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Prehistoric polydactylism: Biological evidence and rock art representation from the Atacama Desert in northern Chile

Vivien G. Standen[a,*], Calogero M. Santoro[b], Bernardo Arriaza[b], Daniela Valenzuela[a], Drew Coleman[c], Susana Monsalve[a]

[a] *Departamento de Antropología, Universidad de Tarapacá, Arica, Chile*
[b] *Instituto de Alta Investigación, Universidad de Tarapacá, Arica, Chile*
[c] *Department of Geological Sciences, University of North Carolina, USA*

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ABSTRACT

A review of the bioarchaeological collections from the site Morro de Arica in northern Chile allowed the identification of two cases of human polydactyly. Both cases are from the Chinchorro culture, hunters, fishers, and gatherers with a maritime orientation who inhabited the coast of the Atacama Desert (9000–3400 BP). Additionally, the analyses of 75 rock art sites in the area, from the Formative to Late Intermediate Periods (3000–550 BP), allowed the identification of hands and feet with six digits. Given the bioarchaeological record of polydactyly, it is highly probable that the rock art images were based on real individuals with polydactyly. However, the Sr chemical signal in a juvenile with polydactyly is the same as the Sr chemical signal in the rest of the individuals buried in the same site, proving that all the individuals were born and lived on the coast. We discuss the idea that, although these anomalies could have been the result of genetic mutations, endogamy and exposition to ecotoxic environments could also be at play within the Chinchorro groups.

1. Introduction

Polydactyly, is a congenital malformation featuring supernumerary or bifid digits in hands or feet. It is most common in modern African populations, followed by Native Americans, Latin Americans (mainly mestizos), and Caucasians. This phenomenon, however, has been rarely observed in bioarchaeological reports for pre-Columbian populations. In contrast, representations of polydactyly in various visual media are common throughout the Americas. Thus, it is worth considering why the low biological frequency of extra fingers and toes contrasts with the more frequent depiction in rock art. Is polydactyly in rock art meant to represent real individuals or just depict a rare anatomical condition whose bioarchaeological remains have not been identified?

We searched for bioarchaeological and visual representations of polydactyly among pre-Columbian Chinchorro groups of fishers, hunters, and gatherers who inhabited the hyperarid Pacific coast of the Atacama Desert. We try to understand the probable causes of the genetic mutation that resulted in polydactyly in these Archaic Late period populations (ca. 4000 BP). Although polydactyly could have occurred as a result of a spontaneous and random genetic mutation, as the case study corresponds to rather small populations, we explored two hypotheses as possible causes of this anomaly: (a) environmental or ecotoxity derived from the chronic contamination of heavy metalloids (i.e. arsenic) in river and spring waters in northern Chile and (b) consanguinity resulting from endogamic behaviors between Chinchorro groups.

Volcanism and hydrothermal activity of the Andean mountains yields water with elevated salinity and high concentrations of heavy metalloids (i.e arsenic, lead, boron, among others) that create ecotoxic environments (Figueroa, 2001). Water drains from the western slope of the Andes carrying As and other metalloids that contaminate aquifers and spring water all the way out to the Pacific littoral zone. Coastal human populations in northern Chile who lived in this ecosystem consumed water with high concentrations of these nonmetallic minerals (Arriaza et al., 2010; Swift et al., 2015). The consumption of water contaminated with heavy metalloids (especially As) may cause chromosomal alterations resulting in teratogenic anomalies during embryonic development (Zhang et al., 2005). The Chinchorro people were dramatically affected by arseniasis for thousands of years (Arriaza et al., 2010; Boston and Arriaza, 2009; Figueroa, 2001; Swift et al., 2015), which hypothetically could have played a role in the presence of the teratogenic condition.

Molecular DNA studies of ancient remains of the Chinchorro people and other pre-Hispanic northern Chilean coastal populations to

* Corresponding author.
E-mail addresses: vstanden@uta.cl (V.G. Standen), csantoro@uta.cl (C.M. Santoro), barriaza@uta.cl (B. Arriaza), dvalenzu@uta.cl (D. Valenzuela), dcoleman@unc.edu (D. Coleman).

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understand the degree of endogamy among these populations have not, to date, been performed. This is due to the degradation of DNA in bioanthropological samples from the hyperarid environments of the Atacama Desert. Recent analyses of strontium (Sr) isotopes show that the sampled Chinchorro individuals were born and raised on the coast until they died (Standen et al., 2018). No foreign individuals were isotopically identified. Recent records of two cases of polydactyly in Andean camelids from a Huari culture site -El Castillo de Huarmey- on the northern coast of Peru have been interpreted as a consequence of low genetic diversity due to selective reproductive practices (Tomczyk and Giersz, 2017). In this context, we pose the question whether endogamy may have been part of the causal factors for polydactyly in the Chinchorro population.

1.1. The biology of polydactyly

Polydactyly is a common congenital malformation expressed by supernumerary or bifid fingers and toes (Malik, 2014). It is transmitted through autosomal dominant inheritance (Radhakrishna et al., 1997) and can occur as an isolated anomaly or in association with other malformations (e.g., Ellis-van Creveld or Rubinstein-Taybi syndromes, among others). Cases of polydactyly related to teratogenic factors, such as the maternal consumption of medication during gestation (e.g., valproic acid), have been reported (Aguinaga-Ríos et al., 2002; Buntinx, 1992; Clayton-Smith and Donnai, 1995). These teratogenic factors can cause chromosomal alterations that result in congenital malformations during embryonic development. Currently, supernumerary digits and toes are linked to “gene regulation and pattern formation in vertebrate limb evolution” (Lange and Müller, 2017).

