# Hereditary Osteochondrodysplasia in Scottish Fold Cats<sup>[1][2]</sup>

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### Abstract

In this study, the clinical progress and the treatment results of cases Istanbul University, Faculty of Veterinary Medicine, Department of Surgery with a complaint of stilted gait, pain, lameness, reluctance to move, kyphosis and miniature appearance have been evaluated. The study materials included eleven Scottish Fold cats. The cases underwent conservative and surgical treatment due to severity of osteochondrodysplasic lesions. The aim of this study is to draw attention to this illness of homozigot and heterozigot Scottish Fold cats whose population is increasing dramatically in our country, to stop the uncontrolled mating of this breed and also to share our foundings and results with our colleauges.

Keywords: Scottish Fold osteochondrodysplasia, Cat

# Scottish Fold Kedilerde Kalıtsal Osteokondrodisplazi

### Özet

Bu çalışmada, İstanbul Üniversitesi Veteriner Fakültesi Cerrahi Anabilim Dalı'na hareket etmede isteksizlik, tutuk yürüyüş, topallık, arka bacaklarında şişlik, kifozis ve minyatür görünüm gibi şikayetlerle getirilen, tanısı kalıtsal osteokondrodisplazi olan, onbir adet Scottish Fold kedinin klinik durumları ve sağaltım sonuçları değerlendirilmiştir. Olgulara osteokondrodisplaziye bağlı lezyonların şiddetine bağlı olarak konservatif ve/veya cerrahi sağaltım uygulanmıştır. Bu çalışmayla, ülkemizde sayıları giderek artmakta olan Scottish Fold kedilerindeki kalıtsal nitelikli bu hastalığa dikkat çekmek, kontrolsüz çiftleşmelerin engellenmesini sağlamak ve ilaveten sağaltım seçeneklerinin bulgu ve sonuçlarını meslektaşlarımızla paylaşmak amaçlanmıştır.

Anahtar sözcükler: Scottish Fold osteokondrodisplazi, Kedi

### INTRODUCTION

The characteristic feature of Scottish Fold cats is their folded ears <sup>[1-4]</sup>. The reason for this morphological difference is an autosomal dominant gene mutation that affects cartilage development. However, this mutation not only creates a simple morphological difference but also presents itself as hereditary osteochondrodysplasia <sup>[1-3]</sup>.

The Scottish Fold breed emerged for the first time in the 1960s as a result of the mating of naturally-mutated local farm cats and British Shorthair cats <sup>[2,3]</sup>. The osteochondrodysplasia developing in all cats with folded ears <sup>[2,3]</sup>, is a developmental abnormality affecting all cartilaginous

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structures in the body and the disease is manifested by deformed bone structure and severe arthritis <sup>[1-4]</sup>.

The abnormal ear structure of Scottish Fold cats is due to the insufficiency of cartilage support. The folded appearance of the ears may become apparent when the kittens are 2-4 weeks old <sup>[1,2]</sup>. Studies have concluded that, this malformation in the pinna is an incomplete dominant hereditary feature of Scottish Fold cats <sup>[4]</sup>.

Osteochondrodysplasia is not seen in homozygote cats alone. Varying degrees of primary developmental deformities are also observed in heterozygote cats <sup>[1-4]</sup>. The earliest and most common finding seen in homozygote cats is a short, thick, unbending tail structure <sup>[24,5]</sup>.

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Findings may emerge in later stages in heterozygote cats <sup>[2,3]</sup>.

Progressive deformities in the tail and distal extremities emerge when the cats are approximately 7-weeks old and these remain throughout their lives. The metaphyses of bones forming the tail and extremities are wide and the bone length is shorter than normal. Deformed tail vertebrae are fused together and the tail loses its flexibility due to this fused structure <sup>[1,2,4]</sup>.

