

## RADIOLOGICAL ASPECTS ASSOCIATED WITH OSTEOCHONDRODYSPLASIA IN A SCOTTISH FOLD CAT: CASE REPORT

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### **Abstract**

*Scottish Fold cats have an unique appearance due to their forward-folded ears, being considered a defining feature of the breed. It is considered that the folded ears are a result of an underlying cartilage disorder, a genetic abnormality that predisposes the affected cats for another genetical condition of the musculo-skeletal system, termed osteochondrodysplasia. Osteochondrodysplasia is a painful and progressive syndrome characteristic only to the Scottish Fold breed due to a mutation in the TRPV4 gene. This condition is characterised by skeletal deformities, such as shortened feet and splayed phalanges, affecting primarily the hindlimbs, short, thick and inflexible tails, shortened caudal vertebrae and ankylosing polyarthropathy of the affected joints. This paper has the objective of characterising the main bone and joint problems observed in a 1 year-old female Scottish Fold cat which presented for lameness and postural abnormalities of the hindlimbs.*

**Key words:** cat, Scottish Fold, radiography, osteochondrodysplasia, congenital anomaly.

### **INTRODUCTION**

The Scottish Fold cat has been developed as a breed starting the 1970s in Scotland, after the mating of a farm cat with naturally occurring folded ears to local cats and British Shorthairs (Wastlhuber, 1991).

Although the breeding of these cats started as early as 1961, the first mention of skeletal abnormalities and deformities was not made until 10 years later, when osteochondrodysplasia was recognised as a painful clinical syndrome, characterised by progressive bony abnormalities and a crippling lameness (Robinson & Pedersen, 1991; Wastlhuber, 1991).

Genetic analyses have designated the gene responsible for the defective endochondral ossification as Fd, suggesting that this is the gene that also causes the articular cartilage to fold and is inherited as a simple autosomal dominant trait (Robinson & Pedersen, 1991). Over several years of genetic testing the conclusion is that kittens that show radiological

lesions are only a product of mating Fold-to-Fold cats (Jackson, 1975).

The breed has been banned in the United Kingdom starting as early as 1974 and breeding in the United States and other countries has been frowned upon ever since by many organisations focused on the welfare of cats (Malik et al., 1999; Brocklehurst, 2017).

Recent DNA testing of 44 Scottish fold cats and 54 control cats suggested that there is a variant of the TRPV4 gene that is associated with ear folding (Selting et al., 2019). TRPV4 is expressed in chondrocytes and other mesenchymal cells and has a great impact in tissue homeostasis and cellular differentiation (Gandolfi et al., 2016).

The first description of osteochondrodysplasia of the Scottish fold cat was made by Jackson, in 1975 (Jackson, 1975). The main radiological findings are abnormally thick and inflexible caudal vertebrae, short feet and splayed phalanges, resulting in reduced ability to support weight, an abnormal gait, reluctance to jump and lameness (Malik et al., 1999; Chang et al., 2007). The joint lesions progress until the

cats become non-ambulatory. The radiographical lesions are evident in cats as young as 7 weeks of age (Jackson, 1975). The metacarpal and metatarsal bones, as well as the phalanges have distorted metaphyses, with widened physes, resulting in decreased length and abnormal shape of the affected bones and shortening of the hindlimbs (Malik, 2001). The caudal vertebrae have reduced length and widened endplates. Cats older than 6 months develop gross plantar exostoses of the tarsal and metatarsal bones, which become evident both clinically and radiographically. Diffuse osteopenia of the bones is also present (Chang et al., 2007).

The histopathological examination of affected tissues shows defective bone formation at the level of the growth plates, affected chondroblast proliferation and irregular groups of cells arranged in a disorganized manner. The osseous physes are grossly expanded, with deficient ossification, irregular mineralisation and remodelling and multiple centres of ossification (Malik et al., 1999).

Due to the fact that the underlying disease has a genetic trait, the methods of therapy used have the primary aim of reducing clinical signs and the accompanying pain (Hubler et al., 2004). One study shows the effects of pentosane polysulfates and complex glycosaminoglycans which provided reduction in lameness and discomfort in some cats (Malik et al., 1999). Another study focuses on the effect of the surgical approach in which bilateral osteotomies and pantarsal arthrodesis of both hindlimbs resolved the lameness in a cat (Mathews et al., 1995). A study from the year 2000 had obtained the same results only by surgical removal of the tarsal exostoses (Simon, 2000). The latest studies involving therapy for cases with osteochondrodysplasia focus on the effects of radiation therapy in conjunction with phosphonic acid, which showed immediate, whole-body improvement in mobility (Selting et al., 2019).

## MATERIALS AND METHODS

A one year old, entire female Scottish fold cat was presented to a private practice near Pitesti, Romania in 2019 due to progressive lameness

in the hindlimbs over the past weeks, abnormal gait and misshapen distal hindlimbs.

