



Physical Anthropology Section – 2007

H31 Sources of Error in Genetic and Osteological Sex Determination: Lessons from Physical Anthropology

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The goal of this presentation is to present forensic scientists with an understanding of the error associated with both the genetic and morphological based tests of sex determination, and to stress the fact that forensic scientists should employ and understand a wide range of scientific tools when analyzing human remains in a forensic context.

This presentation will impact the forensic community and/or humanity by introducing important information on the advantages and limitations of genetic and morphological sex determination, and will present a critical assessment of the interpretational error associated with the techniques using anthropological examples.

An important duty of the forensic scientist is to present their opinion in a judicial setting. An experienced expert witness should be prepared for conflicting opinions presented by opposing counsel. An opportunity for such a situation to arise may involve the determination of sex from skeletal remains. Determining sex from the skeleton can be accomplished by both morphological and genetic analyses. Forensic anthropologists are skilled at an osteological determination of sex and are aware of the error associated with interpretation of their analyses. Such investigations are extremely accurate when the entire skeleton is present for examination, and become more difficult as skeletal elements are missing and/or fragmented. Non-metric analyses examine sexual dimorphic differences in the size and shape of bones, and rely on the training of the investigator. Metric analyses employ classification based on measurements of various skeletal elements. Many forensic anthropologists use the discriminant functions in FORDISC software to classify the individual based on comparison to a data base of individuals of known sex. They should be trained in the interpretation of the results involved in classifications, should have a working knowledge of the statistical methods employed by FORDISC, and should understand that the probability of an individual being misclassified is not random.

Determination of genetic sex employs polymerase chain reaction (PCR) amplification of X and Y chromosome-specific DNA fragments of different sizes. Misclassification can occur with this test and has been attributed often to a deletion in the relevant area of the Y-homologue. Other interpretational issues include contamination, stochastic fluctuation, and preferential amplification. Laboratory contamination is rare; however, remains can become contaminated during collection. Stochastic fluctuation, or sampling error, and preferential amplification are both phenomena that may occur with the low quality and quantity DNA

associated with forensic evidence. Females have two X chromosomes and failed amplification of one X-homologue would still indicate a female. Males have one X and one Y chromosome. In a male sample, failed amplification of the X-homologue would raise concern, but would not lead to misclassification due to the presence of the Y-homologue. However, if the X-homologous fragments are preferentially amplified over the Y-homologous fragments, male samples would be misclassified as female. A forensic DNA analyst must therefore have the ability to analyze and interpret samples that are usually highly degraded, and must have an understanding of the physical evidence as well as the techniques used to analyze that evidence.

Genetic sex tests are automatically included in genetic identification tests like CODIS (combined DNA index system). With what is being called the "CSI Effect" the general public is over confident in scientific evidence, especially genetic evidence. There can be instances where morphological and genetic sex could provide conflicting results, and it is imperative that forensic experts understand the strengths and weaknesses of all of the scientific techniques involved in the analysis of skeletal remains.

Sex Determination, Osteology, Genetics