Characteristics of sexual dysfunction in men with Klinefelter's syndrome: is it all about gonadal dysfunction?

Rastrelli G, Vignozzi L, Maggi M

Azienda Ospedaliero-Universitaria di Careggi; Centro ENDO-ERN

Abstract

Background: Klinefelter syndrome (KS) is the most common sex chromosome aneuploidy in males. While the association between KS and infertility and metabolic alterations has been extensively investigated, very few data on sexual health in KS men are available at the moment.

Aim: To evaluate the characteristics and correlates of sexual dysfunction in KS men compared to men with a gonadal dysfunction non KS-related and men with a normal testicular function.

Methods: A total of 4103 men consulting our outpatient clinic for sexual dysfunction (mean age 50.6 ± 13.7 years) was studied. Among them, 38 were diagnosed with KS, 1718 suffered from testicular dysfunction not KS-related (defined by luteotropic hormone (LH) > 9.4 U/L or follicle-stimulating hormone (FSH) > 8 U/L), while the other 2347 men presented a normal testicular function (total testosterone ≥12 nmol/L and normal LH and FSH).

Several clinical, biochemical, and instrumental (penile color Doppler ultrasound; PCDU) factors were evaluated.

Results: When compared to both men with normal testicular function and those with non KS-related gonadal dysfunction, KS patients showed decreased sexual desire (OR = 2.66 [1.22-5.82], 2.51 [1.14-5.54], respectively) reduced ejaculate volume (OR = 3.52 [1.68-7.37], 3.67 [1.74-7.71], respectively), uneasiness with masturbation (OR = 3.79 [1.64-8.72], 6.44 [2.72-15.26], respectively), and later andrological referral. Conversely, they were not characterized by impairment in morning erection, reduction in intercourse or masturbation frequency or relational issues. KS men less frequently complained of premature ejaculation and severe erectile dysfunction than men with normal testicular function and those with non KS-related gonadal dysfunction (OR = 0.17 [0.08-0.37], 0.26 [0.12-0.55], respectively), although having PCDU parameters comparable with either men with normal gonadal function and non KS-related gonadal dysfunction. When considering metabolic parameters, KS patients presented higher triglycerides and waist circumference (mean difference 6.53 [3.18-9.89], 4.71 [1.32-8.09] cm vs. eugonadal and non-KS gonadal dysfunction respectively) and an increased frequency of metabolic syndrome (OR = 5.53 [2.31-13.2], 3.86 [1.61-9.24] vs. eugonadal and non-KS gonadal dysfunction respectively). The aforementioned data derive from age- and BMI- analyses. The introduction of serum testosterone in the model attenuated most relationships, however lower severity of ED, more severely reduced perception of ejaculate volume and later andrological consultation as compared with men with testicular dysfunction were confirmed as peculiar of KS men.

Conclusion: Most sexual symptoms in KS men are common to men with gonadal dysfunction non KS-related. However, some symptoms may be specific features of KS. In particular, despite a worse metabolic profile and PCDU parameters comparable with the other categories of men with sexual dysfunction, KS men complain less about erectile dysfunction and are referred later to andrological consultation. Conversely, they report more frequently a decrease in ejaculate volume, lower sexual desire, and uneasiness with masturbation. Overall, these characteristics found in adult men consulting for sexual dysfunction and with primary impairment in testicular function should call for a possible diagnosis of KS and may suggest genetic assessment. Further studies are needed to obtain more insights on this topic.

Keywords: Klinefelter syndrome, sexual dysfunction, erectile dysfunction, hypogonadism.