International Journal of Paleopathology 7 (2014) 8–14

Contents lists available at ScienceDirect

International Journal of Paleopathology

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

Case Study

Ancient Down syndrome: An osteological case from Saint-Jean-des-Vignes, northeastern France, from the 5–6th century AD

Maïté Rivollat[a,*], Dominique Castex[a], Laurent Hauret[b], Anne-marie Tillier[a,c]

[a] Université de Bordeaux, UMR 5199 PACEA B8, Allée Geoffroy ST Hilaire, CS 50023, 33615 Pessac Cedex, France
[b] Service de Radiologie générale et pédiatrique,Clinique Tivoli, 33000 Bordeaux, France
[c] University of Pennsylvania Museum of Archaeology and Anthropology, USA

ARTICLE INFO

Article history:
Received 22 January 2014
Received in revised form 19 May 2014
Accepted 20 May 2014

Keywords:
Trisomy 21
Skull
Subadult
France
Early medieval period

ABSTRACT

The pathological skull of a 5–7 year old child from Saint-Jean-des-Vignes (Saône-et-Loire, north-eastern France) dated to the 5–6th century AD is described. Morphological and radiographic features, metrical data and Computed Tomography (CT) scans are used to study the osteological abnormalities in comparison with normal skulls of individuals of similar age and geographic origin. The combination of features is consistent with the diagnosis of Down syndrome (e.g. brachycrany, metopism, hypodontia, periodontitis, a flattened occiput, vault thinness, and an open cranial base angle), although none is pathognomonic of the disease in isolation. Cases of Down syndrome in past populations are rare, frequently poorly described or discovered out of context. This case represents the earliest and youngest example of the condition in the archaeological record. The context and funerary treatment of this child suggests that he/she was not stigmatized by other members of the community, who afforded a normal mode of burial.

1. Introduction

Down syndrome was first clinically described in 1866 by J.L. Down. The disease occurs due to a partially or complete third copy of chromosome 21 in 95% of cases (Lejeune et al., 1959). One of the main factors in the development of the condition is the age of the mother. The average birth rate for Down syndrome is 1 in 2500, but this increases to 1 in 600 in mother's over the age of 30 (Grmek, 1994). This disease might have affected ancient populations in approximately the same proportion, but Down syndrome is not well documented in human history. Archaeological cases are rare, with only two sub-adult cases recorded (Brothwell, 1960; Charlier, 2008; Czarnetzki, 1980; Walker et al., 1991). However, due to the number of dental and skeletal variants seen in Down syndrome, and the spectrum of disease severity, these cases are not without controversy. Interestingly, the existence of the disease in the past is illustrated in painting and sculpture that dates as early as 1500 BC (Starbuck, 2011).

This paper presents a potential archaeological case of Down syndrome in a 5–7 year old child from early medieval France (Castex, 1994; Castex et al., 2009). The morphometric skull features were compared to those of a sample of skulls from healthy archaeological and modern children of similar age to highlight abnormalities beyond normal population variation. Potential differential diagnoses were also considered and ruled out where possible.

2. Material and methods

The child skull under study was excavated in 1989 from a French necropolis dated to the 5–6th century AD. The necropolis was located to the south of the church of Saint-Jean-des-Vignes, Saône-et-Loire, in North-Eastern France and included 94 individuals. The child was buried supine in a single grave cut with three ceramic shards. Bone preservation was good, although most of the thoracic and lumbar vertebrae, and the right hand were missing. Unfortunately, the postcranial skeleton was lost after the excavation, and the only available evidence for these bones comes from field observations and a photograph taken in situ (Fig. 1). With the exception of the *Sella turcica*, the skull was complete and forms the focus of this investigation (Fig. 2).

Two earlier studies mentioned the possibility of Down syndrome for this specimen (Castex, 1994; Castex et al., 2009), based

* Corresponding author. Tel.: +33 5 40 00 25 51.
E-mail addresses: maite.rivollat@u-bordeaux.fr, maite87@live.fr (M. Rivollat), dominique.castex@u-bordeaux.fr (D. Castex), anne-marie.tillier@u-bordeaux.fr (A.-m. Tillier).

