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PSEUDOACHONDROPLASIA IN DOGS – A CASE REPORT

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Abstract

Here we present the radiographic findings of limb and spine in a 15-month-old female Bichon with pseudoachondroplasia (PSACH). PSACH is a rare form of osteochondrodysplasia, the main clinical characteristics are disproportionate short stature, abnormalities of the limbs and spine, abnormal walk, joint laxity and early osteoarthrosis. The animal was evaluated by physical and radiographic exams. Radiographic images of the skull, spine and limbs were obtained, and showed normal appearance of cranial structure and multiple changes in the limbs and spine, including generalized dysplasias of epiphyses and metaphyses of the long and short bones. Epiphyses were incomplete developed and had irregular appearance, separated by a radiolucent line, and metaphyses of the long bone were prominent flared appearance. Carpal and tarsal bones were also affected, showing shape changes.

Key words: *dog, pseudoachondroplasia,skeletal dysplasia.*

INTRODUCTION

Locomotory apparatus includes bones, articular and muscular structures in which are present all categories of tissues used for growing, mecanic suport, vital organs protection, movement and mineral and blood cells reservoir (Predoi et al., 2012; Georgescu and Raita, 2014). Skeletal components of vertebrates have their origins in mesoderm (Docheva et al., 2014) which is the second embryo layer. In ontogenetical development the mesoderm generates the mesenchyme out of which arise all types of conjunctive tissue of adult animals (Cornilă and Manolescu, 1995). Conjunctive tissues contain conjunctive cells and extarcellular matrix (ECM) (consisting of fundamental substance and conjunctive fibres), then after metaplastic processes those components are suffering adaptive phenomena causing appearance of supportive conjunctive tissues, respectively cartilage and bone tissues (Georgescu and Raita, 2014). Under the influence of physical, chemical, nervous and humoral factors, conjunctive cells perform multiple functions such as mechanical, defense (by phagocytosis, antibody release), cytogenetics (generating other cell types) and synthesis of various protein and carbohydrates range composing the ECM (Georgescu and Raita, 2014). Collagens are one of the most important structural proteins which compose the ECM having widespread inside the organism and determinating specific features of tissues depending on the collagen type (Krakov and Rimoin, 2010). Bone formation starts in fetal period with two osteogenetic mechanisms, inside matrix ossification on a conjunctive pattern, the way it happens in forming the bones in cranial vault, maxilla, part of the clavicle and pubis, or on a hyaline cartilage pattern, the way it happens with formation of long bones (Georgescu and Raita, 2014). The bone formation mechanism is under specific genetic and direct control (Colnot, 2005). Mutations in genes encoding collagen fibres formation and cartilage ECM proteins, such as cartilage oligomeric matrix protein (COMP), proteoglycans aggrecan and perlecan usually result in various skeletal dysplasia (Ionita C., 1999; Carter et al., 2009; Krakov and Rimoin, 2010; Cao et al., 2011; Kyöstilä and al., 2013). Skeletal dysplasia found in humans and dogs alike are disorders of varying severity characterized by abnormally skeletal shape and size and long bones, spine and skull disproportion (Warman et al., 2011). Currently in humans based on radiological and clinical criteria are described more types of skeletal dysplasia (Alanay and Lachman, 2011), while in dogs include more entities which sometimes can look similar clinical and radiological but histological and biochemical they are representing a heterogeneous group of disease (Wisner and Pollard, 2007). Among them is

pseudoachondroplasia (PSACH), a rare form of osteochondrodysplasia whose main characteristics are disproportional waist shortening, limbs and spine abnormalities, abnormal walk, articular laxity and early osteoarthritis (Radlović et al., 2013). The purpose of this report is to describe radiographic findings of limb and spine PSACH in a 15-months-old female dog.

MATERIALS AND METHODS

A 15-months-old female Bichon was presented for evaluation with walking disorders, pain and deforming of limb articulation. Faulty movement has been seen since she was 4 months old and in time became more obvious. The animal was evaluated by physical examination (inspection and palpation) and radiographic as previously described (Tudor, 2002; Papuc and Lăcătuş, 2013). The Radiological exam was conducted from orthograde incidents using a computed radiography machine set to 66 kV and 6.3 mAs with a radiation source – film distance of 100 cm. Were obtained radiographic images of the skull, spine and limbs.

