

# JOURNAL OF INTEGRATED OMICS

A METHODOLOGICAL JOURNAL

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SPECIAL ISSUE: SELECTED ABSTRACTS OF THE II INTERNATIONAL CAPARICA CONFERENCE IN TRANSLATIONAL FORENSICS (FORENSICS 2019)

## RNA markers analysis and post-mortem interval: a review of the evidence

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**Available Online:** 31 August 2020

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### ABSTRACT

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One of the most important problems in forensic sciences is the post-mortem interval (PMI) determination. Forensic scientists always provided different methods (physical, chemical, and entomological) to estimate a correct PMI, without success. However, the improvements of last two decades in the field of molecular biology, allowed us to evaluate the time-dependent degradation of biological markers (e.g. proteins, DNA and RNA). Thus, we want to present a review of the recent progress in the estimation of PMI, mainly focusing on the potential usefulness of RNA markers. Therefore, we reviewed 29 studies, each one chosen according to specific inclusion criteria. These studies evaluated the role of endogenous reference genes in different biological samples in order to determine the PMI based on post-mortem RNA degradation in relation with other influencing factors such as time, cause of death and environmental conditions.

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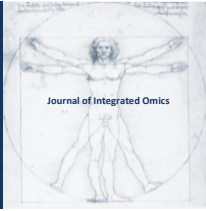
#### Acknowledgments:

We wish to thank Prof. Caenazzo for critical discussion and comments on the different drafts of the present contribution.

#### References:

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## DNA-based victim identification in mass disasters: ethical aspects.

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**Available Online:** 31 August 2020

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### ABSTRACT

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Even if every mass disaster is an unexpected event with specific characteristics that depend on the nature of the event, it always causes human, material, economic and/or environmental losses. Each disaster arises a chain of events in which victims' identification, independently from the absolute number of them, has a wide relevance. DNA analysis is often applied to the identification of mass disasters' victims. In fact, DNA profiling plays a prominent role in the identification of human remains, becoming sometimes indispensable, especially when they are badly damaged, commingled, or decayed. Since everyone involved in forensic work is also involved in an activity characterized by ethical obligations towards the deceased and the bereaved, forensic geneticists can be asked to face some ethical issues concerning DNA profiling. The DNA techniques, in fact, allow identification of an individual, distinction between fragments, and enable fragments to be reassembled and assigned to an already identified body, but raise specific problems, including ethical issues. Even if several guidelines and standards have been published in order to improve identification process and technical aspects of data collection and analysis, the complex implications of DNA typing for the purpose of identifying victims of mass disasters need to be further explored. Authors seek to delineate the principal ethical aspects of individual identification based on DNA technology, ranging from humanitarian importance of identification, through resource allocation in emergency contests with resource limitations, to the appropriateness of using samples collected for identification purposes for the secondary purpose of research.

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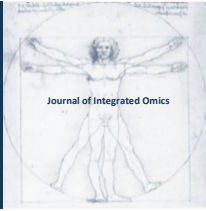
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#### References:

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## Methodological approach to detect HLA-B27 allele in case of degraded human bone remains

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**Available Online:** 31 August 2020

### ABSTRACT

Paleopathology is a subdiscipline of Forensic Anthropology, which is specialized in the study and application of methods and techniques for investigating diseases from skeletal remains [1]. Paleopathology offers a wide range of forensic applications, such as skeletal identification to reconstruct the ante-mortem biological biography, and providing key information to solve criminal investigations [2].

Spondyloarthropathies (e.g. Ankylosing Spondylitis, Reactive Arthritis, Psoriatic Arthritis) are a group of arthropathies (diseases that affects the joints in the body), whose most remarkable feature is the axial involvement of the skeleton [3]. In some cases, it is possible to establish a morphological diagnosis based on the presence or absence of some macroscopic change criteria, which may help to establish the differential diagnosis [4]. However, paleopathological studies face several problems such as the absence, in many cases, of important parts of the skeleton or the presence of some taphonomic alterations. In these cases, the morphological diagnosis is limited and sometimes impossible.

Considering the genetic background of Spondyloarthropathies [5], we are trying to investigate the use of molecular technology in forensic genetics to help to establish the diagnosis of these diseases. This is particular relevant when analyzing degraded human bone remains, in which morphological information is quite limited. Traditionally, the presence/absence of HLA-B27 allele by conventional PCR has been used as a genetic marker associated with Spondyloarthropathies [6]. However, this technique has some important limitations, especially in the case of degraded human bone remains. A negative result for HLA-B27 may be determined as an absence of the allele in that individual, while this result can be a consequence of the scarcity of DNA in the sample to obtain a result.

In the present study, we propose a new methodological approach to detect the presence or absence of HLA-B27 allele by genotyping 2 SNPs of this allele (rs\_116488202 / rs\_4349859) using quantitative-PCR (qPCR). In the case of detecting these 2 SNPs in the DNA sample, we can conclude that HLA-B27 allele is present in the individual. In comparison to conventional PCR, qPCR reduces negative results. This is because by using qPCR, the two alleles of each SNP are determined, obtaining always a result, even from a DNA extracted from degraded human remains.

In conclusion, a new methodological approach is proposed to detect specifically HLA-B27 allele, through the analysis of 2 SNPs by quantitative PCR (rs\_116488202 / rs\_4349859). This methodological tool could help to improve the diagnosis of Spondyloarthropathies, precisely, in degraded human remains, in which morphological diagnosis is quite limited.

**Keywords:** Forensic Anthropology, Forensic Genetics, Paleopathology, Spondyloarthropathies, HLA-B27, SNPs

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**Acknowledgments:**

This work was supported by the Basque Government Grant to Research Groups of the Basque University System (IT1138-16), and the granting of a predoctoral fellowship to Nerea G. Ventades (2018\_1\_0139).

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