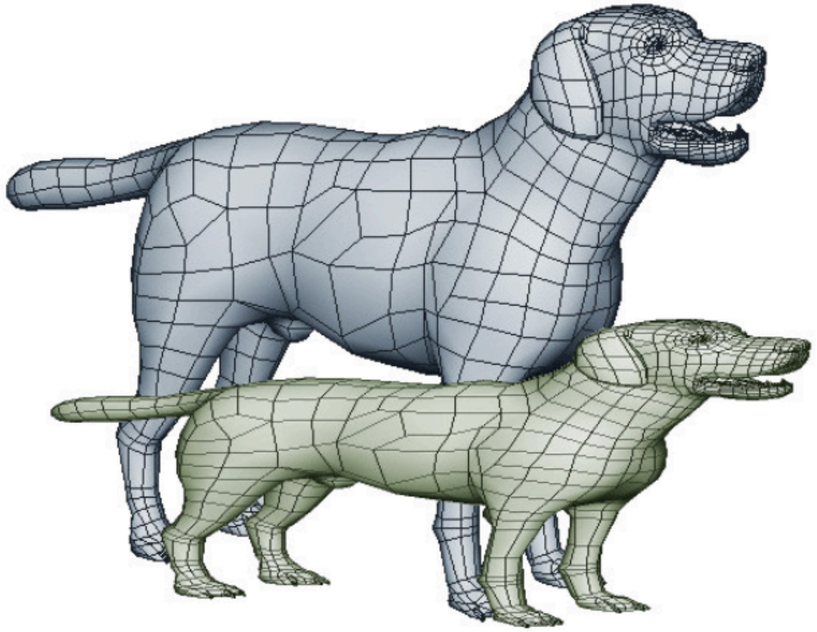
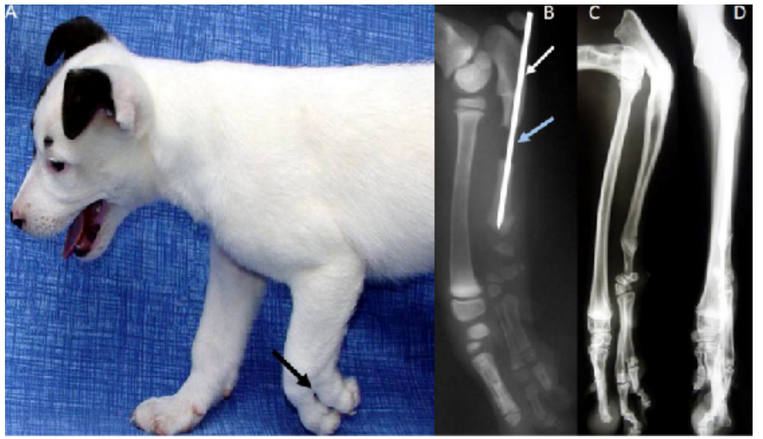
Rare congenital skeletal disorders