RESULTS AND DISCUSSION

Pshysical examination has shown that the limbs were shorter than the normal size with a high sensibility in articular palpation with a grown laxity and a slow movement. There were not found cutaneous, ocular or craniofacial changes. Radiographic images have indicated normal appearance of cranial structures and multiple changes in limb and spine bones. The reduction in dimension of bone rays was accompanied by important bone changes in all ephihyses both vertebral bodies and long bones. The ephiphyses were incompletely developed and had irregular appearance still separated from the shaft by a radiolucent line. The long bone metaphyses were deformed especially the distal humerus, radius and ulna which had a flared apperence (Fig. 1 and 2). Bony growths were noticed at the crest deltoid of humerus and medial humeral epicondyle level. Vertebral end plates have had irregular appearance, distictly separated from the vertebral body. Moreover, the ventral borders of the vertberal bodies were lost the pleated appearance and vertebrae have been shorter than normal. Tarsal and carpal bones were affected also showing shape changes. It was also found deformation of shoulder glenoid cavity bilaterally.

In the pelvis it has been found acetabular cavity and proximal femoral epiphyses agenesis (Fig. 3). Limb articulations were modified mostly in the elbow, knee and hip. As a result of bone ends incongruity involved in the articulation were produced degenerative processes especially in the elbow. Since onsteochondral changes incuded only the axial and appendicular skeleton without affecting craniofacial structures (such as achondroplasia does) it was diagnosed with PSACH.

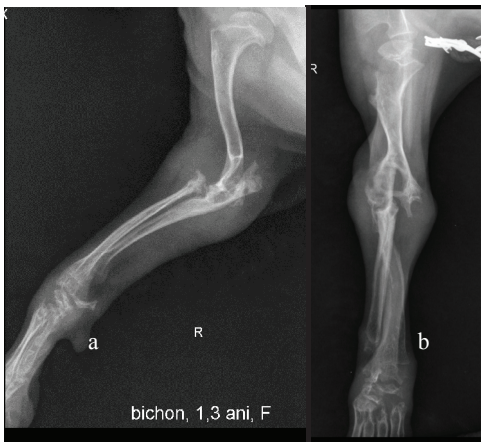

Figure 1. Lateral (a) and craniocaudal (b) views of the right limb. In the lateral view it can saw severe changes on bones: incomplet growth of the femural head; the proximal humerus metaphysis is wider; the elbow joint incongruency is faulty development of the distal epiphysis of the humerus and the proximal epiphysis of radius and ulna; the distal metaphysis enlargement and the incomplete growth of the distal epiphysis of the radius and ulna; carpal bones and proximal epiphysis of the metacarpal bones have incomplete growth. In the craniocaudal view it can be observed the normal appearence of the proximal and distal extremities of the humerus, malformation of the elbow joint, the new bone formation on the medial epicondyle of humerus, the medial angular deformity of the ulna, enlargement of the distal metaphysis of ulna, incomplete growth of the distal epiphysis of radius and ulna like carpal bones and proximal etremities of metacarpal bones.

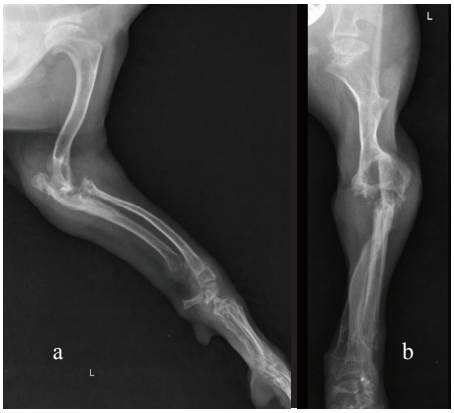

Figure 2. Lateral (a) and craniocaudal (b) views of the left limb. Can see bone changes similar to those of the right forelimb.

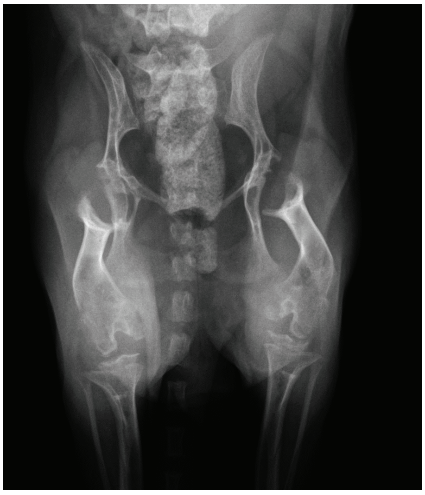

Figure 3. Ventrodorsal view of pelvis and hind limbs. It can be observed the epiphyseal alterations of the endplate in vertebral bodies. On hind limbs it can be observed bilateral bone severe changes: the acetabular cavity and the femural head are not formed, femural diaphysis have angular deformity; the proximal tibial epiphysis is incomplet formed and the proximal tibial metaphysis is wider.

