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IMAGES IN MEDICINE

Image In Medicine- Laurence-Moon Bardet-Biedl Syndrome

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A 13 years old female presented to us with the complaint of diminution of vision mainly at night and slight decreased vision in the day. This vision loss was gradually progressive since last 3 years. She was in 5th standard because she failed thrice. On examination she was obese, short stature, moon like face, [Table/Fig 1] no secondary sexual characteristic (No breast and pubic hair development). There was polydactyly in both upper and lower limbs [Table/Fig 2]. Total fingers were 24. IQ was around 55. She was operated for club foot at the age of four. There was a history of consanguineous marriage of parents. His one younger brother had similar finding though he did not present with any complaint. Investigations and clinical findings were suggestive of hypogonadism. Cardiac, renal and hepatic functions were normal. Echocardiography was normal. USG for Pelvic organs revealed normal female internal genitals. Barr body examination was normal as per female sex. Fundus examination was suggestive of retinitis pigmentosa. All findings like short

stature, round face, mental retardation,

pigmentosa

and retinitis

diagnostic of Laurence-Moon Bardet-Biedl syndrome.



(Table/Fig 1) Female patients with short stature and obese.



(Table/Fig 2) Showing polydactyl in both feet.

polydactyle