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Palaeopathological diagnosis of spondyloarthropathies: Insights from the biomedical literature

Mathilde Samsel*, Sacha Kacki, Sébastien Villotte

PACEA, UMR 5199, Anthropologie des Populations Passées et Présentes, Université de Bordeaux, Bâtiment B8, Allée Geoffroy St Hilaire, CS 50023, 33615 Pessac Cedex, France

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ABSTRACT

In palaeopathology, the diagnosis of spondyloarthropathies traditionally relies on the association of three types of skeletal lesions: erosive and proliferative modifications of the sacroiliac joint, formation of vertebral syndesmophytes and erosive and proliferative changes in peripheral joints. These conditions can therefore be recognised only in well-preserved skeletons that exhibit the most typical pattern of lesions. In order to develop additional criteria for the diagnosis of spondyloarthropathies, a literature survey was conducted as a preliminary step by comparing biomedical data with the palaeopathological literature. We point out musculoskeletal changes and localisations rarely, if ever, used for identification of spondyloarthropathies in skeletal material. Whereas a specific focus has been put on entheseal changes encountered in spondyloarthropathies, the results highlight skeletal changes that may contribute to the diagnosis of the spondyloarthropathies from osseous remains such as erosive lesions of the temporomandibular joint and erosive changes of entheses in the pectoral girdle. Recording of these lesions in future studies of archaeological samples would contribute to discussions of their diagnostic relevance.

1. Introduction

The spondyloarthropathies (SpAs) encompass a group of inflammatory rheumatic conditions, including ankylosing spondylitis (AS), psoriatic arthritis (PsA), reactive arthritis (ReA), inflammatory bowel diseases related to SpAs (IBD-SpAs) and undifferenciated spondyloarthropathies. The SAPHO syndrome (Synovitis, Acne, Pustulosis, Hyperostosis, Osteitis) is sometimes included in this group. However, as its status within SpAs is highly debated, it is not discussed in the present work (Sibilia et al., 2005; Rudwaleit et al., 2011; Zeidler and Amor, 2011). These disorders share common genetic predisposing factors (the strongest association being with HLA-B27), seronegativity for rheumatoid factor and clinical features including axial and peripheral inflammatory arthritis, enthesitis[1] and possible extra-articular manifestations (Moll et al., 1974; Dougados et al., 1991; Resnick, 2002a; Chandran et al., 2013). Males are affected preferentially in AS and ReA, whereas the sex ratio is equal to 1 for PsA and IBD-SpAs forms (Sibilia et al., 2005). The onset of early disease manifestations generally occurs in young adults (Sibilia et al., 2005). As stated by Stolwijk et al. (2012), large variations in the population incidence and prevalence rates of SpAs and their subtypes are seen in the literature. The geographic distribution of the haplotype HLA-B27, the way the target population was sampled and the criteria used to identify cases contribute to such differences. Overall, in Western European countries, SpAs have a prevalence rate between 0.3% and 2.5%. Undifferentiated spondyloarthropathies seem to represent almost 40% of these cases. AS and PsA display prevalence rates which seem quite similar in Western countries, up to 0.53%, with ReA between 0.09 and 1% and IBD-SpAs estimated at 0.01 to 0.5% (Stolwijk et al., 2012). In palaeopathology, one of the more recent epidemiological studies was performed by Martin-Dupont et al. (2006) on a 19th–20th century Portuguese collection. In light of the new set criteria developed by these authors, SpAs were definitively diagnosed for 6.7% of the individuals, as probable for other 6.3% and could not be excluded for another 19.4% of the subjects in this collection. However, using more 'parsimonious' classical criteria, other authors found lower prevalence rates for SpAs of between 1% and 3% in four Italian sites dated before the 15th century (Rothschild et al., 2011) and 2% in a 16th–18th centuries skeletal sample from a French convent cemetery (Villotte and Kacki, 2009).

Today, SpAs constitute a complex set of disorders with polymorphic expression (Sieper et al., 2009). Their diagnosis is based on

* Corresponding author. Tel.: +33 689473399.
E-mail address: mathilde.samsel@gmail.com (M. Samsel).

[1] 'Enthesitis' refers to an inflammatory condition of the enthesis; 'enthesopathy' is the generic term used to refer to any pathological condition of the enthesis. Terms used in the present work correspond to those found in the cited literature.

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classification criteria (e.g. Rudwaleit et al., 2009, 2011) that include various clinical features, with bony lesions representing only a few of them. One can postulate that SpAs also caused varying skeletal and extra-skeletal manifestations in past populations. However, as skeletal remains are generally the only material available for skeletal analysis, the studies performed so far in palaeopathology have mainly focused on the most characteristic skeletal changes of the diseases. The skeletal lesions considered to be suggestive of SpAs in osteological material and which constitute a consensus in palaeopathology have been summarised on one hand by Rothschild and Woods (1991) and Rothschild et al. (1999) and on the other by Rogers et al. (1985, 1987). Rothschild's criteria are (1) sacroiliac or zygoapophyseal joint erosion[2] or fusion by bone proliferation; (2) spinal ankylosis; (3) asymmetrical pattern of oligoarthritis; (4) peripheral joint fusion; (5) reactive new bone formation; (6) enthesopathies; and (7) lesions non exclusively ligamentous. Rogers' criteria are as follows: (1) symmetrical or asymmetrical sacroiliitis; (2) vertebral ossification/ankylosis; (3) frequent zygoapophyseal involvement; (4) asymmetric arthritis of the hip, knee, ankle, foot, shoulder and hand. Additionally, entheseal changes are sometimes discussed in greater detail as an aid to diagnosis (e.g. Rogers et al., 1985, 1987; Rothschild and Woods, 1991; Rothschild et al., 1999; Villotte and Kacki, 2009).