On radiographs, a deformed and irregular structure is observed in the tarsus, carpus, metatarsus, metacarpus, phalanges and tail vertebrae <sup>[4-6]</sup>. Once the kittens have reached 7-weeks of age, the deformity which has developed in the metaphyses of the metacarpal and metatarsal bones is visible on radiographs. Widening, flattening and sclerotic areas may be observed in the physis. Lesions may be less severe in the phalanges. The tail vertebrae appear short and growth plates expand. After 6-months of age, progressive new bone formation, especially in the distal extremities, and exostosis formation on the plantar surface of the calcaneus are observed radiologically. These exostoses may lead to joint ankylosis. Widespread osteopenia also accompanies the lesions <sup>[2-6]</sup>.

The aim of this study is to raise awareness of this disease observed in both homozygote and heterozygote Scottish Fold cats, whose numbers in Turkey are increasing, draw attention to the prevention of their breeding and to present to colleagues the findings and results obtained in the study.

### **MATERIAL and METHODS**

The material of the study comprised eleven Scottish Fold cats brought to the Istanbul University, Veterinary Faculty, Surgery Department between the years of 2010-2014, with complaints such as; reluctance to move, a stilted gait, lameness, swelling in the hind legs, kyphosis and a miniature appearance.

Following obtaining the history of each case from patient owners, physical and radiological examinations of

the cases were performed. In the radiological examination of the cases, craniocaudal (CC) and mediolateral (ML) radiographic views of the distal extremities and phalanges and ventrodorsal (VD) and laterolateral (LL) views of the tail vertebrae were obtained.

Apart from case numbers 1, 9, 10 and 11, radiological follow-ups of the patients still continue at 2-monthly intervals at the present time. Case numbers 2, 3, 4, 6, 7, 8, 9, 10 and 11 received conservative treatment while surgical treatment was performed on case numbers 1 and 5 in order to remove exostoses. Prior to commencement of treatment, full blood count and biochemical parameters of all cases were evaluated.

For treatment, meloxicam was recommended orally at a dose of 0.1 mg/kg on the first day and 0.05 mg/kg thereafter, glycosaminoglycan chondroitin sulphate at a dose of 0.5-1 mg/kg and 65 mg omega-3 oil acid containing tablets once daily.

## RESULTS

Of the 11 cats constituting the study cases, 6 were female and 6 were male. Three of the females had been spayed and one of the males had been castrated. Ages of the cats varied between 5 months and 3.5 years and bodyweight changed between 1.6 kg and 3.8 kg.

During clinical examination, all of the cases had a placid temperament and presented with a stilted gait. With the exception of case numbers 1, 9 and 11, the patient owners reported that all of the cases, aged between 5-months old and 2-years old, had been reluctant to move since they had been homed and constantly displayed a desire to sit, had a miniature appearance together with kyphosis and gave a pain response when manipulated.

In case numbers 1, 9 and 11, patient owners expressed that there had not been any particular muscular complaint except for the lameness which had begun in the past few days, however, that the cats had never been very mobile.

In the physical examination of case numbers 1, 9 and 11, difficulty in walking and a bilateral plantar



**Fig 1.** Case number 1: pre-operative (a, b) and immediately post-operative (c, d) radiographic view of left tarsal joint. Craniocaudal **a**, **c** - and mediolateral **b**, **d** - positions

Şekil 1. Olgu no: 1'e ait preoperatif (a, b) postoperatif hemen (c, d) sol tarsal eklem radyografik görüntüsü. a, c- kraniyokaudal, b,d-mediolateral pozisyonlarda alınan radyografiler stance, together with bilateral, painful, hard masses of sizes ranging between a walnut and a tangerine were identified.

In case number 1, it was discovered that the range of motion in the tarsal joints had decreased and bilateral partial ankylosis had developed in the joints. In case number 9, there was partial ankylosis in the right tarsal joint while complete ankylosis was present in the left tarsal joint. Joint ankylosis in case number 11 had developed in the reverse order to those seen in case number 9. In all three cases, a clear pain response was elicited during physical examination of the tarsal joints. No abnormal findings were encountered in the length of the tail and distal extremities of these cases, however, in case number 11 there was severe pain during palpation of the tail region and difficulty in tail movement.

