

On engagement with Anthropology: A critical evaluation of skeletal and developmental abnormalities in the Atacama preterm baby and issues of forensic and bioarchaeological research ethics. Response to Bhattacharya et al. “Whole-genome sequencing of Atacama skeleton shows novel mutations linked with dysplasia” in Genome Research

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Halcrow S, Killgrove, K., Robbins Schug, G., Huffer, D., Arriaza, B., Jungers, W., & Gunter, J. (2018). On engagement with Anthropology: A critical evaluation of skeletal and developmental abnormalities in the Atacama preterm baby and issues of forensic and bioarchaeological research ethics. Response to Bhattacharya et al. “Whole-genome sequencing of Atacama skeleton shows novel mutations linked with dysplasia” in Genome Research. *International Journal of Paleopathology*, 22, 97–100. <https://doi.org/10.1016/j.ijpp.2018.06.007>

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Abstract:

Here we evaluate Bhattacharya et al.’s (2018) recent paper “Whole-genome sequencing of Atacama skeleton shows novel mutations linked with dysplasia” published in *Genome Research*. In this short report, we examine the hypothesis that the so-called “Atacama skeleton” has skeletal abnormalities indicative of dysplasia, critique the validity of the interpretations of disease based on genomic analyses, and comment on the ethics of research on this partially mummified human foetus. The current paper acts as a case study of the importance of using an anthropological approach for aDNA research on human remains. A critical evaluation of the ethically controversial paper by Bhattacharya et al. highlights how an understanding of skeletal biological processes, including normal and abnormal growth and development, taphonomic processes, environmental context, and close attention to ethical issues of dealing with human remains, is vital to scientific interpretations. To this end, close collaboration with palaeopathologists and local archaeologists through appropriate peer-reviewed journals will add to the rigour of scientific interpretation and circumvent misinterpretation.

Keywords: Ata | Genomic analyses | Foetal skeletal development | Skeletal dysplasia | Research ethics | Chile

Article:

*****Note: Full text of article below**



Brief Communication

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ARTICLE INFO

Keywords:

Ata
Genomic analyses
Foetal skeletal development
Skeletal dysplasia
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ABSTRACT

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1. Introduction

Judging by the sheer amount of press that human skeletons have received in recent years, it is clear that skeletal analysis speaks to many people, easily capturing the public's attention with its potential to help us understand individual lives in the past. Although educating the public about ancient life courses is a goal that we share, the media blitz in early 2018 following the publication of Bhattacharya et al.'s article in *Genome Research* is a prime example of how research that is not rigorous, analytically sound, or performed by appropriately trained researchers can spread misinformation. Further, studies such as these

that do not address ethical considerations of the deceased and their descendant communities threaten to undo the decades of work anthropologists and others have put in to correct past colonialist tendencies. When human skeletal studies that flout standard conventions of science are published, it is imperative for us to demonstrate how collaborative efforts in the analysis and interpretation of remains can counteract incorrect and problematic scientific narratives.

In this brief commentary, we use the Bhattacharya et al. article as an example of the kind of problematic research from which we can learn the importance of taking a holistic perspective in science. Drawing on scientific analytical techniques using human

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<https://doi.org/10.1016/j.ijpp.2018.06.007>

Received 30 March 2018; Received in revised form 28 June 2018; Accepted 28 June 2018

Available online 17 July 2018

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developmental osteology standards, comparative foetal osteological material, and paediatric genetic syndrome literature, we begin by outlining our concerns with the analysis of the age-at-death and ‘abnormalities’ in Ata’s skeletal remains and with the flawed scientific rationale to conduct genomic analysis. We then bring attention to essential ethical concerns and conclude with suggestions for how to engage in rigorous scientific research using human remains.

2. Critical commentary

2.1. Ata’s skeletal morphology

Bhattacharya and colleagues (2018: 1) state in their abstract that the Atacama “specimen carried a strange phenotype – 6-in[ch] stature, fewer than expected ribs, elongated cranium, and accelerated bone age.” The original assessment of the skeleton, however, was never published in a peer-reviewed journal, finding a public audience in *Science Magazine* (Stone, 2013). In their Supplemental Note to their 2018 article, Bhattacharya and colleagues say that the “morphological features include that the specimen has only 10 ribs, mild mid-face hypoplasia, and shows abnormalities of the skull. [...] As represented by a specialist in pediatric human bone and growth disorders, the 6-inch specimen is a human that was likely 6–8 years of age at the time of death (age based on epiphyseal plate X-ray density standards). [...] The specimen was concluded by the medical specialist to be a human child with an apparently severe form of dwarfism and other anomalies.”