Leon, Mexico , 2019



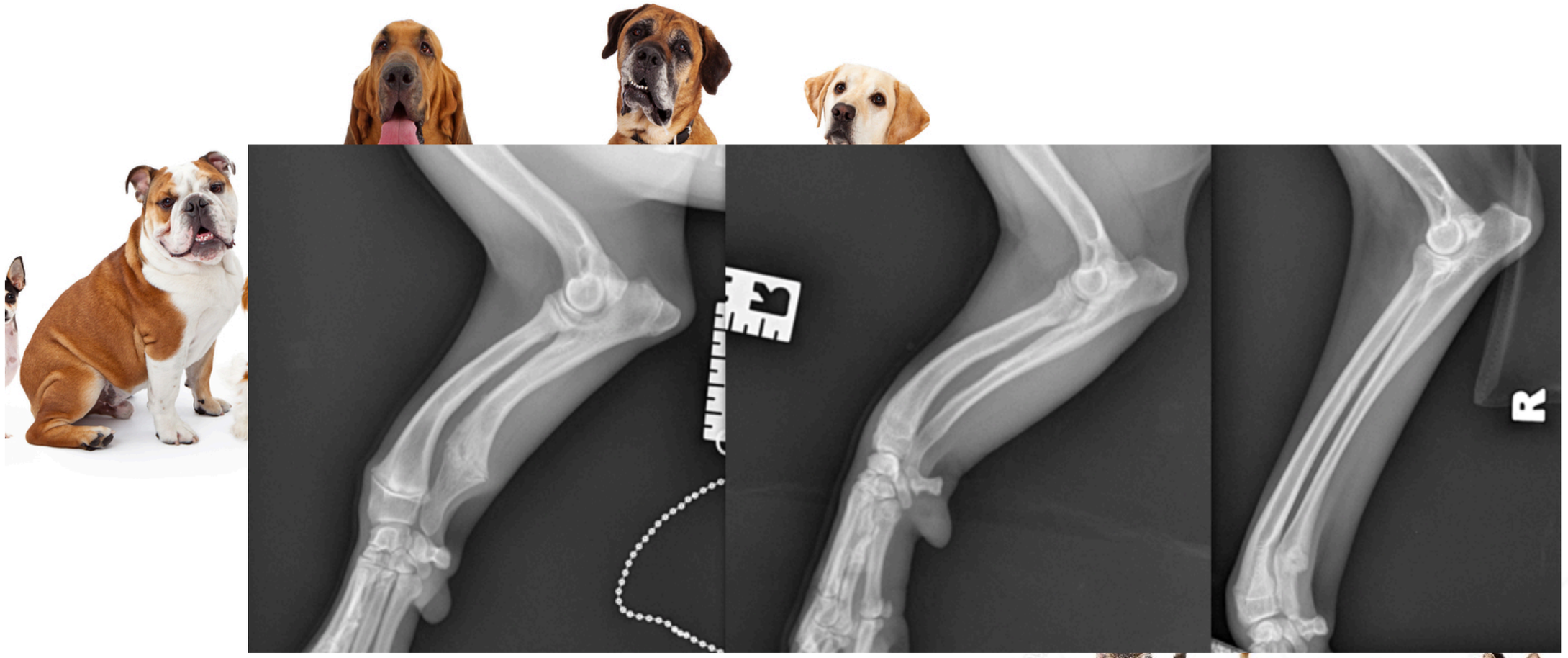


Dr. Vladislav Zlatinov, DVM
Central Vet Clinic- Sofia
Bulgaria

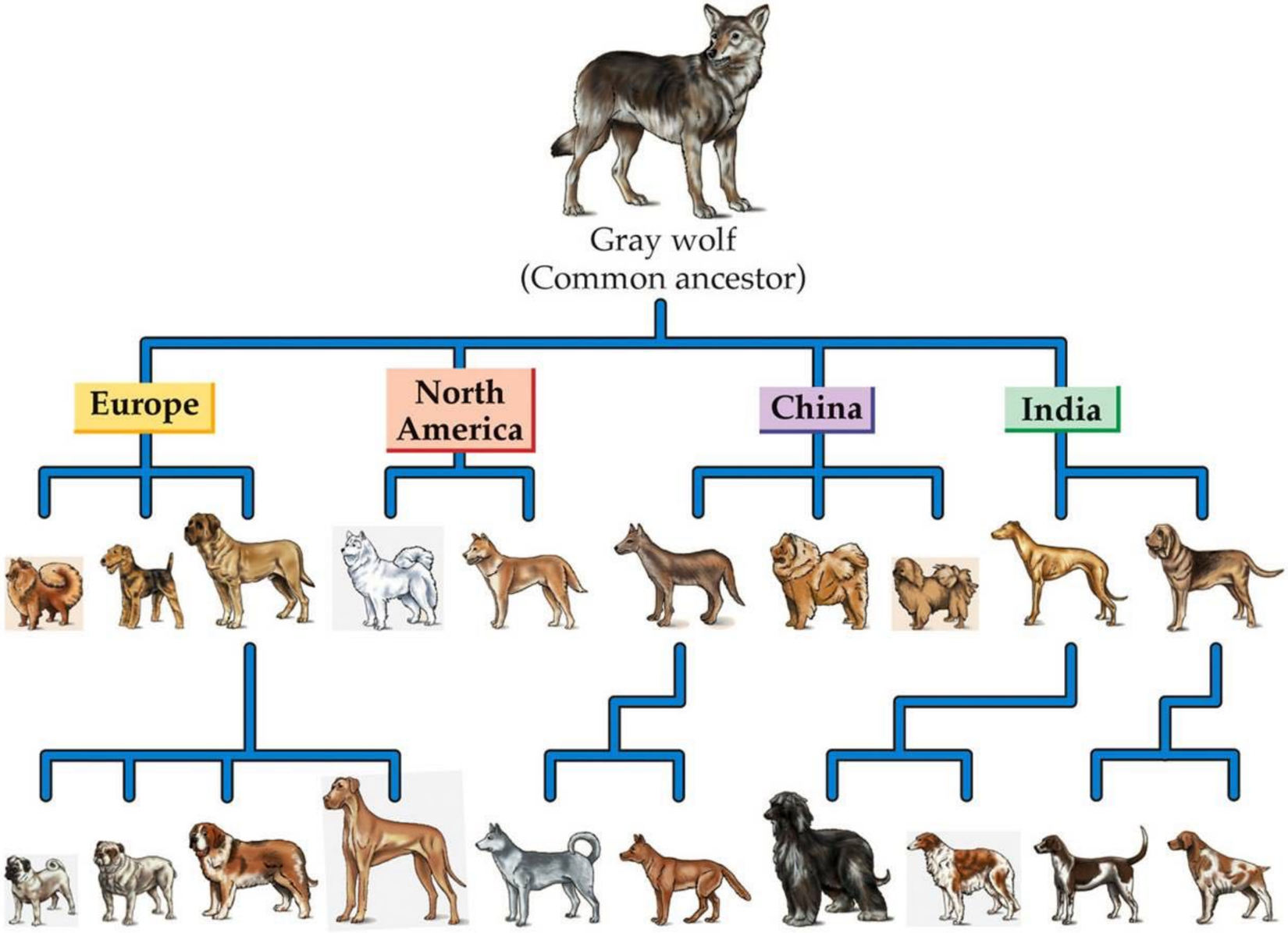
Deformity definition

