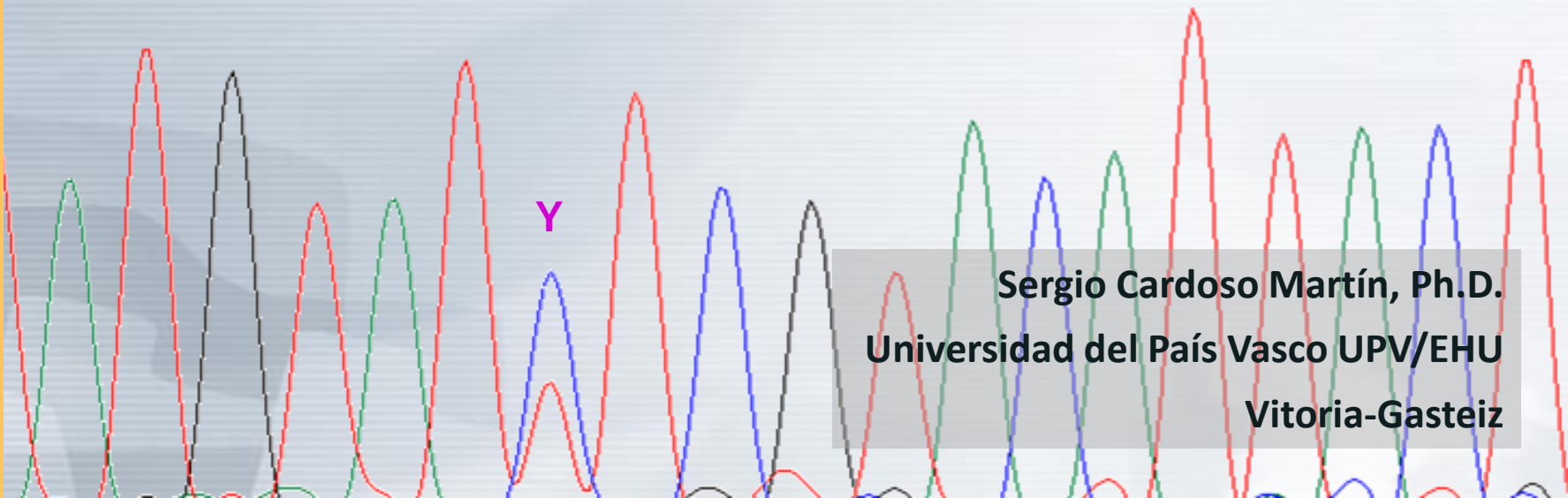
# COLLABORATIVE EXERCISE REGARDING

# MUTATION OF MITOCHONDRIAL DNA IN HAIR SHAFTS



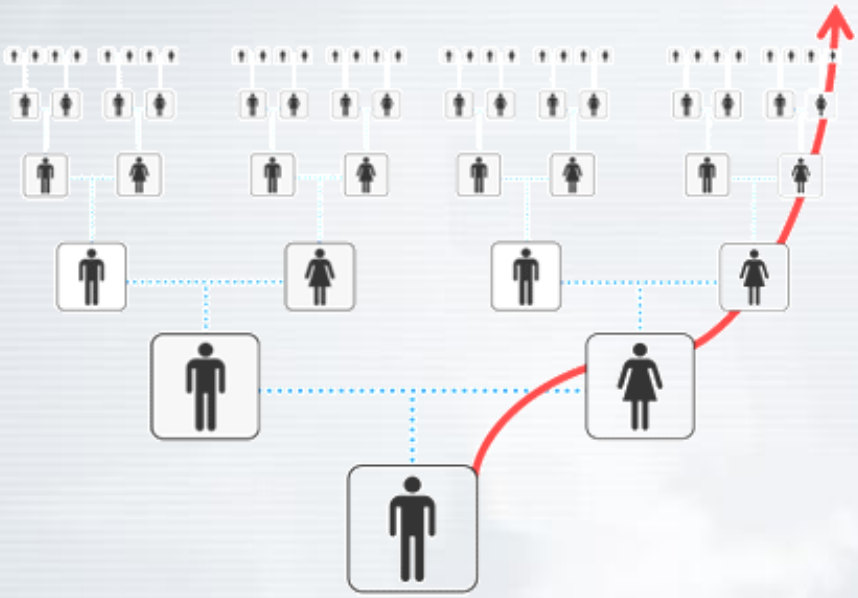

**Sergio Cardoso Martín, Ph.D.**
**Universidad del País Vasco UPV/EHU**
**Vitoria-Gasteiz**

Phylogenetic component of mtDNA lineages.

Importance of classification of haplotypes into haplogroups.

Detection of errors.

Information about phylogeographic origin of any lineage.



**Concept for estimating mitochondrial DNA haplogroups using a maximum likelihood approach (EMMA).** Röck AW, Dür A, van Oven M, Parson W. ***Forensic Sci Int Genet.*** **2013 Aug 12.**

**Phylotree + mtGenomes + maximum likelihood approach** → **EMMA**

EMPOP

**Fluctuation rates**

↓

**Diagnostic weight of a specific mutation is not the same in different haplogroups**

**Implications for haplogrouping of mtDNA sequences, especially when incomplete information of the Dloop is available.**

## Fluctuation rate

- Instability of particular mutations within haplogroups.
- Allows to improve haplogrouping.