As experts in human anatomy and skeletal development, we find no evidence for any of the skeletal anomalies claimed by the authors. Their observations of ‘anomalies’ represent normal skeletal development in the foetus, cranial moulding from delivery, and potential post-mortem taphonomic effects. Specifically:

- 1 Bhattacharya et al. claim the skeleton demonstrates “precocious epiphyseal ossification” (2018: 1) and “was possibly 6–8 yr at the time of demise” (2018: 6). They provide no evidence in the paper to support this claim. Based on the long bone (diaphyseal) lengths published in Gabilondo (2007) of a femur (20 mm) and clavicle (15 mm), we can estimate that this baby died at approximately 15 weeks gestational age (Cunningham et al., 2016). Further, if we accept the 6-inch crown-heel length reported for the Ata specimen as accurate, this also allows us to estimate gestational age at 15 weeks (Archie et al., 2006); however, there may be some reduction of length of the skeleton from desiccation.
- 2 Oblique reference is also made to the *Science Magazine* article (Stone, 2013) in which Nolan noted that Ata was 6–8 years of age-at-death based on an epiphyseal plate density test, a claim repeated in Bhattacharya and colleagues’ (2018) Supplemental Note. The actual methods for reaching this conclusion are not specified, nor is the applicability of the method on desiccated tissue explained. Based on Bhattacharya and colleagues’ (2018) Figure 1, there is no evidence for phenotypic abnormalities in any of the long bones (Baker et al., 2005).
- 3 The authors note (2018: 1) that “after examining the X-ray images, it is concluded that Ata had only 10 pairs of ribs instead of the normal 12 in humans.” The 11th and 12th ribs may not be observable as they are smaller, shorter, ‘floating’ ribs that do not articulate anteriorly at the sternum and are not as robust. There is little information about the formation of ribs *in utero*, but Scheuer and Black (2000: 238) state that “by the eleventh and twelfth weeks of intrauterine life, each rib (often with the exception of the twelfth)” has started to form, which implies that the lower ribs are later forming. All ribs that are visible in the Ata specimen have normal morphology. Interestingly, the clinical literature (e.g., Calder and Offiah, 2015: 539) acknowledges the potential for misdiagnosis of skeletal dysplasia due to normal lack of ossification in early gestation foetuses. This misdiagnosis seems to be the case in

the paper in question.

- 4 Bhattacharya et al. (2018: 1) also argue that the baby has an “elongated cranium.” Although the cranium does appear to be longer than it is wide, this can be better explained in terms of both taphonomic and birth processes. It is common for a process called plastic deformation to alter the shape of cranial remains that have been interred in the ground, where heat and pressure can slowly affect their shape (McPherson and Kriewall, 1980). Additionally, a foetus of this age does not have the same cranial proportions of a full-term foetus (Calder and Offiah, 2015; Campbell and Newman, 1971). Furthermore, during delivery, the relationships between the cranial bones may be altered from compression of the bones in the cervix in a process referred to as moulding. Such moulding can reduce the skull diameter, resulting in an elongated appearance; this has been shown to be more severe in preterm foetuses (McPherson and Kriewall, 1980). Based on the photos provided, the frontal and parietal bones of the Atacama baby indeed show significant moulding; the parietals are compressed, and the superior part of the left parietal bone is passing over the right parietal at the midsagittal suture. Lifting of the parietal bones is often reported in obstetric and paediatric literature (McPherson and Kriewall, 1980; Lapeer and Prager, 2001). The “elongated cranium” of Ata is therefore phenotypically normal for a preterm foetus that has been delivered.
- 5 The authors state that they have identified known mutations in genes associated with cranioectodermal dysplasia and Greenberg skeletal dysplasia (Bhattacharya et al., 2018: 5), both of which they assert may have produced Ata’s supposed phenotype: the inferred cranial dysplasia, the claim that the foetus demonstrates “accelerated bone age” (2018: 1), a “premature ossification phenotype” (2018: 6), and “was possibly 6–8 yr at the time of demise” (2018: 1). Cranioectodermal dysplasia (Sensenbrenner syndrome) is a rare multiple anomaly syndrome with distinctive skeletal changes including craniofacial findings (e.g., forehead bossing, dolichocephaly), and metaphyseal dysplasia (e.g., short limbs, small thorax) (Lin et al., 2013), and Greenberg skeletal dysplasia causes punctate calcification of cartilage and asymmetrical shortening of long bones (Offiah et al., 2003). Given that there is no skeletal evidence for any of these conditions in the Atacama foetus, the basis for this conclusion is questionable.

Taken together, none of the methods or findings regarding Ata’s skeletal age presented by Bhattacharya and colleagues meet the accepted standards for age estimation using bioarchaeological, forensic, or paediatric/obstetric techniques. One of us (WJ) raised these concerns some years ago, saying that “genetic anomalies aren’t evident, probably because there aren’t any” (quoted in Stone, 2013).

