



CASE STUDY

Legg-Calvé-Perthes disease and slipped femoral capital epiphysis in the skeletal remains of the Mediaeval Necropolis of Santa Maria (Sintra, Portugal)

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ABSTRACT

This paper discusses the differential diagnosis of unusual and distinct pathological changes in two left femurs and one left pelvic bone fragment from the Necropolis of Santa Maria, Sintra (Portugal), dated from the 14th-17th centuries. The differential diagnosis of the lesions gave rise to several possible pathological conditions, namely, slipped femoral capital epiphysis, Legg-Calvé-Perthes disease, developmental dysplasia of the hip, and trauma. Various macroscopic and radiological aspects led us to consider slipped femoral capital epiphysis as the most probable diagnosis in one case and Legg-Calvé-Perthes disease in the other. Given the scarcity of reports of both conditions in the osteoarchaeological literature, the identification of new cases becomes important. In fact, this appears to be the first report of slipped femoral capital epiphysis in non-

identified Portuguese human skeletal remains and adds to the only two cases of Legg-Calvé-Perthes disease detected until now.

Keywords: Paleopathology; Femur; Acetabulum; Femoral head osteochondrosis; Hip developmental dysplasia.

RESUMO

Neste artigo é apresentado e discutido o diagnóstico diferencial das alterações patológicas invulgares verificadas em dois fémures e num fragmento de osso pélvico esquerdos, recuperados da Necrópole de Santa Maria, Sintra (Portugal), séculos XIV-XVII. O diagnóstico diferencial considerou várias condições patológicas, nomeadamente, a epifisiólise, a doença de Legg-Calvé-Perthes, a displasia de desenvolvimento da anca e o traumatismo. As análises macroscópica e radiológica das lesões apontam para a epifisiólise como o diagnóstico mais provável para o primeiro caso e a doença de Legg-Calvé-Perthes para o segundo. Dada a escassez de relatos de qualquer uma destas condições na literatura osteoarqueológica, torna-se importante a divulgação e a identificação de novos casos. De facto, até ao momento, este é o primeiro relato de epifisiólise e o terceiro de doença de Legg-Calvé-Perthes em restos esqueléticos humanos não identificados de Portugal.

Palavras-chave: Paleopatologia; Fémur; Acetábulo; Osteocondrose da cabeça femoral; Displasia de desenvolvimento da anca.

Introduction

The skeletal remains under analysis were recovered from the medieval necropolis of *Santa Maria*, in Sintra (around 24 km west of Lisbon, Portugal). The church of *Santa Maria do Arrabalde* was originally built in the 12th century AD. It was founded by D. Afonso Henriques after the conquest of Sintra from the Moors in 1147. The necropolis, located in the south side of the churchyard and dated from the 14th-17th centuries, was excavated between November 1982 and January 1983 under the direction of the archaeologists José Belezza Moreira and José Cardim Ribeiro from

the Cultural Services of the Municipality of Sintra (*Serviços Culturais da Câmara Municipal de Sintra*) (Moreira and Ribeiro, 1983).

The inhumations showed an east-west orientation, with their heads to the west, in accordance to the Christian belief in the resurrection of the soul (Barroca, 1987), with the exception for burial #2, which showed a north-south orientation. The majority of the 41 excavated graves was reused, accommodating more than one individual, with commingled non-articulated bones, making it impossible to identify a single

individual (Moreira and Ribeiro, 1983). The associated archaeological artefacts include ceramics, copper and iron pricks, copper rings, and coins [date range: reigns of D. Sancho II (1223-1247/8) and D. João V (1706-1750)] (Moreira and Ribeiro, 1983).

In January 1999, the human remains were sent to the Department of Anthropology (now incorporated in the Department of Life Sciences) at the University of Coimbra and, in 2000/2001, were analyzed by the two authors of the present study. The MNI (minimum number of individuals) obtained was 120 individuals, including males and females, adults and sub-adults (Umbelino and Wasterlain, 2000; 2001; Wasterlain and Umbelino, 2001). Among the disarticulated human bones were two femurs and a pelvic bone fragment with pathological changes.