“Situation in which a part of the body has not developed in the normal way or with the normal shape.”

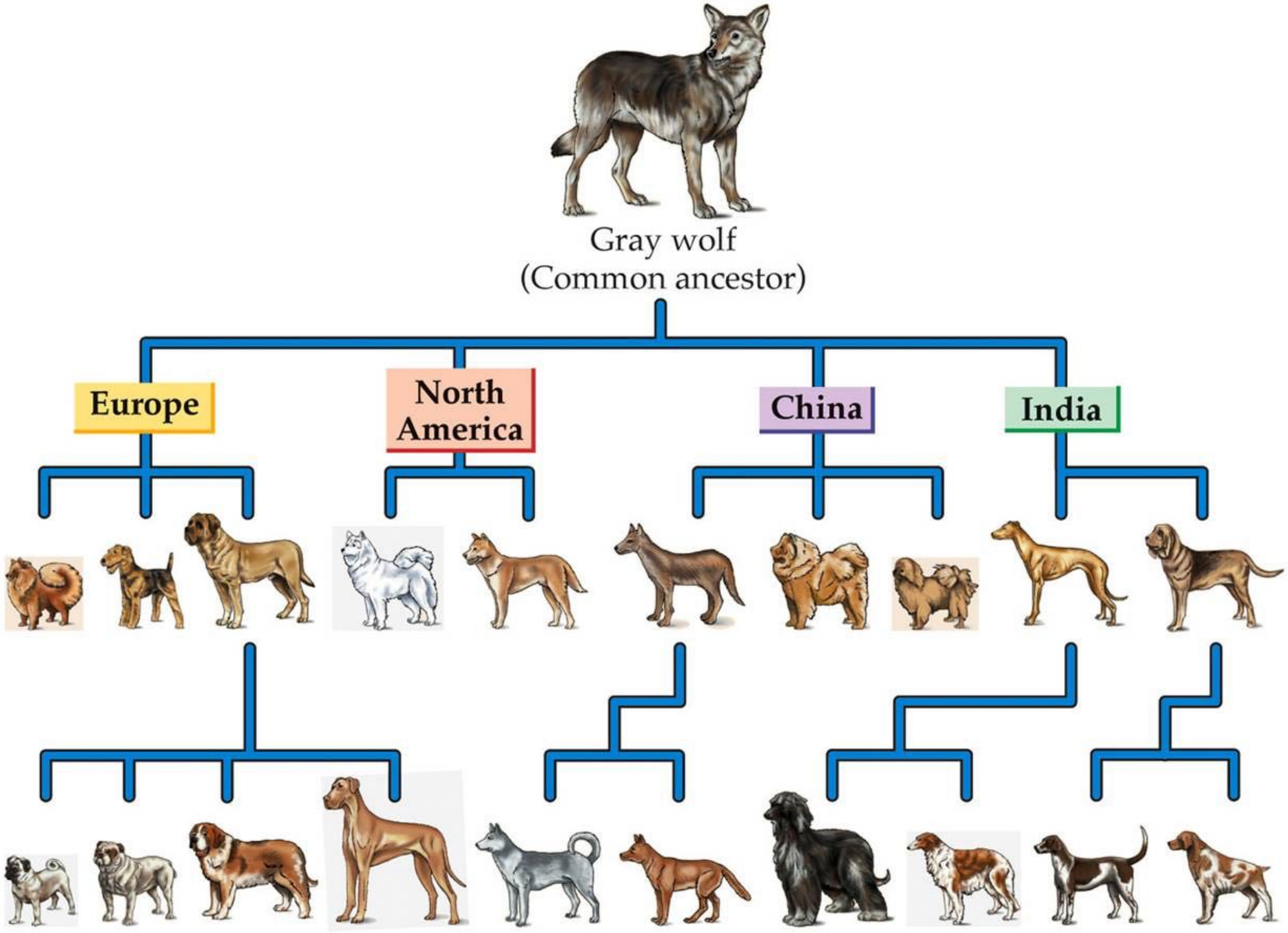
What is NORMAL shape /size?