2.2. Genomic data interpretation

We also want to comment on the genomic results in the Bhattacharya paper, as we are sceptical that the genomic results support morphological anomalies that are not actually present. Although we concede that only one of us (MK) is a specialist in human genomics, we have serious misgivings about the interpretation of the genomic analysis. Specifically:

- 1 According to the authors (2018: 6), the specific variants they have identified are “associated with scoliosis (COL1A1, FLNB, COL2A1, PMP22), Ehlers-Danlos syndrome (COL1A1, FLNB, COL2A1, PMP22), and musculoskeletal abnormalities (COL2A1, WDR65, ASPM, PMP22, FLNB).” We question why the authors have used missense variants in the COL1A1 and COL2A1 genes (rs575285203 and rs768451951) as evidence of a predisposition to dysplasia. These genes provide instructions for making type I collagen; the specific variant found in the COL1A1 gene could possibly influence the development of Ehlers-Danlos syndrome, Caffey’s disease

(infantile cortical hyperostosis), or osteogenesis imperfecta because there are variants in this gene that do cause these diseases. However, the specific variant they found is of uncertain significance in terms of the role it plays in any of these conditions according to a search for rs575285203 in the NCBI search engine under ClinVar. A search of ClinVar for rs768451951 returned no results. In addition, even if the mutation found in the COL1A1 gene did result in Ehlers-Danlos syndrome, it would most likely be the arthrochalasia type, which is associated with hypermobility of joints and potentially encourages the development of hip dysplasia. Neither of these conditions was cited in the osteological evidence for “abnormalities” that prompted this analysis. Furthermore, none of these conditions—Ehler’s-Danlos, Caffey’s, or Osteogenesis Imperfecta—would result in observable phenotypic changes in a foetus of this age.

- 2 The variants the authors found in FLNB, KMT2D, TRIP11, and ATR are novel, and thus their role in the development of skeletal dysplasias is unknown. The rs2070426 variant in PCNT has no demonstrated clinical significance and is classed as a “benign allele” (again, according to a search of NCBI’s search engine under ClinVar for rs2070426). The authors admit that their speculation that the variants would be deleterious is based on a prediction algorithm; however, they immediately follow that statement with a discussion in their paper about how other dominant mutations in these genes can cause dysplastic disorders (Bhattacharya et al., 2018: 5). This discussion is not clearly related to the specific genetic variations reported for the Atacama skeleton. Given there are no skeletal abnormalities and the vast majority of protein substitutions are neutral, the presence of these mutations in the Atacama skeleton’s genome may be genuine, but it is unlikely they had phenotypic consequences, and it is possible they were coincidental.
- 3 The authors use a “negative control,” in which they selected at random a South American individual and looked for deleterious or disease-causing mutations. Their intention, we presume, is to show that the specific mutations in the Atacama foetus are not normal for the South American population. Unsurprisingly, the negative control individual does not show the mutations found in Ata. This individual had other mutations in genes that could potentially predispose them to medical conditions including: amnesia; glandular and epithelial neoplasms; malnutrition; sleep disorders; heart diseases; nonsyndromic deafness; renal diabetes; nerve degeneration; and neurodegenerative diseases (Supplementary Table S7). Rather than bolstering their interpretation of the genetic data, this result demonstrates it is possible to find a similar number of variants as identified in the Atacama foetus in any given human and that these may be associated with a variety of disorders that will never be expressed in the phenotype. Given there is no evidence for skeletal dysplasia, the genomic data do not provide conclusive results that warrant the destructive analysis.
- 4 Bhattacharya and colleagues (2018: 6) write that “although we can only speculate as to the cause for multiple mutations in Ata’s genome, the specimen was found in La Noria, one of the Atacama Desert’s many abandoned nitrate mining towns, which suggests a possible role for prenatal nitrate exposure leading to DNA damage (Andreassi et al. 2001).” We do not know the date of the specimen and therefore cannot speculate whether the authors’ argument that exposure to toxins from nitrate mining was a root cause of genetic mutation. Furthermore, this claim was not tested by this analysis.

The authors draw the following major conclusions (2018: 6): “Taken together, it is entirely plausible that the chance combination of multiple known mutations and novel SNVs identified here may explain Ata’s small stature, inappropriate rib count, abnormal cranial features, and perceived advanced bone age. Given the size of the specimen and the severity of the mutations described above, it seems likely the specimen was a preterm birth.” In our view, it is most likely a coincidence that the authors found this individual had some mutations in genes that are

associated with a predisposition toward dysplasia because: 1) the impetus for their analysis was based on a misinterpretation of the skeletal morphology; 2) the specific variants they discovered have no known functional effect on skeletal morphology at this age; and 3) other variants they found are novel with unknown significance.