The exuberant nature and rareness of the lesions as well as the scarcity of reports in the paleopathological literature led us to publish their differential diagnosis.

Materials and Methods

The bones were examined by gross inspection and radiographic analysis, using standard medical radiography procedures (GE Medical Systems equipment; Agfa Blue film; voltage: 30 kV; exposure: 70 mAs).

Case 1: femur and pelvic bone from grave 19

Inventory and state of preservation

The first case refers to a left femur (head, neck, proximal and middle portions of the shaft), from the grave #19, which presents an

exuberant macroscopic change on its proximal epiphysis. Because of the bone pathology, incompleteness and post mortem damage (lacking the greater trochanter and the distal end), it was not possible to apply sex-determination methods. Probably belonging to the same individual, a left pelvic bone fragment, specifically an acetabulum, was recovered from the same grave.

Description of the condition

The main macroscopic bone changes consist of bone formation at the junction of the femoral head with its neck. The femoral capital epiphysis is placed in an inferior-posterior position (Figure 1) and the head is enlarged with marginal osteophytes, with eburnation present on its surfaces (Figure 2). Subchondral cysts and porosity are present and shortening and widening of the femoral neck is observable.



Figure 1. Left femur (anterior view) of the individual #19 from the Necropolis of Santa Maria (Sintra, Portugal).

The acetabulum was recovered from the same grave and articulates with the femoral head described above. This joint also has osteoarthritis present and no evidence for a false acetabulum on the outer surface of the ilium was observed (Figure 3).



Figure 2. Frontal view of the left femoral head of the individual #19 from the Necropolis of Santa Maria (Sintra, Portugal). Note the enlarged head with exaggerated new bone formation on its margins, along with a polished surface.



Figure 3. Left acetabulum of the individual #19 from the Necropolis of Santa Maria (Sintra, Portugal) showing new bone formation and eburnation.

Radiological features

The X-ray examination of the femur revealed sclerosis in addition to osteophytes surrounding the femur head, and at least one focus of osteopenia. This larger radiolucent area corresponds partially to post mortem damage, but there is also some ante mortem reduction of bone density. The medial cortex of the femoral diaphysis is slightly thickened (Figure 4). The X-ray of the pelvic bone fragment reveals areas of higher bone density and foci of osteopenia (Figure 5).



Figure 4. X-ray image of the left femur of the individual #19 from the Necropolis of Santa Maria (Sintra, Portugal), in anterior view.

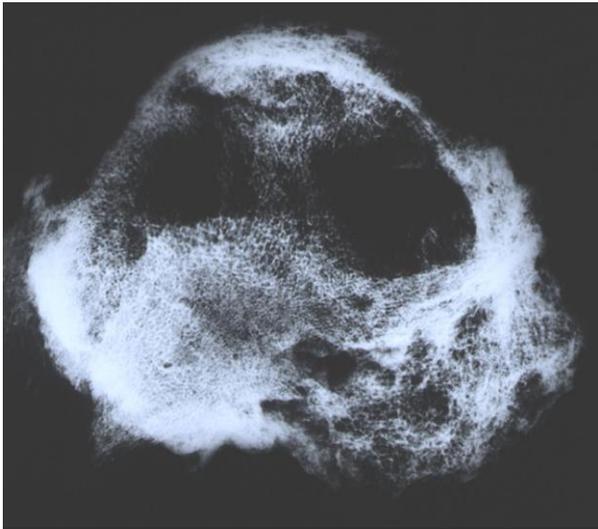


Figure 5. X-ray image of the left acetabulum of the individual #19 from the Necropolis of Santa Maria (Sintra, Portugal), in anterior view.

Case 1: femur from grave 35

Inventory and state of preservation

A lesion was observed on the proximal epiphysis of a second left femur. It is also from an adult individual, excavated from grave #35. No attempt to diagnose sex was made because it was only represented by its proximal diaphyseal region and epiphysis, both areas affected by the disease process.