Breeding breeds



Breeding breeds



Common knowedge

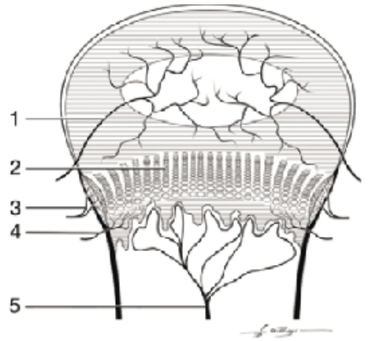
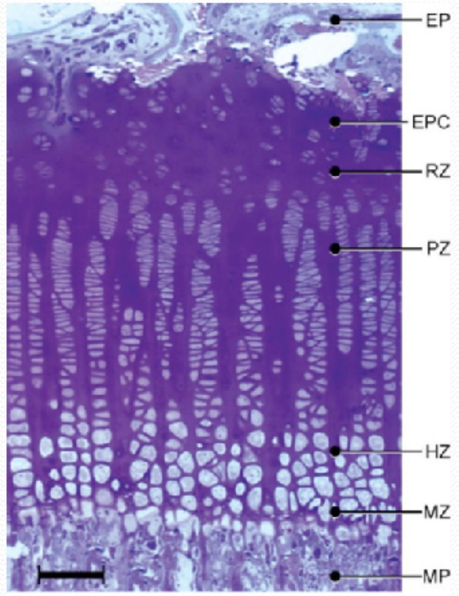
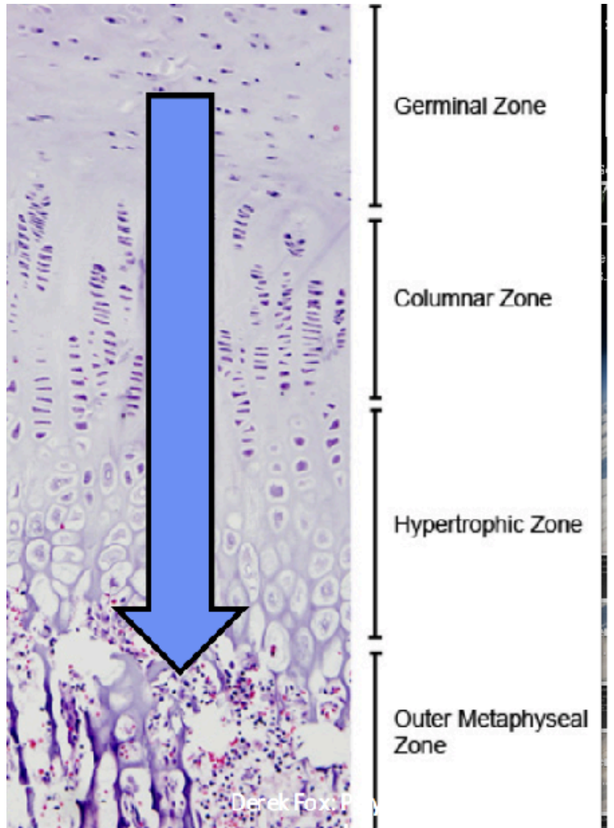


