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Baseline characteristics and hormonal pattern in Klinefelter syndrome patients: first data from the Klinefelter Italian Group (KING)

Giacomo Accardo¹, Daniela Pasquali¹, Paolo Chiodini¹, Vittorio Simeon¹, Carlo Foresta², Alberto Ferlin³, Mario Maggi⁴, Linda Vignozzi⁴, Giovanni Corona⁵, Fabio Lanfranco⁶, Vincenzo Rochira⁷, Aldo E. Calogero⁸, Vito A. Giagulli⁹, Marco Bonomi¹⁰, Rosario Pivonello¹¹, Giancarlo Balercia¹², Alessandro Pizzocaro¹³, Pietro Salacone¹⁴, Antonio Aversa¹⁵, AND Andrea Garolla², on behalf of the Klinefelter Italian Group (KING)

¹University of Campania "L. Vanvitelli", ²University of Padova, ³University of Brescia, ⁴University of Florence, ⁵Endocrinology, Bologna, ⁶University of Turin, ⁷University of Modena & Reggio Emilia, ⁸University of Catania, ⁹Endocrinology, Bari, ¹⁰University of Milan and IRCCS Istituto Auxologico Italiano, Milan, ¹¹University Federico II, Naples, ¹²University of Marche, Ancona, ¹³IRCCS, Istituto Clinico Humanitas, Rozzano-Milan, ¹⁴Santa Maria Goretti Hospital, Latina, ¹⁵University of Catanzaro Magna Graecia.

Introduction

Klinefelter syndrome (KS) is the most frequent chromosomal disorder, occurring in 1:500 to 1:1000 live male births, associated to male infertility. Although significant research has been conducted, KS remains frustratingly underdiagnosed with a remarkable portion of cases being unidentified, among which only 10% are in the prepubertal age while 25-50% in the adulthood.

As a consequence, medical research results are limited for the relatively small number of patients reported in scientific papers.

Aim

To better characterize KS in a nationwide cohort, here we describe the baseline characteristics and the hormonal patterns of patients currently attending a national network of academic or general hospitals of the Klinefelter Italian Group (KING).

Methods

A multicenter, observational study of 587 KS was performed among the patients regularly attending the KING centers, after written informed consent has been obtained.

Results

Up to now, 587 KS from 14 units have been registered. The mean age was 37.2 ± 13.5 years, and median was 36.0 (IQR 28–46). LH and FSH levels were 16.6 UI/L (median IQR 8.8–22.5) and 28.5 UI/L (median IQR 17.5 – 39), respectively (Fig. 1). Mean total testosterone was 350.1 ± 9.1 ng/dl (Fig. 2A). Estradiol and SHBG were in the normal range (Fig. 2B and C). The median testicular volume was 3 ml in both testis (Fig. 3A), BMI was 26.6 ± 5.8 (Fig. 3B) and 25.5% of the patients met the diagnostic criteria for metabolic syndrome (Mets) (Fig. 4).

Conclusions

Data from the present large cohort is demonstrating that diagnosis was reached very late in the adulthood, mostly following test for fertility problems. We can therefore imagine that KS is often underdiagnosed and raise the question about the true prevalence of the syndrome in Italy. Our patients presented with a wide spectrum of the classical Klinefelter symptoms. KS were overweight and, surprisingly, only 25,5% of them were diagnosed with Mets. This figure is very close to the Mets prevalence in the Italian general population quoted around 26%. In adulthood, two features were consistently present in every subject: small testes and high FSH and LH/ testosterone ratio, despite normal testosterone levels. We confirm that such biochemical parameters combined with small testes should lead to a suspicion of KS.

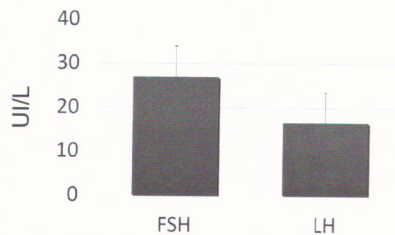


Fig. 1 FSH and LH plasma levels in 587 men with KS

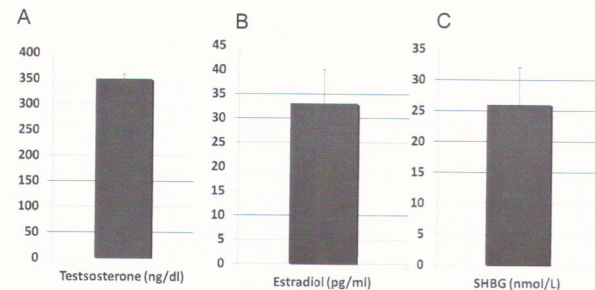


Fig. 2 Total testosterone (A), estradiol (B) and SHBG (C) plasma levels in 587 men with KS

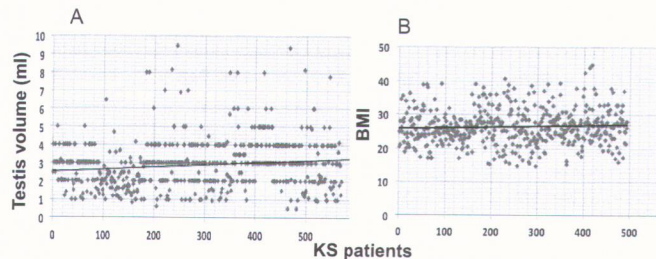


Fig. 3 Testis volume (A) and BMI (B) in 587 patients with KS

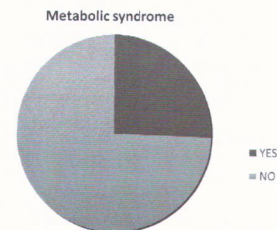


Fig. 4 Prevalence of Metabolic syndrome in 352 patients with KS