http://dx.doi.org/10.1016/j.ijpp.2014.05.004
1879-9817/

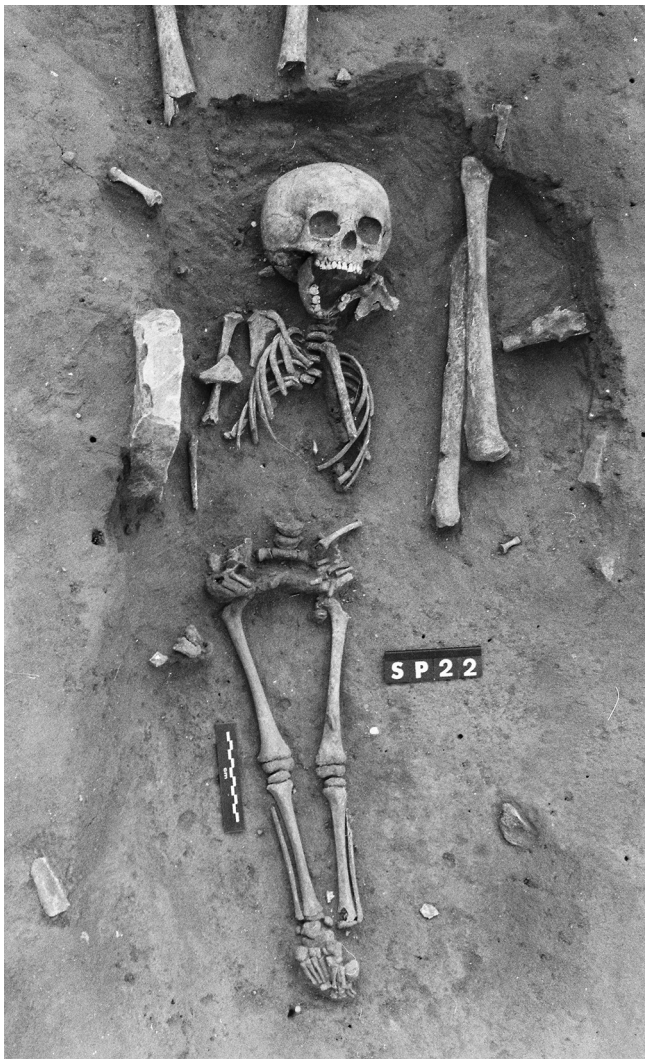

Fig. 1. In situ photograph of the Saint-Jean-des-Vignes individual (Cl. Afan, 1989).

upon the identification of shape of the cranial base, flattening of the occiput, brachycrany and thinness of vault bones. In the current study, the dimensions and morphology of the skull was compared with 78 skulls of children of similar age and backgrounds (Table 1). Due to the lack of postcranial bones, the age-at-death was estimated using dental criteria (Moorrees et al., 1963), maturation of the skull including closure of the Huschke foramen (Humphrey and Scheuer, 2006), and the intra-occipital synchondrosis (Coqueugniot and Le Minor, 2002).

Morphometric observations and comparisons were used to determine the Down syndrome diagnostic characteristics outlined in the extensive clinical literature (Table 2), using the Frankfurt plane as reference. In regard to pathological features, every anatomical part of the skull was studied independently. Radiographic and CT scan images allowed access to the internal features. A 3D reconstruction of the endocranial volume was made with Avizo v.7 software (Visualization Science Group Inc.). Bivariate and principal component statistical analyses were performed, as well as Student's *t*-test.

3. Results

Based on the development of the second deciduous molar (Res¼: 5–8 years old) and first permanent molar (R¼ left, R½ right: 4–6 years old), the dental age at death was estimated to be between

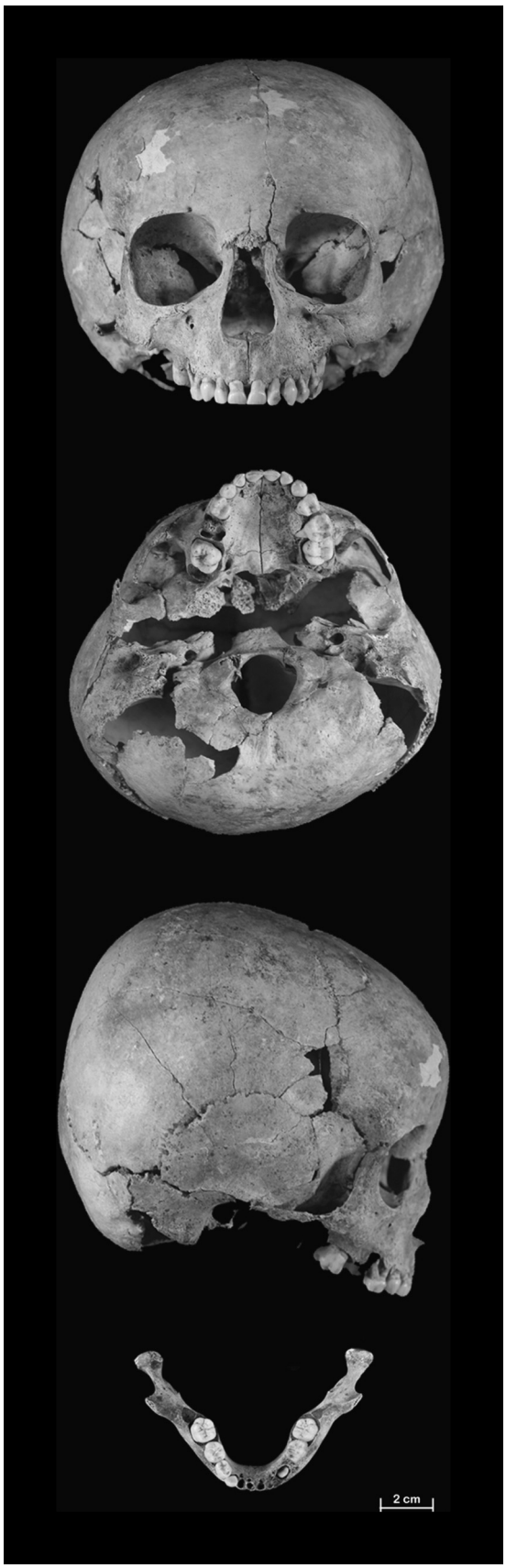

Fig. 2. Anterior, inferior and lateral view of the skull, and superior view of the mandible.