Plate anatomy and physiology

Physeal Growth Plate

- Epiphysis (EP)
- Epiphyseal cartilage (EPC)
- Resting zone (RZ)
- Proliferative zone (PZ)
- Hypertrophic zone (HZ)
- Mineralisation zone (MZ)
- Metaphysis (MP)





Physeal plate growth

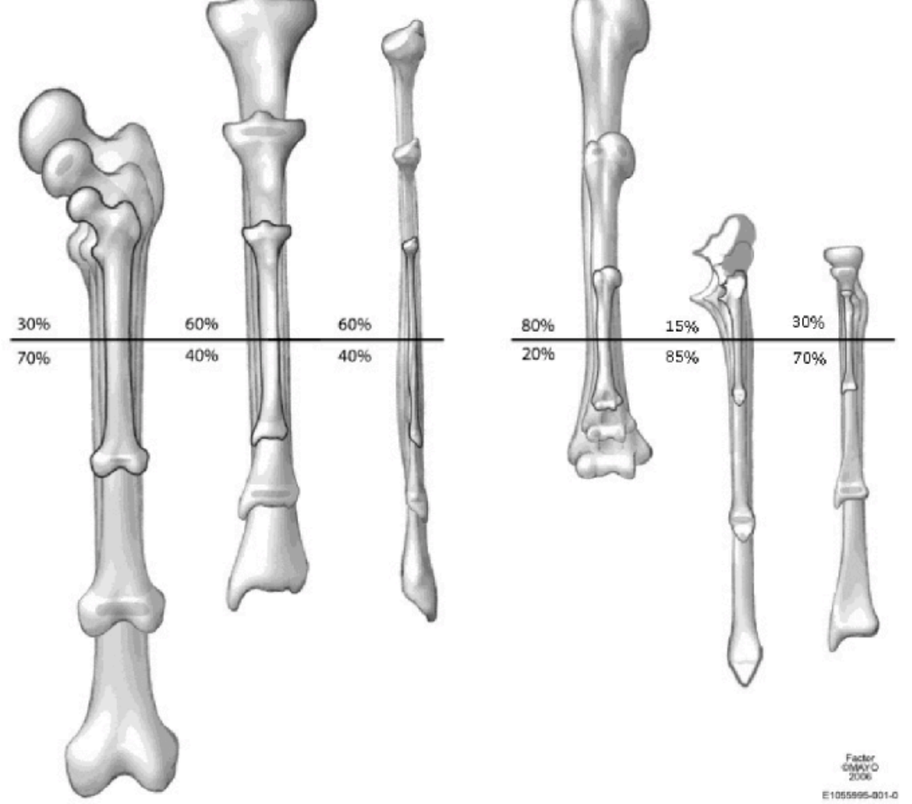
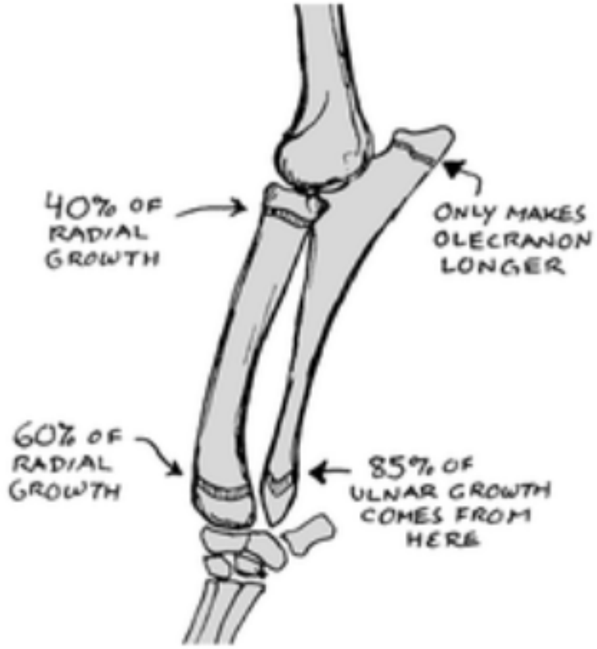