Description of the condition

The femoral head, in this case, is most affected on the superior and anterolateral aspects, being extremely flattened and widened, creating a “mushroom-shape” appearance (Figure 6). It is located inferiorly to the greater trochanter, and the femoral neck is relatively short and thick, producing coxa vara. Furthermore, the head presents signs of severe degenerative joint disease,

and lacks the fovea capitis for ligamentum teres attachment (Figure 7). It is possible, though, that the fovea capitis was obliterated by the osteoarthritic changes. It is worthwhile to note the enlargement in diameter and the extensive presence of pitting and subchondral cysts.



Figure 6. Left femoral proximal end (anterior view) of the individual #35 from the Necropolis of Santa Maria (Sintra, Portugal).



Figure 7. Frontal view of the left femoral head of the individual #35 from the Necropolis of Santa Maria (Sintra, Portugal).

Radiological features

Once again an X-ray was performed, showing flattening and increased radiological density of the femoral head, and confirmed its mushroom-shape (Figure 8). There is considerable reduction in the mediolateral diameter of the head. The femoral neck is relatively short and wide, and a curved line that crosses its base, commonly known as “sagging rope” sign, was caused by the superimposition of the anterior margin of the flattened femoral neck. The radiological examination demonstrates that there was no fracture of the femoral head or of the neck.

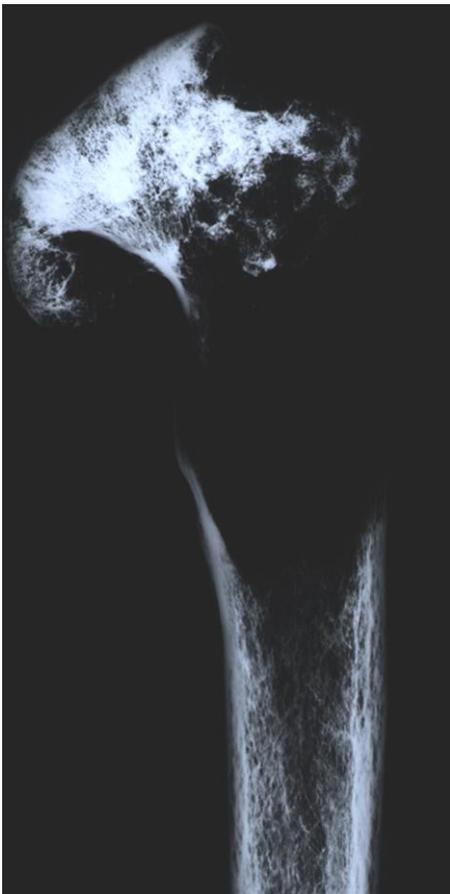


Figure 8. X-ray image of the left femoral head of the individual #35 from the Necropolis of Santa Maria (Sintra, Portugal), in anterior view, revealing areas of higher bone density and foci of osteopenia.

Discussion

Differential diagnosis

The differential diagnosis of the lesions gave rise to several possible pathological conditions; namely, slipped femoral capital epiphysis, Legg-Calvé-Perthes disease, developmental dysplasia of the hip, and trauma.

Slipped femoral capital epiphysis

Slipped femoral capital epiphysis (SFCE) is characterized by an inferior-posterior displacement of the femoral capital epiphysis, with the consequent fusion of the neck in that position (Ortner and Putschar, 1985). Etiologically, besides the genetic factor several others have been recognized, such as trauma, the adolescent growth spurt and obesity (Ortner, 2003).

A male predominance (sex ratio 2.5:1) is observed in this condition. It manifests itself around the onset of puberty, between 10-15 years in boys and two years earlier in girls. It rarely occurs under age 8 or over 17 years (Ortner and Putschar, 1985), and is bilateral in only 25% of the cases (Aufderheide and Rodríguez-Martín, 1998).

In life, it generally involves hip or knee pain and alterations in gait (Waldron, 2009). SFCE is characterized by a stress fracture between the metaphyseal side of the growth plate and the neck of the femur. This gives rise to a medial posterior and downward

displacement of the femoral head that can produce aseptic necrosis in the epiphyseal bone. The fracture, subsequent abrasion and resorption can lead to irregularities of the proximal end of the femoral neck. With healing, the center of the head presents some dislocation toward the axis of the neck. In marked contrast with Legg-Calvé-Perthes disease, the femoral neck is almost non-existent on the superior aspect and greatly shortened inferiorly. Degenerative osteoarthritis can occur. Sometimes, a new "acetabulum" is produced on the lateral aspect of the ilium as a result of the upward displacement of the femur, whereas the head is kept in the anatomical acetabulum by the ligamentum teres (Ortner, 2003).