3. Archaeological legislation and research ethics

The antiquity of this preterm baby remains unclear. Bhattacharya and colleagues (2018: 2) write that “taken together with the DNA damage results above, this indicates that the Ata DNA was relatively free of DNA damage and contaminants. Moreover, the average DNA fragment size for Ata is ~300 bp which, based on a DNA-decay model (Allentoft et al. 2012), is consistent with a sample younger than 500 yr.” Elsewhere, one of the researchers, Garry Nolan, suggested that Ata was only a few decades old (Stone, 2013). Scientific study of this human is therefore bound by either archaeological or forensic ethics. Further, the context in which it was found is unclear, apart from reports that it was discovered in a grave next to a church in the abandoned town of La Noria in the Atacama Desert in Northern Chile (Dorador and Harrod, 2018). Dorador and Harrod (2018) aptly lay out the legal and ethical issues with respect to analysis of this relatively recent human foetus from Chile. They note that, “like many other countries, human remains and historical objects are protected by law in Chile, including the girl from La Noria. This protection comes from legislation passed in 1970 (N°17288) that protects National Monuments.” Further, Dorador and Harrod (2018) explain that “the DNA used for genome sequencing came from destructive sampling of some of the girl’s bones. As such, her body was damaged, illegal under Article N°38 of the law. Moreover, this regulation states that any study by foreign research groups using Chilean materials covered under this law must include Chilean researchers; no Chilean authors are included in the article (Law N°17288: Articles 22 and 23).”

Irrespective of the time period, the research and publication of data from this specimen does not follow current ethical standards in anthropology (e.g., Kintigh, 1996; COPE, 1997; AAPA, 2003; Turner, 2004; Bardill et al., 2018). There are no human ethical consents, nor archaeological permits cited by Bhattacharya and colleagues, despite the fact that one or both of these are necessary. Further, this mummified human foetus was sold to the current “owner,” which is also illegal under Chilean Law No. 17288. This aspect of its ownership history by itself places Ata within a global, complex trade in numerous categories of human remains, conducted both on- and off-line (Huffer and Chappell, 2014; Huffer and Graham, 2017). Additional human ethics issues therefore occur with the trade of these human remains on the “red market” (*sensu* Carney, 2011). *Genome Research* does not currently require an ethical statement of research with submission of research manuscripts, according to the journal editor (Zimmer, 2018). Whether or not a required ethics statement would have flagged the issues with the Bhattacharya paper prior to publication, however, is unclear.

Finally, there is an ethical question of whether the ends justify the means; that is, whether a study undertaken without appropriate ethical or legal considerations substantially addressed an important anthropological, medical, or genomics research question. We have shown that there is nothing to suggest that Ata had any skeletal abnormalities. On the basis of incorrectly perceived phenotypic anomalies and an incorrect age-at-death estimate, Nolan and colleagues undertook a DNA analysis in 2013 and unsurprisingly confirmed the mummy was human. Although this testing was not *sensu stricto* necessary, once her humanity was confirmed, analysis should have stopped and her body should have been repatriated to Chile. Had these researchers involved, from the beginning, a biological anthropologist who specialises in human remains, we are certain that ethical concerns would have been raised regarding the potentially living relatives of Ata (Dorador and Harrod, 2018) and the illegal removal of the mummy from Chile. We therefore cannot conclude that the ends justify the means. In the end, even the

novel genetic variations discovered in Ata's genome are of uncertain significance.

4. Conclusion

Close collaboration with archaeologists and/or palaeopathologists is a vital part of informed scientific research on human remains from the past. A nuanced understanding of skeletal biological processes and environmental context is essential for accurate scientific interpretation and for acting as a check on the ethics and legality of such research. Unfortunately, there was no scientific rationale to undertake genomic analyses of Ata because the skeleton is normal, the identified genetic mutations are possibly coincidental, and none of them are known to be strongly associated with skeletal dysplasias that would affect the phenotype at this young age. We caution DNA researchers about getting involved in cases that lack clear context and legality, or where the remains have resided in private collections. In the case of Ata, costly and time-consuming scientific testing using whole genome techniques was unnecessary and unethical.

Legislation

Chilean Government Law N° 17.288 of National Monuments. 1925 (1970). <https://www.leychile.cl/Navegar?idNorma=28892>

Notes

Two of us (SH and KK) contacted the editor of *Genome Research* to ask about submitting a response to the article and research in question. We were both told that *Genome Research* does not publish letters to the editor, only original research papers, despite senior authors Nolan and Butte's (2018) later response statement in which they attempt to justify the ethics of their analyses. For the scientific process to advance it is essential to have *open* debate through peer-reviewed journals.

Acknowledgement

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

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