

CO 036 - CHARACTERISTICS, GEOGRAPHICAL DISTRIBUTION AND AGE AT DIAGNOSIS OF PATIENTS WITH KLINEFELTER SYNDROME IN ITALY: A COHORT STUDY FROM THE KLINEFELTER ITALIAN GROUP (KING)

D. Pasquali¹, A. Garolla², G. Accardo¹, P. Chiodini³, V. Simeon⁴, A. Ferlin², M. Maggi⁵, L. Vignozzi⁵, G. Corona⁶, F. Lanfranco⁷, V. Rochira⁸, A. Calogero⁹, V. Giagulli¹⁰, M. Bonomi¹¹, R. Pivonello¹², G. Balercia¹³, A. Pizzocaro¹⁴, P. Salacone¹⁵, A. Aversa¹⁶

¹Dipartimento di Scienze Mediche e Chirurgiche Avanzate Napoli, ²Università di Padova Padova, ³Dipartimento di Salute Mentale e Fisica E medicina Preventiva Napoli, ⁴Università di Brescia Bologna, ⁵Università di Firenze Firenze, ⁶AUSL di Bologna Bologna, ⁷Dipartimento di Scienze Mediche di Torino Torino, ⁸Università di Modena e Reggio Emilia Modena, ⁹Università di Catania Catania, ¹⁰ASL di Bari Bari, ¹¹Università di Milano e IRCCS Istituto Auxologico Milano, ¹²Università di Medicina Clinica e Chirurgica Napoli, ¹³Università di Ancona e delle Marche Ancona, ¹⁴IRCCS, Istituto Clinico Humanitas, Rozzano-Milan Milano, ¹⁵Santa Maria Goretti Hospital Latina, ¹⁶Università di Catanzaro Catanzaro

Introduction

Klinefelter syndrome (KS) is the most frequent chromosomal disorders, 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. Under diagnosis may be due to men's hesitancy about seeking medical attention, low awareness of KS among health professionals, and failure by health professionals to perform routine genital examinations in adult men.

Aim Our purpose was to describe the phenotypic characteristics and the hormonal patterns of a large cohort of patients currently attending a national network of academic or general hospitals of the Klinefelter Italian Group (KING). Moreover, we focused our interest on the geographical distribution, and age at diagnosis of KS patients in Italy.

Methods

A multicenter, observational study of 594 KS was performed among the patients regularly attending the KING centers, after written informed consent has been obtained.

Results

Five hundred and ninety four KS from 14 units have been registered. The mean age was 37.4±13.4 years (median IQR 28–46). The mean testicular volume was 3 ml in both testis (fig 1 A), BMI was 26.6±5.8 (fig 1 B) and 25.5% of KS meet the diagnostic criteria for metabolic syndrome (Mets). Mean total testosterone was 350±9.1 ng/dl, and LH and FSH mean levels were 16,6 (median IQR 8.8-22.5) and 28.5 (median IQR (17.5 - 39), respectively. A descriptive analysis performed in 594 KS, showed that 329 KS were referred to KING centers of Northern Italy, 65 and 200 KS patients to KING facilities in Central and Southern Italy, respectively. Analysis of variance showed significant statistical differences (p<00000) between the ages at diagnosis of the KS of the three geographical groups. In particular the age of KS patients was significantly lower in Southern Italy (33.3 ± 13 SD) compared to Central and Northern Italy (40.2 ± 12.5 SD and 39.2 ± 13.3 SD).

Conclusions

Our preliminary data showed that KS is highly underdiagnosed in Italy, raising the question of the true prevalence of KS. 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. The differences of KS age between geographical groups highlight the need for increased awareness leading to timely detection.