One radiological sign that may occasionally be helpful is the appearance of cystic lesions in the femoral head which are considered typical of SFCE (Waldron, 2009).

Legg-Calvé-Perthes disease

Legg-Calvé-Perthes disease (LCPD) is a childhood osteochondrosis of the femoral head (Aufderheide and Rodríguez-Martín, 1998; Salter, 1999; Smrčka et al., 2009). It is caused by an obstruction to the blood flow that reaches the epiphysis of the femoral head, with resulting avascular necrosis (Aufderheide and Rodríguez-Martín, 1998; Ortner, 2003; Smrčka et al., 2009; Tripathy et al., 2010; Herrerín and Garralda, 2012). Despite being known for over 100 years, the etiology of this condition remains unclear (Hosalkar and Mulpuri, 2012), although there has been speculation regarding the potential role of traumatic, genetic, metabolic,

nutrition, environmental, hormonal, and hematologic factors as causative (Smrčka et al., 2009; Herrerín and Garralda, 2012). Of the many proposed theories, the one that seems most probable, and for which there is some experimental proof, is that the original occlusion of the precarious blood supply to the femoral head is caused by excessive fluid pressure of a synovial effusion in the hip, either inflammatory or traumatic. Approximately 5% of children with transient synovitis of the hip and an associated synovial effusion in the joint develop the complication of Perthes disease (Salter, 1999).

Boys (particularly those physically active) are more affected than girls (sex ratio 4:1). Approximately 10-20% of cases show bilateral involvement (Waldron, 2009; Tripathy et al., 2010). In the majority of these cases the condition does not occur synchronously in each hip. It has an age of onset from 3-10 years of age (average about 6 years) (Zimmerman and Kelley, 1982; Aufderheide and Rodríguez-Martín, 1998; Salter, 1999; Ortner, 2003; Waldron, 2009). The prognosis is good when onset occurs before the age of 5 years, fair when onset occurs from 5 to 9 years of age with more than half of the head involved and poor when onset occurs after the age of 10. In general, the younger the child is when affected, the wider and better the remodeling because of the length of time remaining until skeletal maturity for remodeling of the femoral head. It is perhaps this factor that explains why girls appear to have a worse prognosis than boys because of the more advanced skeletal maturation at onset (Wedge, 2003).

The most common clinical presentation of the disease is limp, with pain at hip, thigh and knee. These symptoms worsen with physical activity and at the later part of the day. As the disease progresses the limp, pain and restriction of movement increase, which correlates with the collapse of the femoral head. Progress thereafter is influenced by the nature and extent of ensuing femoral head deformity (Tripathy et al., 2010).

This malformation is frequently depicted as a “mushroom-shaped” femoral head (the superior and anterolateral aspects are deformed, flattened and widened). The femoral head is usually at a lower position than the greater trochanter and it is characterized by the absence of the fovea capitis for the insertion of ligament teres (Aufderheide and Rodríguez-Martín, 1998). Early severe degenerative arthritis changes the appearance, hampering differentiation from other conditions (i.e., the end stage of slipped femoral capital epiphysis) (Ortner, 2003). The acetabulum is also affected, becoming wider, flatter, and presenting an irregular articular surface (Herrerín and Garralda, 2012). Afterwards, there may be a decrease in the diameter of the obturator foramen due to a medial displacement of the ischium associated to an ischium varum deformation (Aufderheide and Rodríguez-Martín, 1998).

Radiographically, at first the affected epiphyseal center appears more opaque than the surrounding bone. Posteriorly, there is continued opacity and flattening of the epiphyseal center. In the last stage, radiolucent areas may be observable where

new replacement bone has not yet formed (Zimmerman and Kelley, 1982).