In radiographic assessments, distinct osteochondrodysplastic lesions were observed bilaterally in the tarsal joint areas. On radiographs, new bone formation with a cauliflower appearance was seen along the caudal surface starting from the proximal left calcaneous until the proximal metatarsus in case number 1 (*Fig. 1*), and on the proximal plantar surface of the metatarsals in case numbers 9 (*Fig. 2*) and 11 (*Fig. 3*). On the tail radiograph of case number 11, new bone formation was observed between the tail vertebrae (*Fig. 4*).



Fig 3. Case number 11: Radiographs taken at the time of first physical examination. a- mediolateral view of left tarsal joint, b- mediolateral view of right tarsal joint, ccraniocaudal radiographic view of bilateral tarsal joint

Şekil 3. Olgu no: 11'in muayeneye getirildiğinde alınan radyografileri. a- sol tarsal eklem mediolateral, b- sağ tarsal eklem mediolateral, c- bilateral tarsal eklem kraniyokaudal radyografik görüntüsü





**Fig 4.** Case no: 11, laterolateral radiographic view of tail

**Şekil 4.** Olgu no: 11'e ait laterolateral kuyruk radyografisi





In the physical examination of case numbers 2, 4 and 5, all under 1-year of age, the distal extremities were

shorter than normal, thick and the joints were swollen and bulging. The tail structure of case numbers 2 and 4

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**Fig 9.** Case number 11, Operation views. **a**- new bone formation (*black arrow*), **b**- after removal of new bone formation, **c**- removed bony parts

Şekil 9. Olgu no: 5'e ait operasyon görüntüleri. a- yeni kemik oluşumu (*siyah ok*), b- yeni kemik oluşumunun uzaklaştırıldıktan sonraki hali, c- uzaklaştırılan kemik parçaları

was shorter than normal, thick and lacked flexibility. In radiographic images obtained at 2-monthly intervals, in case numbers (*Fig. 5*), 4 (*Fig. 6*) and 5 (*Fig. 7*), osteochondrodysplastic lesions first appeared as widening and proliferation on the plantar surface of the calcaneus and metatarsal bones and gradually

Fig 8. Case number 2, Craniocaudal radiographic view of

Şekil 8. Olgu no: 2'ye ait bilateral karpal eklem kraniyokaudal

bilateral carpal joints

görüntüsü

became distinct exostoses. At the end of 6 months, it was seen that the epiphyseal and metaphyseal regions of distal extremities had flattened extensively, sclerotic areas had increased, intertarsal and tarsometatarsal joint spaces had become indiscernible and irregular bone structure was very advanced. Deformation in the forelegs and metacarpal bones was seen only in case numbers 2 (*Fig. 8*) and 11. There were no obvious osteochondrodysplastic deformities in the forelegs of the other cases. In the palpation of distal extremities of all the cases, deformation was palpable and pain was present.

The severity of symptoms and radiographic lesions related to osteochondrodysplasia was relatively moderate in case numbers 3, 6, 7 and 8 compared to lesions in the remaining seven cases. The areas where proliferative lesions were observed were limited to the proximal part of the caudal surface of the metatarsals.

Following the assessment of case numbers 1 and 5, (*Fig. 9*) it was decided to perform surgical treatment in order to remove proliferative bone formations. Long-term clinical and radiological follow-up of case number 1 could not be done. In telephone conversations held with the owner of case number 5, it was established that the painful swellings in the hindlegs had recurred a few months after the operation. In case numbers 2 and 4, the lesions continue to advance. In case numbers 2, 4, 9 and 11, patient owners were explained that treatment was not curative and lesions could recur. Ostectomy was advised for ease of walking in the short-term, however, the patient owners declined this suggestion.

