



Contents lists available at ScienceDirect

journal homepage: www.elsevier.com/locate/humimm

Research article

HLA allelic diversity in the Waorani population of Ecuador: Its significance to their ancestry and migration

Samantha Saenz Hinojosa ^{a,1}, Timothy Adrian Jinam ^{b,1}, Kazuyoshi Hosomichi ^c, Vanessa I Romero ^{a,*}^a School of Medicine, Universidad San Francisco de Quito, Quito, Ecuador^b Department of Para-Clinical Sciences, Faculty of Medicine & Health Sciences, University Malaysia Sarawak, Malaysia^c Laboratory of Computational Genomics, Tokyo University of Pharmacy and Life Sciences, Tokyo, Japan

ARTICLE INFO

Keywords:

HLA alleles
Waorani population
Ecuador
Ancestry

ABSTRACT

The Waorani, an isolated indigenous tribe in Ecuador, have long been characterized by limited genetic diversity, with few studies delving into their genetic background. Human Leukocyte Antigen (HLA) genes which are located in the human major histocompatibility complex (MHC) provides valuable insights into population evolution due to its highly polymorphic nature. However, little is known about the HLA diversity and ancestry of the Waorani population. In this study, we sequenced eight HLA genes using Next Generation Sequencing (NGS) from 134 Waorani individuals and obtained up to four-field HLA allele resolution. Cluster and phylogenetic analysis show that the Waorani are genetically distant from other Ecuador populations, but instead show genetic affinities with the Puyanawa and Terena tribes from Brazil, as well as the Mixe tribe from Mexico. The identification of alleles common within the Waorani population, previously linked to specific health conditions, notably paves the way for future association analyses. This extensive study, employing Next-Generation Sequencing (NGS) technology, significantly enriches the sparse and segmented understanding of HLA diversity in the South American region. Our findings enhance the global comprehension of human genetic diversity and underscore the value of studying indigenous populations. Such research is vital for deepening our insights into human migration patterns and evolutionary processes.

1. Introduction

Ecuador, a country situated on the equator in South America, is globally recognized for its extensive biodiversity, encompassing both its ecosystems and its diverse human populations. Despite its modest dimensions, Ecuador boasts a remarkable geographical and cultural diversity, encompassing four distinct regions. Each region is characterized by its own unique flora and fauna: the Sierra, with its mountainous highlands; the coastal lowlands; the Amazon rainforest; and the Galápagos Islands. Notably, Ecuador exhibits significant population diversity, as demonstrated by the 2010 population census, the latest available as of now, which delineated various ethnic groups. These groups include Mestizos (71 %), a mixed population of White and Native Americans; Montubio (7.4 %), representing farmers from the Ecuadorian coast; Amerindians (7 %); White individuals (6.1 %); Afro-Ecuadorians (4.3 %); Mulato (1.9 %); Black (1 %); and 0.4 %

identifying with other ethnic groups [1]. The Amerindians in Ecuador are divided in 13 indigenous nationalities [2] being one of those the Waorani.

The Waorani, an indigenous tribe located in the Amazon region of Ecuador, specifically within the provinces of Napo, Orellana, and Pastaza (see Fig. 1), play a vital role in the country's demographic diversity. The current estimated population of the Waorani is approximately 3000 individuals, this figure includes those of mixed ancestry. They self-identify using the term “wao,” which translates to “people,” thereby distinguishing themselves from the rest of society, which they refer to as “cowode,” meaning “non-people” [3]. Traditionally, the Waorani followed a nomadic hunter-gatherer lifestyle, residing in small clan settlements, which contributed to their previously homogeneous population [4–6]. The tribe's first encounter with the outside world occurred in 1958 through interactions with American missionaries [4]. In contemporary times, some modern Waorani individuals

Abbreviations: DNA, Deoxyribonucleic Acid; HLA, Human Leukocyte Antigen; MHC, Major Histocompatibility Complex, PCA, Principal Component Analysis.

* Corresponding author.

E-mail address: vromero@usfq.edu.ec (V.I Romero).

¹ Authors contributed equally to the work.

<https://doi.org/10.1016/j.humimm.2024.110771>

Received 31 July 2023; Revised 9 February 2024; Accepted 28 February 2024

Available online xxx

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