## Somatic mutation rate

- Instability of mutations across the entire mtDNA tree and in different tissues.
- Is of special interest in Forensic Genetics.

**To study mutation of mitochondrial DNA in specific haplotypes is of interest, since it represents an exploratory approach to deepen knowledge about the variability of the bases which constitute the mtDNA.**

## BACKGROUND

Oftentimes it is possible to observe that mtDNA sequence of the same individual obtained from different tissues shows some differences. In fact, some tissues such as hair shafts have a higher tendency to undergo mutations. Thus, depending on the tissue available to carry out mtDNA analysis, the potential presence of mutations may affect the characterization of a question sample and lead to erroneous conclusions. On this basis, it is essential to deepen knowledge about the behaviour of mtDNA in different tissues of forensic interest.

Universidad del País Vasco
Euskal Herriko Unibertsitatea

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Universidad del País Vasco
Euskal Herriko Unibertsitatea

Institute for Legal Medicine,
Innsbruck Medical University

# PROPOSAL

## MAIN OBJECTIVE

**To study behaviour of mtDNA in hair shafts, a tissue of forensic interest frequently subjected to analysis of mtDNA. Thus, we aim to deepen knowledge about variability of specific positions of control region in this tissue and in a specific haplotype.**

## SPECIFIC OBJECTIVES

**To analyze the complete mtDNA control region in hair shafts from a single donor who shows point heteroplasmy/ies in this tissue, but does not in the reference samples (blood and saliva).**

## PARTICIPANTS

- **Those laboratories that successfully passed the analysis of complete control region of mtDNA in sample M5 (hair shaft) in the last Intercomparison Program (2013).**
- **Those laboratories that successfully passed the analysis of sample M5 by studying at least HVS1 and HVS2, only if they previously analyze the complete control region of an additional hair shaft as a "validation" test.**

## BASIC ASPECTS

- On the basis of 8 possible laboratories:
- 52 hair shafts from a single donor, divided into 4 fragments: 208 total fragments.
  - Each laboratory should analyzed 26 fragments belonging to 13 different hair shafts.
  - Each hair shaft would be analyzed by at least two laboratories (2 fragments from the same hair shaft to each lab).
  - If a hair shaft is divided into four fragments (1, 2, 3 y 4), one lab would analyzed fragments 1 and 3 and another lab fragments 2 and 4.
- Possibility to use self designed primers/protocols.
- Submission of haplotypes in EMPOP format and raw data (.ab1).

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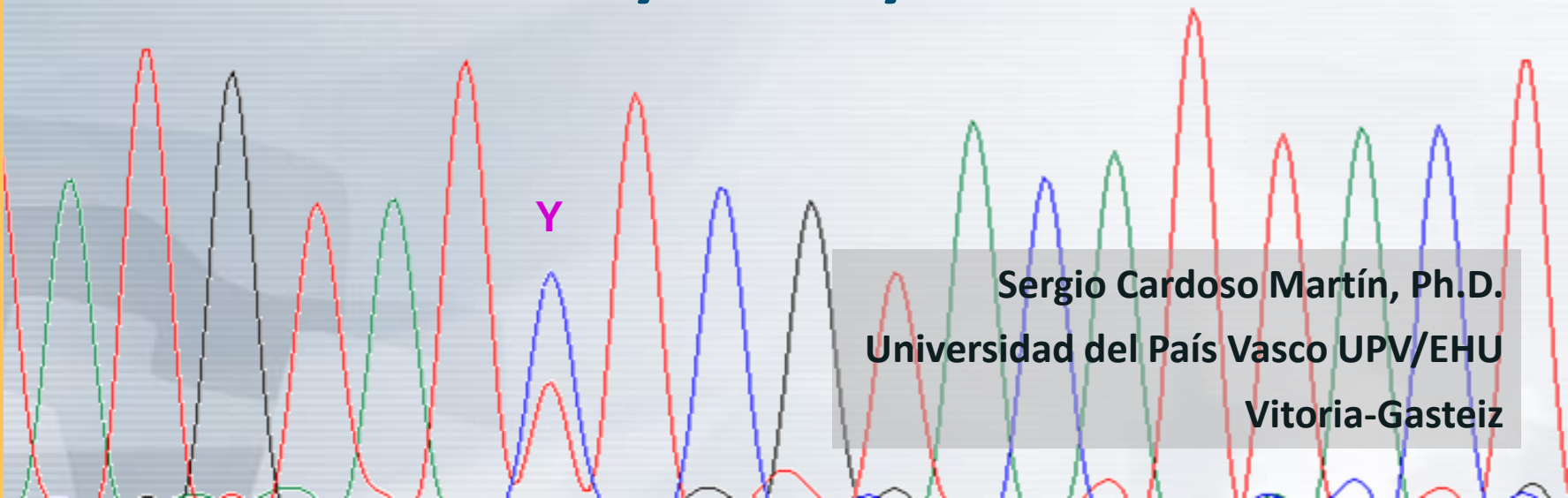
# Muchas gracias

# Thank you very much

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