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Relation between morphological and karyotypic sex in individuals with intersex

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Aim: The main aim of the study was to determine the frequency of different karyotypes in intersex individuals and to find out the relation between their morphological and karyotypic sex.

Method: Using a non-probability purposive sampling technique a cross sectional study was done in the Department of Genetics, Children Hospital Lahore with the facility of performing karyotypic analysis. This study was completed in one year by taking 140 cases. Patients were examined for their morphological sex and its comparison was done with sex on karyotypic analysis.

Result: When physical examination was studied in coordination with karyotyping the agreement was very good with p-value<0.001. The karyotypic analysis of 140 intersex individuals revealed that 40.7% of them were genetically males (46 XY) while 17.8% were genetically males but phenotypically had more of female characters (male pseudo hermaphrodites). In contrast to above 30.7% were genetically females (46 XX) while 2.1% displayed genetically 46 XX chromosomal compliment with physical appearance more of maleness (female pseudo hermaphrodites). Turner's syndrome 45 X0 was found in 5% while 0.7% of individuals had Klinefelter's syndrome. Remaining 0.7 % individuals were presented as XXX female.

Conclusion: Frequency of different karyotypes was studied in individuals with intersex. Relation between morphological, karyotypic and sex of rearing sex was also established. Sex of rearing co-relates with phenotypic sex in most of the cases of intersex. However, sex of rearing maybe altogether different from karyotypic sex. If karyotypic sex is determined late, changing the sex of rearing becomes almost impossible as it bears grave impact on psychosocial aspects of personality.

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