According to the location of the duplicated digit, polydactyly is classified as preaxial when it affects the first digit of the hand (thumb) or foot (large toe) and postaxial when it affects the fifth digit of the hand (little finger) or foot (Temtamy, 1990). Postaxial polydactyly is classified as type A when an osseous structure is present and type B when there is only soft tissue forming a hint or remnant of the extra digit (Bingle and Niswander, 1975; Temtamy, 1990). Polydactyly type A displays a gradient of expressions ranging from the complete duplication of the fifth digit including the metacarpal to only the duplication of the distal phalange. Other variants can affect other fingers or toes, although they are less frequent (Phelps and Grogan, 1985).

1.2. Epidemiological studies of polydactyly in current populations, its heredity, and responsible genes

Epidemiological studies demonstrate that the frequency of polydactyly varies among populations (Bingle and Niswander, 1975; Castilla et al., 1973; Kromberg and Jenkins, 1982; Scott-Emuakpor and Madueke, 1976; Stevenson et al., 1966). The highest incidence of polydactyly is found in Africans (Stevenson et al., 1966). In particular, Scott-Emuakpor and Madueke (1976) report a prevalence of 22.7 per 1000 live births in Nigeria in West Africa. In Uganda, East Africa, a prevalence of 13.5 per 1000 live births is reported (Simpkiss and Lowe, 1961), and in South Africa, polydactyly shows a prevalence of 10.4 for every 1000 live births (Kromberg and Jenkins, 1982). The presence of polydactyly in African American communities in North America (12.8/1000 births) shows values similar to results from studies in African countries (Bingle and Niswander, 1975).

In Native Americans (North Americans) a relatively high frequency of polydactyly is also reported, although it is much lower than that of African or African American populations. A study by Bingle and Niswander (1975) shows a rate of 2.4 per 1000 live births among Native Americans and 1.32 per 1000 in Caucasian Americans. By contrast, in data collected by ECLAMC (Latin American Collaborative Study of Congenital Malformations) that included Argentina, Uruguay, and Chile (without specifying ethnic affiliation), the rate was 1.01 per 1000 live births (Castilla et al., 1973). ECLAMC’s more recent research broadened the study to nine South American countries and showed rates of polydactyly that fluctuated between 15.7 and 39.9 per 10,000 live births (Nazer and Cifuentes, 2011:74); the countries with the largest ancestral African population (Brazil, Colombia, and Venezuela) were those with the highest rates of polydactyly (Table 1). Another ECLAMC study specific for a sample of Chilean population (without ethnic affiliation) shows a rate of 1.3–1.9 per 1000 live births (Cifuentes et al., 2007). In sum, African populations have the highest rates of polydactyly, followed by Native Americans, Latin Americans (mainly mestizos), and Caucasians.

Table 1
Rates of polydactyly in South American countries per 10,000 live births (1995–2008).
(Data adapted from Nazer and Cifuentes 2011:74)

Country	Rate
Brazil	39.9
Colombia	36.3
Argentina	15.7
Bolivia	18.6
Venezuela	27.1
Paraguay	20.9
Chile	18.6
Ecuador	18.4
Uruguay	15.6

The difference in the rate of polydactyly between populations demonstrates the role of genetic variability factors in this anomaly. Moreover, genetic studies have demonstrated that polydactyly can be caused by the mutation of more than one gene, with variations in the long or short arm of chromosomes 7, 13, and 19 (Akarsu et al., 1997; Radhakrishna et al., 1997)

1.3. Bioarchaeological evidence versus material culture of polydactyly in pre-Columbian times

Although some pre-Columbian societies display material culture related to polydactyly (e.g., rock art, pottery figurines, sandals, lithic stelae), reported bioarchaeological cases are rare. An exception are those cases described for the North American southwest (Barnes, 1994; Case et al., 2006; Crown et al., 2016), one in Belize in Maya territory (Wrobel et al., 2012) and most recently, in the highlands of northern Peru (Titelbaum et al., 2017).

The scarcity of osteological records of polydactyly at a continental level (N = 8) (Fig. 1, Table 2) contrasts with rock engraving images, the primary medium used by pre-Columbian societies to depict this feature. In Chaco Canyon, naturalistic footprints and handprints with extra digits (Barnes, 1994; Case et al., 2006; Crown et al., 2016) are spread through a vast territorial extension in Utah, Arizona, New Mexico, and Colorado (Malotki and Weaver, 2002). In the Pueblo Bonito site in New Mexico (AD 900–1100), there are handprint and footprint impressions with six digits on the clay floors and walls of some houses (Crown et al., 2016). Additional cultural objects from other sites include sandals widened in the toe area and effigies modeled in clay with six-toed feet (Crown et al., 2016).

In Mesoamerica, human representations with six-digit hands and feet are identified in lithic sculptures from Palenque (AD 400–800), clay modeled figures from Teotihuacan (Early Classic Period, AD 300–500), and Zapoteca ceramics (Late Classic Period, AD 500–700) (Wrobel et al., 2012). By contrast, there is only one reported osteological case of polydactyly, which came from the Peligroso Maya site in Belize (Wrobel et al., 2012). The iconographic evidence, however, demonstrates that polydactyly did not go unnoticed among the pre-Columbian populations of southwest North America and Mesoamerica. Rather, the depictions of extra digits in rock art, stelae, and ceramic figures suggest