International Journal of Paleopathology 24 (2019) 41–47

Contents lists available at ScienceDirect

International Journal of Paleopathology

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

A Possible case of Facio-Auriculo-Vertebral sequence (FAVs) in an adult female from medieval Iceland (13th–16th Century)

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ARTICLE INFO

Keywords:
FAVs
Facio-Auriculo-Vertebral sequence
Congenital
Medieval
Iceland
Disability

ABSTRACT

Objective: This paper presents a possible case of Facio-Auriculo-Vertebral sequence (FAVs) in an adult female from Haffjarðarey, Western Iceland (1200–1563 CE) and a brief review of associated terminology.
Materials: The skeletal remains of a single adult female (HFE-A-34, 18–24 years old), excavated in 1945 by the National Museum of Iceland.
Methods: We carried out macroscopic examination of the cranium and mandible in 2017.
Results: Right side unilateral asymmetric craniofacial dysplasia was identified on the cranium and mandible of HFE-A-34.
Conclusions: This individual presents with anomalous craniofacial asymmetry consistent with a clinical diagnosis of FAVs.
Significance: This paper offers a visually distinct case of an under-represented and under-documented congenital condition for future identification within paleopathology.
Limitations: Infra-cranial skeletal manifestations of FAVs would strengthen this possible diagnosis, but at this time it is not possible to definitively link the cranium and mandible of HFE-A-34 to any of the infra-cranial remains excavated from Haffjarðarey.
Suggestions for further research: In addition to further clarifying the variable nature of FAVs in archaeological remains, a detailed discussion of disability and the perception of disabled individuals within the medieval North Atlantic is necessary in order to understand the lived experiences of affected individuals.

1. Introduction

Facio-Auriculo-Vertebral sequence (FAVs) is a highly variable spectrum of craniofacial and skeletal asymmetries with accompanying organ, central nervous system, and/or sensory defects. It is typically unilateral but may be bilateral (Pillay et al., 2003). In the clinical and paleopathological literature, FAVs has been interchangeably referred to as Oculo-Auriculo-Vertebral spectrum (OAVs), Goldenhar syndrome, first and second branchial arch syndrome, and hemifacial microsomia (Shprintzen et al., 1980; Reddy et al., 2005; D'Alessandro et al., 2006; Muñoz-Pedroza and Arenas-Sordo, 2013). These conditions, however, may have distinct underlying causes.

Skeletal features of FAVs include variable craniofacial asymmetric dysplasia[1] of the maxilla and mandible as well as of the cervical vertebrae, and sacrum (Shprintzen et al., 1980; Wulfsberg and Grigsby, 1990; Pillay et al., 2003; Reddy et al., 2005; Dabir and Morrison, 2006; Beleza-Meireles et al., 2014). In clinical studies, the skeletal asymmetry of FAVs may also be accompanied by hearing loss and/or ocular abnormalities, as well as cardiac, pulmonary, renal, and/or central nervous system anomalies (Wulfsberg and Grigsby, 1990; Pillay et al., 2003; Reddy et al., 2005; Dabir and Morrison, 2006; D'Alessandro et al., 2006; Beleza-Meireles et al., 2014).

In the paleopathological literature, information on FAVs and its associated features is extremely limited (Castro et al., 1997; Nagar and Arensburg, 2000; Pachajoa et al., 2010; Panzer et al., 2008; Viciano and D'Anastasio, 2018). This paucity of information and identification may stem from the variable phenotypic spectrum, which ranges from barely detectable skeletal asymmetry to severe unilateral craniofacial

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[1] Although there are disagreements over the application of the term 'dysplasia' to the skeletal manifestations of OAVs and FAVs (Cohen et al., 1989), it is used here to simply refer to the abnormal development of bone.

https://doi.org/10.1016/j.ijpp.2018.08.009
Received 8 June 2018; Received in revised form 21 August 2018; Accepted 22 August 2018

hypoplasia (Beleza-Meireles et al., 2014). This is further complicated by the varying names used for the same phenotypic spectrum (OAVs, Goldenhar syndrome, and hemifacial microsomia) (Cohen et al., 1989). Publications of related congenital conditions such as aural atresia (Hrdlička, 1933; Hodges et al., 1990; Masnicová and Benuš, 2001; Keenleyside, 2011; Swanston et al., 2013; van Duijvenbode et al., 2015) and hemifacial microsomia (Ginestà et al., 2013) associated with OAVs (Viciano and D'Anastasio, 2018) are more common in the paleopathological literature than are FAVs. It is therefore necessary to define FAVs, OAVs, Goldenhar syndrome and hemifacial microsomia as well as their distinct skeletal manifestations prior to the presentation of this case study.

In the 1950s, Dr. Maurice Goldenhar described a variable pattern of skeletal and soft tissue abnormalities, including asymmetrical craniofacial dysplasia, epibulbar dermoids (or choristomas), preauricular appendages, and pretragal fistulas, now known as Goldenhar syndrome (Baum and Feingold, 1973; Shprintzen et al., 1980; D'Alessandro et al., 2006). In 1963 the features of this condition were expanded to include associated anomalies of the kidneys, heart, and central nervous system under the new nomenclature of OAVs (Gorlin et al., 1963). OAVs characteristics include "asymmetric ear anomalies…with or without hearing loss…hemifacial microsomia resulting in facial asymmetry; orofacial clefts; ocular defects…and vertebral abnormalities" (Beleza-Meireles et al., 2014). Many of these features also characterize FAVs, with the specific exception of epibulbar dermoids and choristomas, which are benign soft tissue ocular tumors or ocular skin tags and represent a requisite for a diagnosis of OAVs/Goldenhar syndrome (D'Alessandro et al., 2006).