As highlighted by Rothschild and Woods (1991: p. 125), 'recognizing spondyloarthropathy in the absence of sacroiliac/spine involvement and distinguishing it from other erosive disorders (. . .) is the challenge'. It is a truism, however, that skeletons derived from archaeological contexts are not always well preserved, or even complete. Thus, in many cases, the state of preservation prevents the observation of one or several of the areas of interest. Diagnosis would therefore benefit from the development of supplementary criteria to permit differential diagnostic discussion even in such cases, in addition to the 'major' criteria traditionally used in palaeopathology. Here, we present an overview of the data from the biomedical and palaeopathological literature and provide a synthesis of the skeletal modifications likely to be encountered in SpAs. By comparing information from these two sources, we point out some musculoskeletal modifications reported in the clinical literature, but rarely, if ever, precisely described for diagnosis of SpAs in the palaeopathological literature.

2. Materials and methods

This literature survey was conducted primarily from September 2012 to January 2013 as part of a master's thesis project which was completed in January 2014. The overview of biomedical and palaeopathological sources, primarily of English and French origin, did not aim to be exhaustive but endeavoured to embrace the spectrum of features encountered in clinical data and detail the musculoskeletal modifications used by palaeopathologists to identify SpAs in osteological collections. A search of the clinical literature was performed using the search tools Google Scholar (http://scholar.google.com/) and PubMed (http://www.ncbi.nlm.nih.gov/pubmed) and specialised textbooks (e.g. Rothschild, 1982). Palaeopathological sources were searched via Google Scholar and through a review of the main palaeopathology textbooks (Rogers and Waldron, 1995; Aufderheide and Rodriguez-Martin, 1998; Ortner, 2003; Roberts and Manchester, 2005). No start date was set for searches of web databases. However, due to the shifts occurring on SpAs classification, diagnosis and nomenclature over time (for a summary, see for example Rostom et al., 2010; Wendling et al., 2010; Stolwijk et al., 2012), papers were not selected if they did not comply with classification criteria developed since the 1990s in the medical literature: Amor criteria for SpAs (Amor et al., 1990), ESSG criteria for SpAs (Dougados et al., 1991), ASAS axial SpAs criteria (Rudwaleit et al., 2009) and ASAS peripheral SpAs criteria (Rudwaleit et al., 2011). A set of English and French terms was predefined (Table 1).

Table 1
English and French search terms used in the study.

	English terms	French terms
Conditions	Spondyloarthropathy(ies)	spondylarthropathie(s)
	Spondylarthropathy(ies)	
	Spondylarthritides	
	Ankylosing spondylitis	spondylarthrite ankylosante
	Psoriatic arthritis	rhumatisme psoriasique
	Psoriatic arthropathy	
	Reiter syndrome	syndrome de Reiter
	Reactive arthritis	arthrite réactive
	SAPHO syndrome	syndrome SAPHO
	Inflammatory rheumatic diseases	maladies rhumatismales inflammatoires
	Polyarthropathy	polyarthropathie
Features	Spondyloarthritis	spondylarthrite
	Arthritis	arthrite
	Enthesitis	enthésite
	Enthesopathy(/ies)	enthésopathie(s)
	Inflammatory	enthésopathie(s)
	Enthesopathy(/ies)	inflammatoire(s)
	Entheseal changes	modifications enthésiques
	Fibrocartilaginous enthese	enthèse fibrocartilagineuse
	Sacroiliitis	sacro-iliite
	Syndesmophyte(s)	syndesmophyte(s)

The aim of this research was to be as complete as possible, but papers which dealt with medical treatments and specific pathogenesis were excluded in order to restrict the search to our topic (i.e. musculoskeletal features encountered in SpAs' patients). All forms of literature (fundamental research, case reports, skeletal studies and general palaeopathology textbooks) were, however, included. Alteration was selected if it was mentioned once at least, and more precisely as follows: in general palaeopathology and specialised textbooks as criterion or manifestation used for diagnosis; in case reports or skeletal studies as a feature described in a SpAs' case or used for the diagnosis of SpAs; as a criterion used to define SpAs in the differential diagnosis. No chronological periods or geographic areas were excluded from the palaeopathological studies selection. Only adult forms of SpAs were analysed in our study, although there are some forms of juvenile SpAs (e.g. Rothschild et al., 1997; Burgos-Vagas, 2012).

3. Results

Whereas the lesions affecting some skeletal areas are well-described in the palaeopathological literature (e.g. changes in the spine, the sacroiliac and the peripheral joints), others are only documented in biomedical sources. We analysed 32 documents from the clinical literature and 34 from the palaeopathological literature. All of the features described in the palaeopathological references were also noted in biomedical settings. Conversely, the survey indicates that 25 skeletal changes and/or localisations recorded in the biomedical literature have not previously been mentioned in palaeopathological studies (Fig. 1). Many of these affect the cranium or the pectoral girdle while others are located in the thorax, pelvic girdle and upper and lower limbs. In the following sub-sections, the skeletal changes and/or localisations are outlined by anatomical area. The overall results of the literature survey are presented in Appendix A as Supplementary Data.

[2] As used here, the term 'erosion' applies only to pathological lesions in or around joints that are characterised by a loss of cortical bone and the exposure of the underlying trabeculae, according Rogers and Waldron (1995: p. 12).