Although LCPD has been recognized in archaeological material for a century, it has been rarely reported. In 1982, Zimmerman and Kelley presented a probable case of ischemic necrosis in the femoral heads of a prehistoric American Indian. No further details are given. Ortner and Putschar (1985) described a right femur from the Valley of Chicama in Peru as a possible example of LCPD. The chronology of their specimen is unknown. In a review of the paleopathological literature, we identified only four cases of LCPD (Nevelös, 1986; Đurić et al., 2004; Smrčka et al., 2009; Herrerín and Garralda, 2012).

Developmental dysplasia of the hip

Developmental dysplasia of the hip (DDH), previously known as congenital dislocation of the hip, is an ancient disease characterized by a shortened lower limb and tilted pelvis (Zimmerman and Kelley, 1982; Mitchell and Redfern, 2008; 2011). In this condition, the acetabulum on one or both sides fails to deepen (Badgley, 2008; Waldron, 2009). In fact, the condition is bilateral in more than half of the affected individuals (Roberts and Manchester, 2005) and some cases are associated with other skeletal changes at the pelvis, the femur, the tibia and the spine (Mitchell and Redfern, 2008). Both modern clinical studies and past skeletal studies suggest that the prevalence of this condition is 1-2 per thousand. Lower prevalence of DDH in past skeletal series might be explained by preferential loss of dislocated

hips due to taphonomic processes, or the fact that human osteologists are missing the diagnosis (Mitchell and Redfern, 2008). Girls are six to eight times more likely to be affected than boys (Zimmerman and Kelley, 1982; Waldron, 2009).

Numerous well-known theories for the development of this condition have been advanced (for a revision, see Badgley, 2008). After an extensive revision of the available studies, Badgley (2008: 91) concluded that developmental dysplasia of the hip is “the result of faulty development, due to environmental factors extrinsic to the hip joint. An inherited fault in the timing of development may produce these extrinsic changes.”

If left untreated, the consequence of the hypoplastic shallow acetabulum is the formation of a bony reaction on the lateral cortex of the ilium, creating a new acetabulum. Subsequent degenerative osteoarthritis in this abnormal joint may lead to eburnation of the bony surface. The femoral head is flattened and presents a vertical groove for the flattened round ligament. There is frequently a large marginal exostosis present, pointing downward. The acetabulum is small, flat, and triangular indicating that it never articulated with nor supported a mature femoral head. In addition to the formation of a secondary acetabulum, some abnormalities of the hip may result in only chronic slippage of the femoral head within the shallow acetabulum, with no permanent dislocation nor formation of a new joint (Ortner, 2003). Even if present as a unilateral condition, the contralateral hip joint will also be abnormal compared with a

normal healthy hip (Mitchell and Redfern, 2011).

In past populations this condition became apparent only when the child started to walk, as limb shortening is the main problem that underlies many of the skeletal effects of DDH. Young affected children learn to walk at the same age as their counterparts but the sawing gait due to the recurrent dislocation of the joint on weight bearing and the inability to run or jump are characteristic (Mitchell and Redfern, 2008). By that time treatment, had it been available, would have been ineffective (Roberts and Manchester, 2005).

Previous research has shown that in archaeologically derived samples of human remains, DDH can be clearly differentiated from other pathological lesions (Blondiaux and Millot, 1991; Stirland, 1997; Mafart et al., 2007; Mitchell and Redfern, 2008; 2011). In fact, this condition has been recognized in skeletons from many periods (Blondiaux and Millot, 1991; Wakely, 1993; Masnicová and Benus, 2003; Mitchell and Redfern, 2008; 2011).

Differential diagnosis of case 1

The pathological alterations observed in the left femur of the grave #19 (case 1) are consistent with SFCE. Part of the femoral head was damaged post-mortem, although enough remains to indicate the nature of the pathology. The femoral neck is almost non-existent on the superior aspect and greatly shortened inferiorly. The eburnated femoral head is characteristic of secondary degenerative joint disease, which is a

consequence of SFCE. Radiological findings reflect the pathological changes; osteophytes are present to the margin of the articular surface, and the eburnation is evidenced as an area of sclerosis in the subchondral bone (Chhem and Brothwell, 2008).