## DISCUSSION

Despite being identified as a cat breed by The Governing Council of the Cat Fancy, the largest organisation for registering pedigree cats in the United Kingdom, the Scottish Fold cat was removed from the registered breed list for the first time in 1974, due to the deformities in its extremities and tail. Furthermore, the International Feline Federation (Federation Internationale Feline) deemed the reproduction of this breed unethical due to the distinct pain these cats suffer because of the genetic mutation and banned breeding of these cats carrying a hereditary disease <sup>[2,7]</sup>.

The fact that the folded-eared appearance of this breed originates from generalised cartilage deformation <sup>[1,8-10]</sup> is either disregarded or not known and the endearing round faces and expressions of cats of this fenotype come across as likeable and attractive <sup>[11]</sup>. Regrettably, breeding of Scottish Fold cats continues in the United States and other countries around the world. This breed of cat used to be rare for Turkey at the beginning of the 21<sup>st</sup> Century, however, in recent years their numbers have been increasing.

All of the patient owners in this study expressed that they had chosen this breed of cat because of its folded ears. Also, it emerged that most of the patient owners were unaware of this disease while some patient owners explained that they knew about the disease but believed that they would not come across it since they thought their cat was a Scottish Fold-British Shorthair cross. The patient owners were strongly advised that the five unneutered cats should never be used for breeding.

Scottish Fold cats possess a miniature physiognomy. The distal extremities of affected cats may be insufficient to bear the bodyweight. In these cats, lameness, a stilted and strained walk together with reluctance to jump and move is observed in relation to the deformities<sup>[1,4,8-10]</sup>.

In case number 1, a 3,5-year old cat, swelling had been noticed in the left hind leg several weeks before being presented to our clinic and it was established that, as well as the lesions progressing subclinically, the cat lived on a farm and the patient owner saw it very rarely. The owner of the other case (case number 9) with distinctive bilateral exostoses related to osteochondrodysplasia stated that they had also noticed the lesions at a late stage and had brought the cat in for physical examination once the mass on the left side had started obstructing the cat's walk. The fact that no abnormal findings were discovered in the tail structure and extremity length was compatible with data stating that heterozygote cats have less severe lesions <sup>[24,5,11]</sup>that emerge in later stages.

In heterozygote cats, radiologic follow-ups revealed new bone formation to be less widespread and of solitary character compared to homozygote cats. In the authors' opinion, the procedure for removal of new bone formation will present better results in heterozygote cats compared to homozygote cats.

The fact that there is no effective treatment procedure to eliminate osteochondrodysplasia in Scottish Fold cats causes treatment options to remain on a palliative level. Pentosan polysulfate <sup>[12]</sup> and chondroitin sulfate <sup>[6]</sup> have been recommended for cats affected by the disease. Furthermore, in cases accompanied by pain due to arthritis, meloxicam is a non-steriodal anti-inflammatory drug recommended for its safe long-term use <sup>[6,13]</sup>. In another study, the minimally invasive technique of radiotherapy was applied for its prevention of excessive bone formation and suppression of the inflammatory process <sup>[5,14]</sup>. Removal of exostoses can be suggested as a surgical treatment option. In one of the cases in this study, bilateral plantar exostoses were removed and pantarsal arthrodesis performed <sup>[15]</sup>.

It is a fact that patients of this type will begin to suffer from chronic arthritic pain from a very early age. All the cats in this study, whether receiving surgical treatment or conservative treatment, were advised to use a long-term non-steriodal anti-inflammatory drug combined with Omega-3 fatty acid. In addition, the diet was altered by recommending commercial cat food containing joint and cartilage-protecting factors. Setting out from these docile, attractive-faced cats' soft temperament and low-level physical activity, it is erroneous to assume that they can cope with the chronic pain caused by the disease or that they can adapt to an uncomfortable life. In the authors' opinion, the docile nature of these cats is due to the pain from which they suffer.

In conclusion, neither surgical nor conservative curative treatment is possible for osteochondrodysplasia in Scottish Fold cats and quality of life can only be improved using temporary methods.

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