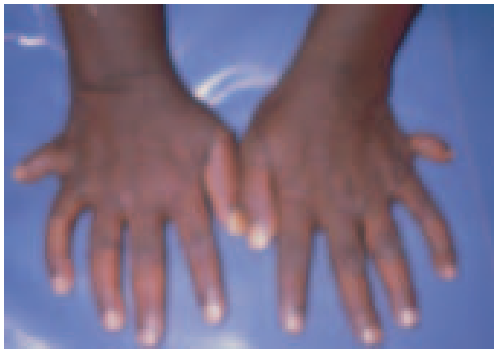
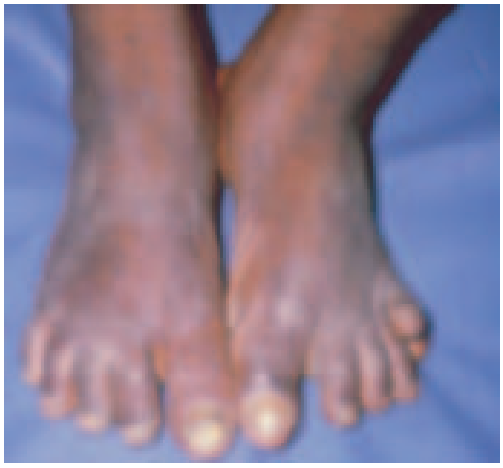
Bardet Biedl Syndrome





Figs.1 and 2: Picture showing post-axial polydactyly which is complete in the left foot and right hand and incomplete in the left hand

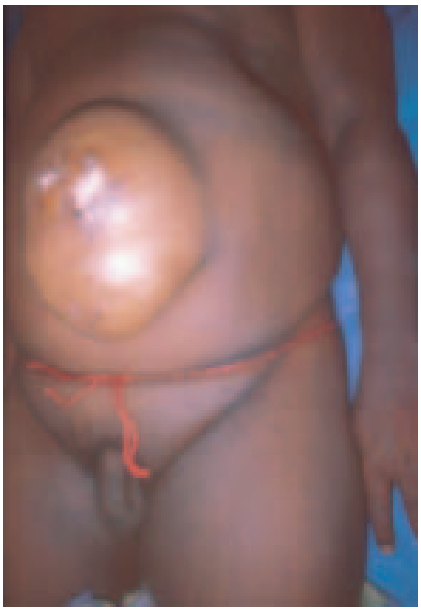


Fig. 3: Picture showing sparsely distributed body hair, central obesity and underdeveloped external genitalia – a smaller than normal testes and a microphallus.

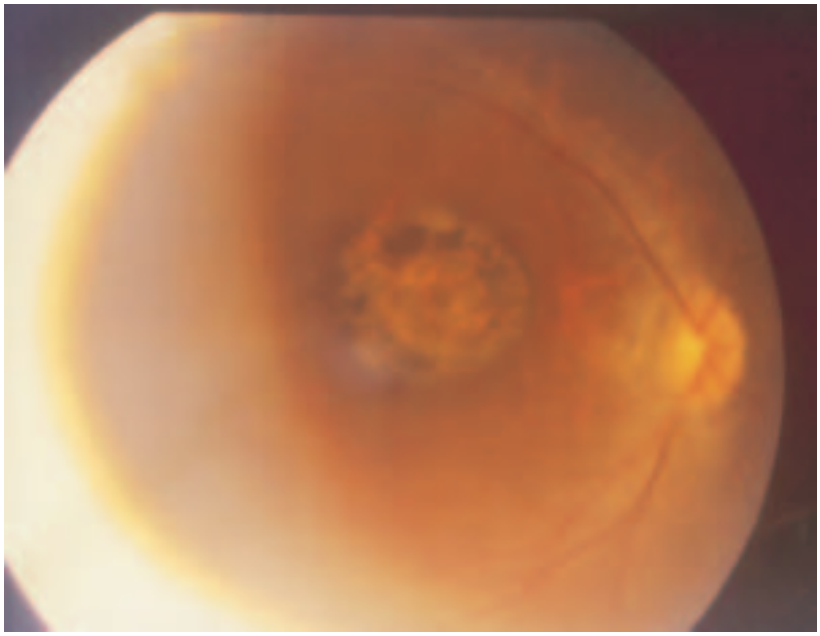


Fig. 4 : Picture of the right optic fundus showing atypical sectorial pigmentary retinopathy involving the macula and the myopic crescent.

A 30-year-old washerman was admitted for paraumbilical hernia repair. Patient was born of non-consanguineous parents with 6 fingers in his both hands and 6 toes on his left foot. He had impaired vision since childhood and had difficulty in learning and did not attend school. Other two siblings are normal except for short stature. He was obese and had a BMI of 34. Examination showed post-axial polydactyly which was complete in the left foot and right hand and foot and incomplete in the left hand (Figs. 1 and 2). He had sparsely distributed facial and body hair. He had underdeveloped external genitalia – a smaller than normal testes and a microphallus with hypospadias (Fig. 3). Examinations of the cardiovascular and respiratory system were unremarkable. Patient had a partially reducible paraumbilical hernia. Ultrasonogram showed a corticomedullary cyst in the left kidney. Ophthalmic evaluation revealed bilateral cortical cataracts, myopia and bilateral atypical sectorial pigmentary retinopathy (Fig. 4). Examination of the CNS was otherwise normal. A diagnosis of Bardet Biedl Syndrome was made as the patient had all the 6 cardinal features of the syndrome *viz.*: mental retardation (only a learning impairment in this instance as in most), post-axial polydactyly, hypogonadism, central obesity, pigmentary retinopathy (typical pigmentary retinopathy sparing the macula is unusual and occurs in only 10-15% of cases) and renal involvement. The absence of neurological abnormalities distinguishes this syndrome from the Laurence-Moon Syndrome.

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