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A Data Profile of Phenotypic Features in 72 Klinefelter Syndrome (KFS) Males

Vallabhajosyula Ranganath[*] and Sayee Rajangam[**]

[*]Newcastle University Medicine Malaysia, Nusajaya, Johor, Malaysia
[**]Department of Anatomy, International Medical School MSRIT post, New BEL Road, Bangalore 560 054, Karnataka, India

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ABSTRACT Klinefelter syndrome phenotype is associated with hypogonadism and infertility that results from 47,XXY or 46,XY/47,XXY karyotype. Men with mosaic status show milder phenotype than those of non-mosaics. The present study aimed to report, a data profile on the observed phenotypic features in 72 cytogenetically confirmed Klinefelter syndrome male gathered from duly filled proforma. The reported phenotype from the literature were categorized into 14 groups (highly arched palate, winged scapula, thin long fingers, flat feet, prognathism, liver cirrohsis, seizures, mental illness, penis, gonads, axillary hair growth, and pubic hair growth, presence of gynaecomastia and semen analysis). The calculated total number of the 14 features multiplied for the 72 samples was 1,008. Of the 1,008 features (14X72), KFS male manifested only 16.56% of abnormal features (167/1,008). Scanty axillary hair growth (25%, 18), scanty pubic hair growth (26.38%, 19), small sized penis (25%, 18), small sized gonads (55.56%, 40), presence of gynaecomastia (45.83%, 33) were of highest percentage. It was noticed that, for the entire sample of 72, the manifestation of the 14 categorised features was only 16.56%, irrespective of the karyotype; out of which, with 47,XXY, the manifestation of the phenotypic features was observed to be highest (18.52%, 153/826). The findings confirmed the reported observations that in Klinefelter syndrome, there seemed to be a wide variability in the phenotype.