International Journal of Paleopathology 20 (2018) 12–19

Contents lists available at ScienceDirect

International Journal of Paleopathology

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

Invited commentary

How should we diagnose disease in palaeopathology? Some epistemological considerations

Simon Mays

Historic England, United Kingdom

ARTICLE INFO

Keywords:
Theory
Epistemology
Reference sample
Target sample
Pathophysiology
Skeletal lesion

ABSTRACT

This paper analyses some of the epistemological frameworks that underpin diagnosis in palaeopathology. Currently, the dominant approach is comparative: relationships between skeletal lesions and disease in a reference group in which there is independent evidence of the diseases present in individuals are used to identify disease in unknown archaeological skeletons on the basis of the lesions present. This is essentially a reference sample – target sample approach, analogous to that used to develop methodology in other areas of biological anthropology (e.g. age estimation in palaeodemography). As well as considerable strengths, this approach also has significant weaknesses. Many of these arise from the nature of the reference material (mainly pathology museum and other skeletal collections, and published collations of medical imaging data) used to develop diagnostic criteria. There may also be a tendency toward over-emphasis on pattern-matching between reference and target material, and an under-emphasis on developing our understanding of the biology of bone lesions. Despite its shortcomings, the comparative approach is likely to remain the foundation of most palaeopathological work, but we should increasingly augment it with other diagnostic approaches, especially those grounded in the pathophysiology of bony responses to disease.

1. Introduction

In palaeopathology, the diagnosis of disease is of central importance. In a skeletal population, it is a step toward understanding the nature of the disease burden in that group and toward the testing of biocultural hypotheses. These are core goals of the field. As a discipline, we are adept at evaluating and applying new laboratory methods – for example biomolecular techniques (Brown & Brown, 2011: 242–263) – to disease diagnosis. Important steps have been made toward rigour in use of nomenclature (Manchester et al., 2017), description of lesions (Buikstra and Ubelaker, 1994; Roberts, in press), and differential diagnosis (Ortner, 2012; Klaus, in press). However, higher level theoretical considerations relating to palaeopathology have not received similar attention. In particular, the epistemological[1] frameworks that underpin our work, and within which our identification of disease takes place, have tended to remain implicit, and the assumptions behind them unexamined. This work identifies some of the conceptual frameworks within which diagnosis in palaeopathology takes place. In particular, it describes the two most important approaches and critically considers their strengths and weaknesses. The paper is principally concerned with studies of skeletal remains, although some of the more general considerations may also have relevance for studies involving ancient soft tissue.

Palaeopathology originated as a discipline when workers began to bring medical knowledge to bear upon ancient bones (Armelagos, 1997), and the relationship with medical sciences has remained important (Ortner, 2011; Mays, 2012). As palaeopathologists, we need to be aware, not only of methodological developments in biomedical and other disciplines, but also of higher level theoretical debates. As regards diagnosis, some epistemological debates in clinical medicine have resonance for palaeopathology. These include: questions of the relationship between heuristic (intuitive) and formal analytical reasoning in diagnosis (Gøtzsche, 2007; Norman et al., 2007; Stolper et al., 2010; Hofmeijer, 2014; van Baalen and Boon, 2015; Mark and Wong, 2015); how different types of knowledge, for example, from pathophysiology, population-based empirical studies in the published scientific literature, and a practitioner's own experience and judgement, might best be combined to inform decision-making in individual cases (Macnaughten, 2004; Bowen, 2006; Norman et al., 2006); and the value of Bayesian reasoning to deal with diagnostic uncertainty (see below). Although it is important that we should be aware of such debates, their potential for direct cross-over to palaeopathology is limited by a number of key differences in approach. A physician may draw upon both clinical and paraclinical data in order to arrive at a diagnosis. Clinical data comprise

E-mail address: simon.mays@historicengland.org.uk.

[1] Concerned with the nature of knowledge.

https://doi.org/10.1016/j.ijpp.2017.10.006
Received 8 June 2017; Received in revised form 20 October 2017; Accepted 22 October 2017

symptoms reported by the patient, together with signs observed by the physician at examination. Paraclinical data include imaging studies and laboratory tests that the physician may, in the light of clinical data, decide to request (Gøtzsche, 2007: 4–7). The importance of clinical data means that the diagnostic process must be viewed as a human social practice (Sadegh-Zadeh, 2000; van Baalen and Boon, 2014) as well as a scientific endeavor. The humanistic element means that, in the philosophy of medicine, epistemic techniques from the humanities, as well as the sciences, assume importance (Solomon, 2008). Evidence contributing to diagnosis in palaeopathology is limited to paraclinical data: gross observation of remains plus a minor subset of the paraclinical data available to the physician (skeletal imaging, some biomolecular analyses). The lack of a humanistic element in palaeopathological diagnosis renders it a more purely scientific endeavor. Since the 1990s, the normative scientific paradigm in Western clinical practice has been evidence based medicine (EBM) (Evidence Based Medicine Working Group, 1992; Bingeman, 2016; Hanemaayer, 2016; Wieringa et al., 2017). In EBM[2] there is an emphasis on evaluation according to formal protocols of systematically-gathered evidence as a basis for clinical decision-making. Much of the focus in EBM has been on the evaluation of drug and other therapuetic interventions, with an emphasis on the desirability of randomised controlled trials. The impact of EBM upon the diagnostic process has been rather less, perhaps reflecting the latter's complex nature with heuristic and well as analytical reasoning being involved. In addition, and in contrast to palaeopathology, the time depth provided by the development of a disease in a patient means that clinical diagnosis may often be an iterative process, diagnosis being open to revision in the light of (*inter alia*) the patient's response to treatment regimes (Dinant and van Leeuwen, 2004; Willis et al., 2013; Kennedy, 2017).

