



Physical Anthropology Section – 2003

H6 Sex, Size, and Genetic Mistakes: Identifying Disorders of Sexual Differentiation in Human Skeletal Remains

Linda O'Connell, BM, MSc, Joy Steven, MSc*, and Margaret Cox, PhD*, School of Conservation Sciences, Bournemouth University, Poole, Dorset, United Kingdom*

This presentation will assess whether disorders of sexual differentiation can be identified in human skeletal remains and whether such changes affect the determination of sex. Results show that a number of skeletal manifestations associated with each of the disorders and that these may compromise the effectiveness of sex assignment.

One of the most fundamental demographic parameters to be ascertained as part of any forensic or biological anthropological analysis is that of sex determination. The term sex can be defined in one of five ways: chromosomal sex, gonadal sex, phenotypic sex, gender, and sexual orientation. However, when forensic and biological anthropologists refer to this term, they are alluding to the biological sex of an individual, which is fixed by the X and Y-chromosomes, and dictated by morphological characteristics. The potential effects of disorders of sexual differentiation upon elucidation of this demographic parameter, (particularly with respect to the effects on the pelvis and skull) has not been considered within this context.

A desk-based assessment extensively reviewed five selected disorders of sexual differentiation - Turner's syndrome, Klinefelter's syndrome, congenital adrenal hyperplasia, testicular feminization, and true hermaphroditism. The pathogenesis, aetiology, epidemiology, clinical features, and treatments of these conditions were investigated, together with any other mimicking conditions.

Results showed that there are a number of skeletal manifestations associated with each of the disorders. Examination of these traits revealed that the normal expression of skeletal sexual dimorphism could be compromised as a result, with individuals being incorrectly assigned a sex.

Furnished with this knowledge, the forensic or biological anthropologist will be in a position to not only offer a potential diagnosis, but also to fully evaluate any effects that this may have on the features employed for sex determination.

Disorders of Sexual Differentiation, Sex Determination, Skeletal Analysis