



A3 Genetic Analyses on Bone Remains: The University of Rome “Sapienza” Laboratory of Forensic Genetics’ Experience

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After attending this presentation, attendees will understand some principles of genetic analysis on bone remains. The challenges related to this kind of investigation, especially for what concerns criminal cases and the importance of the honesty of the forensic scientist when a certain and unequivocal interpretation of the DNA profile obtained cannot be provided.

This presentation will impact the forensic science community by highlighting the importance of bone remains as an evidentiary sample in forensic caseworks and the difficulties related to the genetic analysis of such samples due to degradation and/or inhibition factors. In these cases it is fundamental for the scientist to consider asserting that a complete and interpretable genetic profile not obtained from a sample (thus the sample cannot be considered useful for a comparison) does not mean a failure but, on the contrary, reveals scientific honesty and should stimulate the necessary progress in this field.

Genetic analyses on bone remains in the field of human identification represent one of the most stimulating and complex challenges for forensic geneticists. Unlike the analysis of biological materials such as blood, semen, saliva or urine that usually do not present any particular technical and operational difficulty so that personal identification can be achieved, the identification of bone remains forces the analysts to face multiple and complex variable factors (e.g., the degradation of genetic material and the environmental contamination of the samples) that can affect the success of obtaining a complete and interpretable STR profile. In such cases an accurate evaluation of the characteristics of the sample and the environmental conditions to which this finding has been exposed is extremely important.

This presentation describes the methods used in the Laboratory of Forensic Genetics of the Department S.A.I.M.L.A.L. of the University of Rome “Sapienza” for the analysis of bone remains for the purpose of either personal identification or the assessment of a parental relationship. A selection of twenty cases during the years 2007-2011 where genetic analyses were performed on different bone samples (femur, tibia, humerus, mandible) using different DNA extraction, amplification, and STR typing methods will be presented. The results obtained from each case will be compared in order to assess the specific role of three important variable factors: the age of the remains, the environmental conditions of storage/finding, and the cause of death.

Six out of the twenty cases showed interpretation problems related to the DNA degradation caused by environmental factors (for instance, the effects of high temperatures in case of charred remains and the acceleration of autolytic processes in case of hexumation of a corpse) and/or the presence of PCR inhibitors (e.g., calcium phosphate, humic acid) that likely were co-extracted with the DNA from the evidence sample. The results show that the possibility of obtaining a complete and interpretable genetic profile depends largely on the three variable factors mentioned above, particularly with regard to the environmental conditions of storage/finding of the remains, thus confirming the need to optimize the analytical methods to minimize the effects of environmental inhibitors.

Genetic Analyses, Personal Identification, Bone Remains