There was no previous medical history associated with the patient. On clinical examination, the cat was bright, alert and responsive. The heart rate was 160, with no murmurs or arrhythmias present. Body condition score was 4/9 and the patient was ambulatory, although it had a stiff gait. The tail was observed by the owners to be moving only at the tip ever since it was a kitten. When palpating the hind limbs, the tarsal region appeared firm on the plantar part and elicited pain upon palpation. The region was stiff and could not be manipulated easily. The hind paws had an abnormal shape, with the first and last toes being in front of the other. The middle part of the tail was fused and could not be flexed easily in any direction. The abdomen was within normal limits.

Conscious radiographs of the hindlimbs, forelimbs and tail were obtained in latero-lateral and dorso-palmar projections.

## RESULTS AND DISCUSSIONS

The changes observed as a result of the radiological examination were consistent with osteochondrodysplasia, including shortened coccygeal vertebrae with widened endplates (Figure 1), malformed tarsal and metatarsal bones, exuberant exostoses of the tarsal region (Figure 2 A, B) and shortened and splayed phalanges (Figure 3). The interphalangeal joints spaces were irregular and widened (Figure 3). The forelimbs presented radiological signs of incipient bilateral elbow osteoarthritis, although not clinically significant at the time of the examination (Figure 4 A, B).

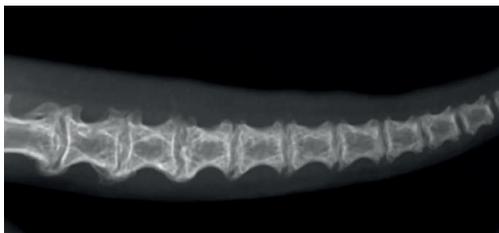


Figure 1. Lateral radiograph of the tail. The middle caudal vertebrae are short, thickened and the intervertebral spaces are reduced. There is periarticular new bone formation observed



Figure 2. Lateral (A) and dorso-palmar (B) radiographs of the left hindlimb. Extensive new bone formation originating from the tarsus and extending to the metatarsals is present. The metatarsal bones are short, thick and misshapen



Figure 3. Dorsopalmar radiograph of the paw of the right hindlimb. The phalanges are short, misshapen and there is diffuse reduced opacity of the bones. The interphalangeal joints are irregular and widened



Figure 4. Medio-lateral radiographs of the right (A) and left (B) forelimbs. There are reduced signs of elbow osteoarthritis present

Metacarpals were somewhat shortened and with an abnormal shape compared to the distal limb of a domestic shorthaired cat (Figure 5 A, B).



Figure 5. Dorsopalmar radiograph of the distal forelimb region of the cat in this study (A) and a domestic shorthair cat (B) for comparison. There is evident shortening and thickening of the metacarpal bones and phalanges in A compared to B and widening of the interphalangeal joints

The changes observed were bilaterally symmetric and affecting primarily the tarsal region. There was massive new bone formation extending from the proximal part of the calcaneus to the metatarsus, leading to bone fusion and tarsal ankylosis.

The new bone was smoothly marginated, with a typical trabecular pattern. There was diffuse decreased opacity of the bones affected (osteopenia). The metatarsal bones were short, thickened, malformed and had a splayed appearance. The interphalangeal joints were irregular. In the shortened and thickened tail, several caudal vertebrae were shorter than normal, with widened end plates, reduced intervertebral joints and new bone formation, tending towards ankylosis.

The forelimbs were thought to have minor changes related to osteochondrodysplasia in the distal parts and mild changes of degenerative joint disease of both elbows – not clear if related to osteochondrodysplasia or not, further studies needing to be conducted.

The defining phenotypic feature which identifies a cat as being part of the Scottish fold breed is characterised by the forward folding of the ears, suggesting a developmental cartilage defect (Malik et al., 1999).

It has been previously reported that osteochondrodysplasia of the Scottish Fold cat can be diagnosed easily through survey radiographs in animals as young as 7 weeks old, older individuals developing ankylosis, as can be seen in the tarsal region of the cat in our study (Chang, 2007; Malik et al., 1999).

Treatment for cats suffering from this disease has not been determined convincingly to date, although some studies have shown the benefits of using polysulphated glycosaminoglycans, chondroprotective agents or non-steroidal anti-inflammatory agents safe for cats (Chang, 2007). Other studies have shown the benefits of osteotomies or arthrodeses (Matthews et al., 1995) and of using palliative irradiation (Hubler et al., 2004) or novel radiation therapies (Selting et al., 2019), but to this date the main proposed solution would be removal of the affected individuals from the mating pool and to restrict breeding to cats with a normal ear conformation (Allan, 2000).

## CONCLUSIONS

The clinical presentation, the onset of clinical signs and the radiological changes observed make the diagnosis of Scottish fold osteochondrodysplasia certain for the cat in our study.

Prospective owners should be warned about the possibility of Fold developing musculoskeletal abnormalities and about the implications that this disease has on the quality of life of their animal.

Additional studies should be conducted using serial computed tomography or magnetic resonance imaging in order to obtain further information regarding bone and joint changes in a non-invasive manner.

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