

Abstract

Klinefelter's syndrome is the commonest chromosomal abnormality seen in males occurring in 1 in 500-1000 phenotypically normal men.(1,2) Yet, it goes undetected and undiagnosed in a majority of affected patients.(3) It accounts for significant proportion of primary subfertility. We present a case of previously undiagnosed Klinefelter's syndromic patient who presented with osmotic symptoms to our unit. He was diagnosed with type 2 diabetes mellitus, central obesity, metabolic syndrome and investigated for lack of secondary sexual characteristics. He had erectile dysfunction and poor libido from childhood and there was no facial hair growth. Gynecomastia and tanner stage 1 and 2 of genitalia and pubic hair development was noted respectively. His FSH and LH was high and testosterone low and karyotyping revealed XXY aneuploidy. He was diagnosed with Klinefelter's syndrome and IM testosterone was started with a plan to gradually increase the dose along with life style modifications and optimized medical management for DM and metabolic syndrome. It is important to diagnose and treat these patients at least pre pubertally since it will help them avoid unwanted effects of hypogonadism such as gynecomastia, reduced insulin sensitivity and metabolic syndrome, obesity, achieve expected bone mass, enable them to receive special learning programs and receive necessary psychological support, overall, have a better quality of life.