Generally osteoachondrodysplasia occurs as a result of faulty endochondral ossification causing disproportionate dwarfism and morphological defects of appendicular and axial skeleton (Tanase and Craciunescu, 2003; Wisner and Pollard, 2007; Ionita L., 2008; Cao et al., 2011). Traditionally some dog breeds were classified as chondrodysplastic breeds based on their phenotypic aspect such as Basset Hound, Dachshund, Bulldog etc. (Hoelen, 2010), and gene intervention in producing these defining breed features was demonstrated (Parker et al., 2009). It was reported that osteochondrodysplasia appears also in some non-chondrodysplatic breeds such as Alaskan Malamutes (Sande et al ., 1982), Norwegian Elkhounds (Bingel and Sande, 1982), Great Pyrenees (Bingel and Sande, 1994), Scottish Deerhounds (Breur et al., 1989), German Shepherd (Mosallanejad et al., 2007), Miniature Poodles (Riser et al., 1980) and Iris Setter (Hanssen et al., 1998). This condition is considered to be hereditary having different phenotypic expressions (Wisner and Pollard, 2007). Besides skeletal defects the disease can affect other organs like the eye causing blindness as is was pointed in Labrador Retrievers (Goldstein et al., 2010) and Samoyeds (Meyers et al., 1983).

Radiographic features of skeleton in the case described here were represented by generalized dyspalsia of limb bone epiphysis and metaphysis and vertebral bodies without involving craniofacial bones are compatible with PSACH (Wisner and Pollard, 2007; Radlović et al., 2013). All limb bones were shorter than they normally are and the most affected were humerus and femur typical changes for rhizomelic dwarfism (Radlović et al., 2013). Moreover, were indicated angular deformities of the limbs showing asychronous growth of bone rays (Tanase, and Craciunescu, 2003). Thus the femur and radius have presented the most obvious changes. Radiographic images have shown articular deformation with the production of new periepifizar bone and the articular laxity seen in palpation sugests affecting tendon and ligament structures. PSACH diagnosis is based firslty on familly history of the individual and characteristic findings pshysical and radiographic. When possible genetic verification of the skeletal disorder must also be made. Skeletal changes met in this case were simillar with those described previously at dog and man (Wisner and Pollard, 2007; Radlović et al., 2013).

PSACH is a condition framed in the osteochondrodysplasia group which contains more than 150 distinct conditions characterized

by abnormal developement of bone and cartilage (Spranger, 1992). Previous studies established that in humans this condition happens consequently to a structural gene mutation positioned on 19p12-13.1 choromosome (Briggs et al., 1995; Cao et al., 2011) and encodes COMP, an ECM protein primarly expressed in cartilage, tendon and ligament, but also in many other tissues (Kracov and Rimoin, 2010). In dogs PSACH has been described in Miniature Poodle (Riser et al., 1980) and Scotish Deerhound (Breur et al., 1989) beeing considered by some authors to be a form of multiple epipyseal dysplasia (Riser et al., 1980), but skeletal abnormalities from PSACH are more generalized than in MED (Jezyk, 1985). In this study were described skeletal adnormalities of limbs and spine in Bichon dog, which is a rare case.

The causes of the appereance of this condition in the dog from the present report are still unclear because of the unknowingly of any data about the dogs parents or brothers or the conditions in which the female gestation took place. The involvement of genetic mutations in the appereance of this skeletal abnormality were previously presented (Carter et al., 2009; Goldstein et al., 2010; Frischnecht et al., 2013; Neff et al., 2012).

PSACH was differentiated from other condrodysplasic abnormalities such as chondrodysplasia of Alaskan Malamutes, chondrodysplasia of Norwegian Elkhounds or ocular chondrodysplasia of Labrador Retrievers and Samoyed dogs based on the radiographic and clinical features. Where in the case of Alaskan Malamutes skeletal changes affect limb bones without involving the skull and the spine. The psysis of all limb bones can be affected but obvious radiographic changes are pointed in the distal physis and metaphysis of ulna and radius. Asychronous growth is causing angular deformations of the limbs (Sande et al., 1982). Chondrodysplasia of Norwegian Elkhounds is characterized by disproportionate dwarfism but the forelimbs are more frequently affected than the hindlimbs. In addition radiographical are found abnormalities of the vertebrate bodies and constochondral junction whithout affecting the skull (Bingel and Sande, 1982). In Labrador Retriever breed skeletal expression of the limbs, varied in this breed (Smit et al., 2011; Frischknecht et al., 2013), may be accompanied sometimes by ocular manifestations such as cataracts, retinal dysplasia and retinal detachment (Goldstein et al., 2010).

CONCLUSIONS

PSACH represents a form of osteochondrodysplasia characterized clinically by dwarfism and abnormal locomotion and radiographic by generalized dysplasias of epyphyses and metaphyses of the long and short tubular bones.

Avoiding the appereance of such compounds should be traced by breeding control and inbreeding avoidance.

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