Clinically, OAVs and/or Goldenhar syndrome are considered to be a subgroup of FAVs, featuring ocular-specific abnormalities not present in FAVs generally (D'Alessandro et al., 2006). Therefore, in paleopathology, when there is no soft tissue preservation that would allow for the confirmation of these ocular abnormalities, it is more accurate to consider a diagnosis of FAVs rather than OAVs/Goldenhar syndrome.

The prevalence of FAVs (together with OAVs) in modern populations is 1 in 3500 live births, with a male to female ratio of 3:2 (Reddy et al., 2005; Beleza-Meireles et al., 2014). FAVs is etiologically heterogeneous and has been associated with prenatal exposures to retinoic acid and primidone, as well as maternal diabetes (Pillay et al., 2003), although many cases are unrelated to teratogenic exposures (Reddy et al., 2005; Dabir and Morrison, 2006; Lakshman et al., 2017). That more than a single case of FAVs occurs in some families suggests a potential genetic predisposition (D'Alessandro et al., 2006), although no specific gene has yet been identified (Haratz et al., 2011).

This paper presents a visually distinctive case of possible Facio-Auriculo-Vertebral sequence (FAVs) in a medieval community. A better understanding of this condition and its variable phenotypic manifestations contributes to the accurate identification of FAVs and OAVs in paleopathology and contributes to on-going discussion of social and physical disability within past communities.

2. Materials and methods

2.1. Haffjarðarey

The cranium and mandible labelled HFE-A-34 were collected in 1945 by Kristján Eldjárn and Jon Steffensen during excavations on the island of Haffjarðarey by the National Museum of Iceland. Haffjarðarey is a small island (less than 0.25km^2 in area) off the western coast of Iceland, just south of the Snaefellsnes Peninsula and north of modern Borgarnes (Fig. 1). Historical records place a Catholic church dedicated to St. Nicholas and an associated cemetery on the island from approximately 1200–1563 CE (Níelsson, 1869; Sigurðsson and Þorkelsson, 1876; Þorkelsson, 1888), although use of the site probably began much earlier in the 11th century (Hoffman, 2018a). Use of both the church and cemetery ceased in 1563 after the effects of coastal erosion became increasingly problematic (Steffensen, 1946). The abandonment of the island church was later storied as the result of a catastrophe in which the last priest and all of the parishioners died on Christmas Eve (Kristjánsson, 1935). In reality, however, abandonment was likely driven by a combination of this coastal erosion and religious reformation (Steffensen, 1946; Hoffman, 2018a).

Human remains were removed from the site in three phases: first by local inhabitants when remains were exposed on the surface by erosion in 1886 (MNI unknown) (Steffensen, 1946), secondly by two American scientists working out of Harvard University in 1905 (MNI = 61) (Pálsson, 2005), and finally by an Icelandic archaeological team conducting a salvage excavation of the site in 1945 (MNI = 67) (Steffensen, 1946). At least one-hundred and twenty-eight (128) primary inhumations have been recovered from the site so far, making it the one of the largest cemeteries known for this time period in Iceland.

While the 1905 expedition did not record their collection process, the 1945 excavations revealed a densely populated cemetery with multiple instances of intercut, overlapping, disturbed, and vertically stacked burials (Eldjárn, 1945, handwritten notes; Steffensen, 1946). The individual discussed in this paper (HFE-A-34) was recovered during the 1945 excavation in association with the remains of at least two individuals (labelled HFE-A-6 and HFE-A-7 in Fig. 2). (Eldjárn, 1945, handwritten notes; Steffensen, 1946).

2.2. Age and sex determination

Analysis of the human skeletal remains from Haffjarðarey, including age-at-death, biological sex and macroscopic analysis of pathological lesions, was carried out at The National Museum of Iceland (Þjóðminjasafn Íslands) in 2017. Age-at-death for the collection was determined through macroscopic observation and recording of epiphyseal fusion, dental eruption, and scoring of the age-related morphological changes of the pubic symphysis and auricular surface of the *ossa coxae* (Brooks and Suchey, 1990; Buikstra and Ubelaker, 1997; Lovejoy et al., 1985; Meindl and Lovejoy, 1989; Ubelaker, 1989). Biological sex was determined through a combination of sexually dimorphic features of the cranium and mandible and *ossa coxae* (Acsádi and Nemeskéri, 1970; Buikstra and Ubelaker, 1997; Milner, 1992).

3. Results

3.1. HFE-A-34

Following analysis of the cranium and mandible, the remains were determined to belong to a young adult female between the ages of 18 and 24 (Hoffman, 2018b). The infra-cranial remains of HFE-A-6 and HFE-A-7, are considered to be in possible association with HFE-A-34 (Steffensen, 1946). Both sets of infra-cranial remains display features of right-sided skeletal asymmetry and were both determined to be within the same age and sex demographic as the cranium and mandible of HFE-A-34. As a result, it is not currently possible to definitively link the cranium and mandible with either set of infra-cranial remains.

3.2. The cranium and mandible

The cranium and mandible of HFE-A-34 are in very good condition, with only minor post-mortem damage of the right maxillary process of the maxilla, right lacrimal, and right and left orbital surfaces of the greater wings of the sphenoid. Several teeth were lost both ante-mortem and post-mortem. Maxillary teeth lost ante-mortem were the right incisors (RI1, RI2), canine (RC), first molar (RM1), and potentially the first premolar (RPM1). Mandibular teeth lost ante-mortem were the right incisors (RI1, RI2). The face is characterized by unilateral asymmetric craniofacial dysplasia affecting the right side (Fig. 3). Dysplasia of the right maxilla includes inferior displacement of the right nasal aperture to the level of the alveolar process and right lateral