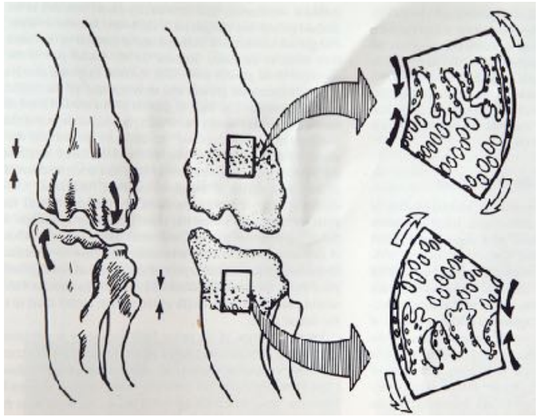
TABLE 3 Approximate Contribution of the Growth Plates to the Growth of Canine Long Bones

Forelimb		Hindlimb	
Growth Plate	**Percentage**	**Growth Plate**	**Percentage**
Proximal humerus	80	Proximal femur	25
Distal humerus	20	Distal femur	75
Proximal radius	40	Proximal tibia	55
Distal radius	60	Distal tibia	45
Proximal ulna	0–15	Proximal fibula	60
Distal ulna	85–100	Distal fibula	40

Physeal plate growth

- **One or two bone systems?**
- **Heuter-Volkmann law/**Wolf's law

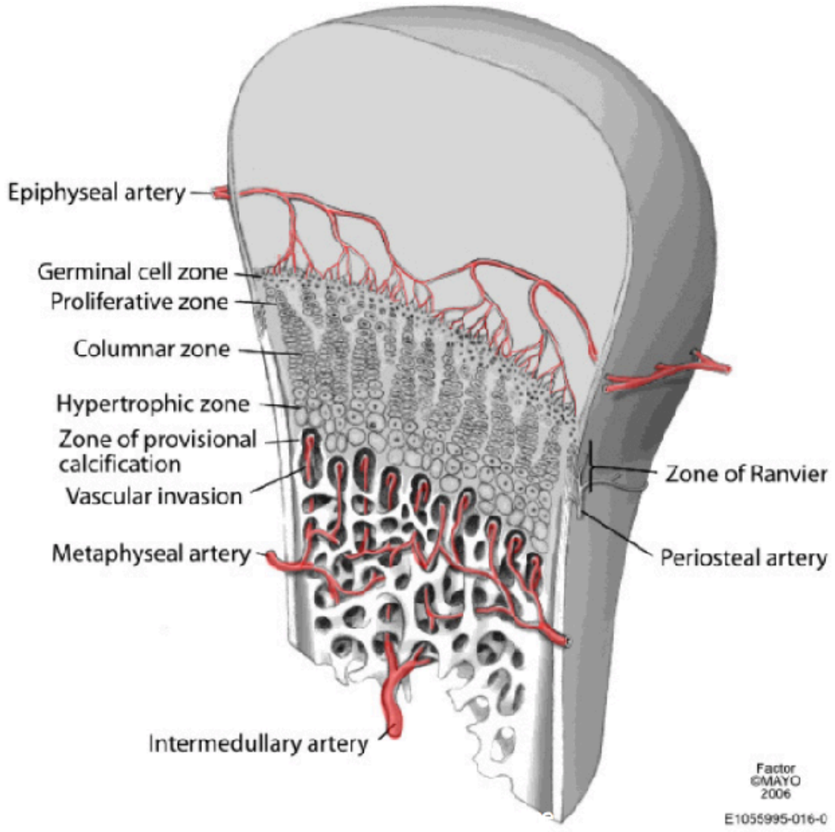




Ethiopathogeneis

Abnormal physeal growth , caused by:

- Metaboloic
- Traumatic
- Developmental
- Inflammatory
- Endocrine
- Genetic



Presentation goals

To focus on some inherited diseases that you may not be familiar with.