Differences between modern clinical practice and palaeopathology in evidence base, methods and aims are clearly considerable. It is therefore suggested that, in order to mature as a discipline, palaeopathology needs to develop epistemological models that properly reflect its nature, rather than attempting to conform to those that were developed for other purposes in modern medical practice. Conceptual frameworks which appear to me to be descriptive of the way we work toward the identification of disease in palaeopathology comprise the following:

1 The comparative approach
2 The biological approach
3 Direct measurement of a diagnostic parameter
4 Direct identification of the causative microrganism (for infectious disease)

The first two pertain to lesion-based palaeopathology; that is, diagnosis based upon the morphology and distribution of lesions. Traditionally, this was the sole basis of palaeopathological diagnosis, at least as far as it used human remains rather than other sources of evidence, such as artistic depictions of disease (Grmek and Gourevitch, 1998). Recently, two non-lesion based approaches have augmented lesion-based studies. Firstly, direct measurement of bone mineral density, cortical thickness or quantitative aspects of bone microstructure are pivotal to studying osteoporosis (Curate, 2014). Secondly, DNA, and to a lesser extent other biomolecular markers, such as mycolic acids, as well as identification of parasite remains, have provided key evidence for identification of microorganisms responsible for infectious disease, enabling diagnosis where lesions are ambiguous or non-existent (e.g. Hershkovitz et al., 2008; Mitchell, 2015; Feldman et al., 2016). Although such methods are important, and will probably become increasingly so, the foundation of palaeopathology is likely to remain the study of skeletal lesions. The focus of the present paper is therefore on the first two frameworks.

2. The comparative approach to diagnosis

This involves study of skeletal alterations in reference material from cases where there is independent evidence concerning what disease(s) were present. The information thus gained on lesion morphology and distribution provides the basis for identification of diseases from lesions in unknown archaeological remains. This method of working from the 'known' to the 'unknown' links our work to clinical science. Indeed, it is the intuitively obvious way to proceed, and has underpinned palaeopathology since the dawn of the discipline (Jarcho, 1966; papers in Buikstra and Roberts, 2012). In essence, this is a reference sample − target sample approach, analogous in some ways to methodology in other areas of osteoarchaeology and forensic science. For example, in age at death estimation, knowledge about the age distribution of skeletal age indicator(s) in a reference group of known age is used to infer age at death in an unknown forensic or archaeological skeleton from the indicator states observed in that skeleton (Chamberlain, 2000). In palaeopathology, we are using knowledge about the relationship between 'disease indicators' (lesions) and disease in a reference sample to try and identify the disease present in an unknown skeleton from the lesions present. In age estimation, the relationship between age-indicator and age is imperfect; similarly, in palaeopathology there is a paucity of pathognomonic skeletal lesions: different diseases may produce similar lesions, and a given disease may produce a range of different skeletal alterations.

As well as considerable strengths, the comparative method in palaeopathology also has important weaknesses. To a great extent these are inherent in any reference sample/target sample methodology where the relationship between the indicator and the parameter of interest is imperfect. Compared to workers in palaeodemography, palaeopathologists have analysed these difficulties rather less. In the sections that follow, I will attempt to elucidate some of the pitfalls of the approach, drawing upon analogies from the field of palaeodemography as appropriate.

2.1. What are the reference samples in palaeopathology?

Gross examination of remains, together with radiographic or other imaging, are the dominant methods in palaeopathology (Grauer, 2008; Mays, 2008; Wanek et al., 2012). Therefore, although there are resources such as documented collections of histopathology slides (Spatola et al., 2012), the key types of reference collections for palaeopathologists are skeletal remains housed in medical museums or other institutions, and published collations of radiographic or other images made on living patients usually as part of diagnostic work-up. These types of reference data form the main basis for textbooks aimed at identification of disease in skeletal remains (e.g. Steinbock, 1976; Zimmerman and Kelley, 1982; Aufderheide and Rodriguez-Martin, 1998; Ortner, 2003). No reference skeletal material was gathered, nor imaging study conducted, with the express aim of facilitating palaeopathological diagnoses, and reference samples may contain biasses that potentially undermine their value for that purpose.

Ideally, the attributes that a palaeopathologist might wish a reference sample to have might include:

- Reliable disease diagnoses
- The full range of disease expression that we might potentially expect to encounter in archaeological skeletal remains
- Additional background information about individuals that would facilitate the identification of factors, other than what particular disease is present, that might potentially affect lesion expression.
- To consist of complete skeletons or full body imaging

No reference sample is likely to fulfil all the above criteria and many

[2] Also known as EBHC, Evidence-Based Health Care.