CLINICAL IMAGES

Bardet-Biedl Syndrome-Rare Disorder

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Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder first described by Bardet and Biedl in the 1920. The principal manifestations are rod-cone dystrophy, postaxial polydactyly, central obesity, mental retardation and hypogonadism. Other features, not always present, include hepatic fibrosis, diabetes mellitus, neurological, deficits, behavioral traits, facial dysmorphism, dental anomalies and developmental delay (1, 2). A 22 year old male was admitted complaining of loss of vision, weight gain, and speech deficit. He was the third child of consanguineous parents (first- degree relatives). His one sibling had died at the age of 1, had polydactyly and the etiology of death was unknown.

On physical examination patient had BMI of 33 kg/m^2 horizontal nystagmus, rod-cone dystrophy (atypical retinitis pigmentosa) in his left eye, noticed on ophthalmologic examination (3). (*Fig 1*), on genital examination he had low testicular volume and his penis was small, buried in adipose tissue (*Fig 2*). He had a postaxial polydactyly (*Fig 3*)& mild mental retardation (4).

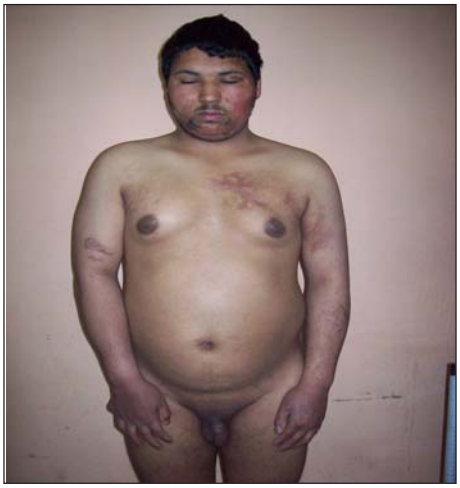

Fig 1. Obesity, Hypogonasdism, Horizontal Nystagmus, Rod-cone Dystrophy Left Eye of the Patient

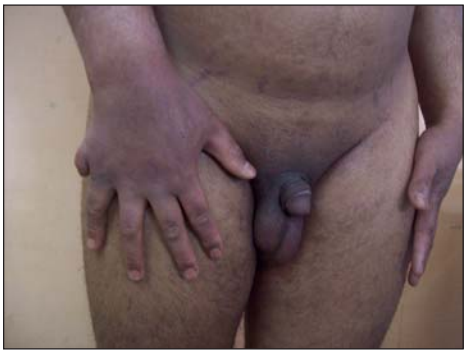

Fig 2. Small Testis & Penis Burried in Adipose Tissue

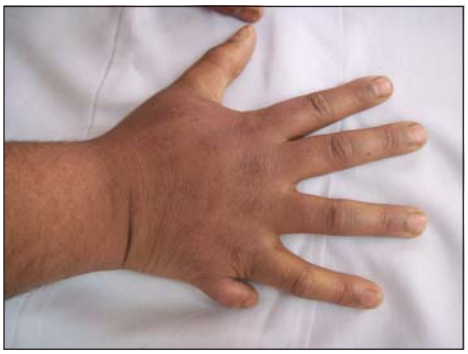

Fig 3. Extra Digit on Patients Right Hand

References

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