Other pathological conditions, namely LCP disease and DDH, were considered but rejected (see Table 1). The existence of a well-defined depression for ligamentum teres excludes Perthes disease. The possibility of being a developmental dysplasia of the hip

was, as well, discarded since the acetabulum found, despite presenting pathological changes, did not show the development of a new joint for the femoral head, a false acetabulum. Trauma was also considered, nevertheless the absence of a fracture line in the radiological image does not favour this diagnosis. However, it is important to highlight that a traumatic etiology cannot be definitely excluded since the fracture line could have been already resorbed.

Table 1. Criteria for differentiating Legg-Calvé-Perthes disease (LCPD), slipped femoral capital epiphysis (SFCE), and developmental dysplasia of the hip (DDH) in archaeological material (according to Ortner and Putschar, 1985; Mitchell and Redfern, 2008; 2011).

Criteria	LCPD	SFCE	DDH	Case 1	Case 2
Mushroom-shaped femoral head	+	-	+/-	-	+
Head center dislocated towards the neck axis	-	+	-	+	-
Thickening of the neck	+	+/-	-	+	+/-
Shortening of the neck	+/-	+	+	+	+/-
Steep wide acetabulum	+	-	-	-	NA
New acetabulum at the ilium	-	+/-	+	-	NA
Porous lesion of the femoral head	+	-	+	-	+

NA – not observable

Differential diagnosis of Case 2

The pathological alterations observed in the left femur of the grave #35 (case 2) are

consistent with LCP disease. The femoral head is deformed and flattened, mushroom-shaped, with a porous surface and it is slightly displaced downwards. There is

extensive bone destruction as well as reactive new bone formation. Finally, the possibility of Perthes disease is also favored by the inexistence of a well-defined depression for ligamentum teres. This pathology can be misdiagnosed with SFCE, DDH and trauma. The first condition was excluded, due to the characteristic mushroom-like appearance resulting from the exuberant bony growth, with signs of porosity, which resulted in a complete obliteration of the depression for the ligamentum teres (Table 1). SFCE was excluded also due to the rather normal femoral neck length. According to Mann and Murphy (1990), the examination of the femoral neck length is extremely important, since, if it is normal, it will probably be a case of LCPD, whereas if it is short, it would be suggestive of a SFCE. The radiological examination (Figure 8) demonstrates that there was no fracture on the femoral head nor on the neck, which could suggest a possible traumatic origin of this pathology. Regarding DDH, this cannot be completely discarded due to the absence of the pelvic bone which made it impossible to accomplish the paleopathological evaluation.

Conclusion

In the present study, the absence of complete skeletons precluded any definitive diagnosis, as important factors such as unilaterally/bilaterally of the conditions could not be evaluated. Besides, the absence of important bones (e.g., tibia, vertebrae, and pelvic bones), usually affected by some of these diseases and used for its discussion and

positive diagnosis, was a limiting factor. Also, it should be highlighted that the rather poor state of preservation can indeed compromise definitive diagnosis in paleopathological analysis, which seems to have taken place with the two cases reported. Nevertheless, the diagnoses presented here, slipped femoral capital epiphysis and Legg-Calvé Perthes disease, seem the most probable. If, at first sight, both diseases affecting the femoral head appear similar, after a more careful evaluation it becomes evident that some macroscopic signs differ, a fact corroborated by radiological analysis, leading to the presumption that we are in the presence of two different pathological conditions.

Since there are only a few reported cases of both these conditions in the osteoarchaeological literature (Knüsel et al., 1992; Đurić et al., 2004; Smrčka et al., 2009; Herrerín and Garralda, 2012; Walker, 2013), we find it to be interesting and important to report these two cases. In fact, this appears to be the first report of slipped capital femoral epiphysis in non-identified Portuguese human skeletal remains. Regarding Legg-Calvé-Perthes disease, there are only two other cases described (Marques and Cunha, 2001; Ferreira et al., 2013).

Finally, in this paper, two rare conditions are compared and contrasted, representing therefore a significant contribution to the paleopathological field.

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