Initial approach

A thorough history:

- Age at which the disease became apparent
- Littermates
- Breeding environment
- Exposure to medications
- Maternal health

Thorough examination

Imaging

Knowedge

Genetic (or other) tests

Treatment

-palliative

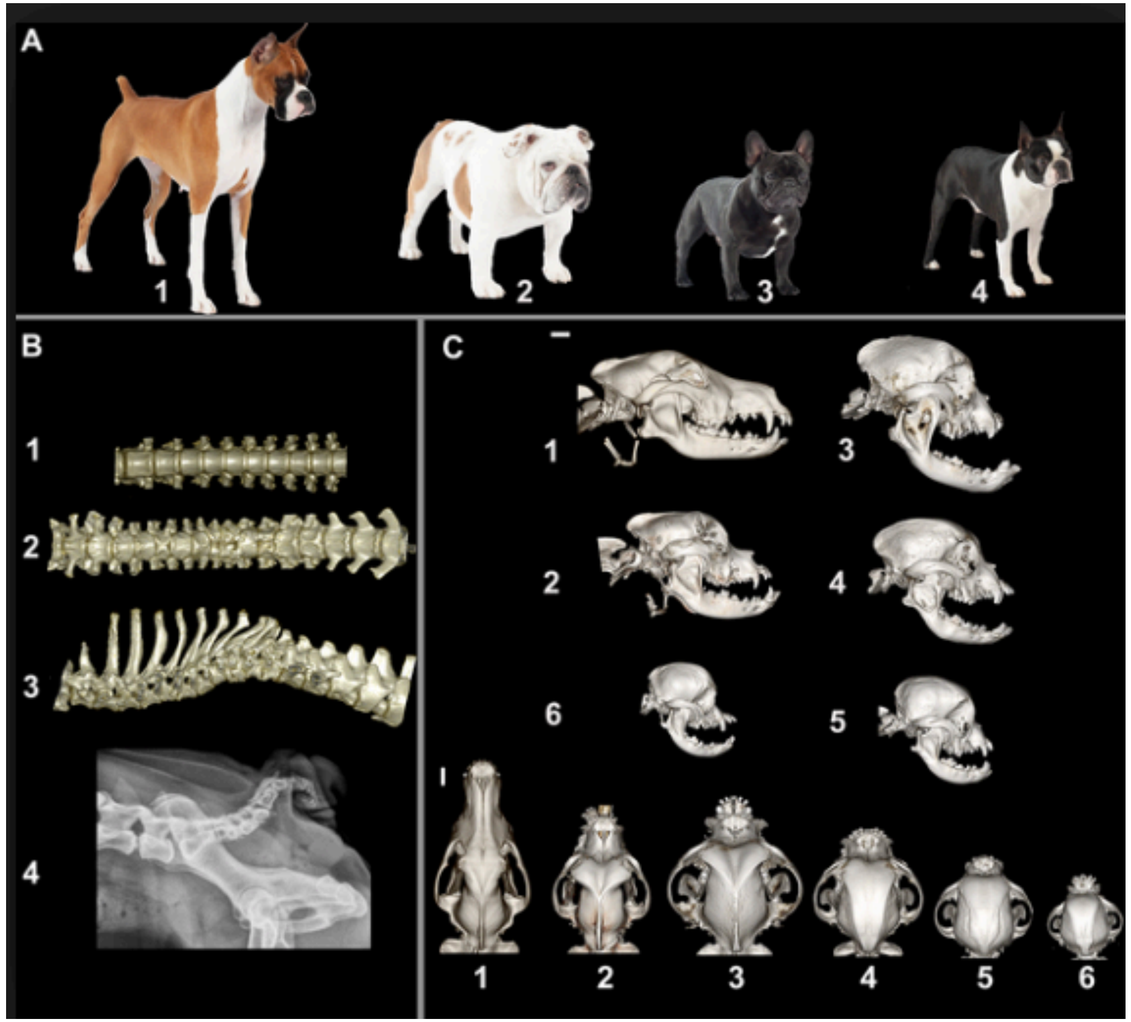
-curative

Dysostoses

- S*ynostosis*= fusion, dysostosis the opposite.
- **Defect in the formation** of a bone or part of a bone.
- **Transformation** of the mesenchymal bone model into **cartilage**, or conversion of cartilage into bone.
- **Spinal or appendicular** skeleton.

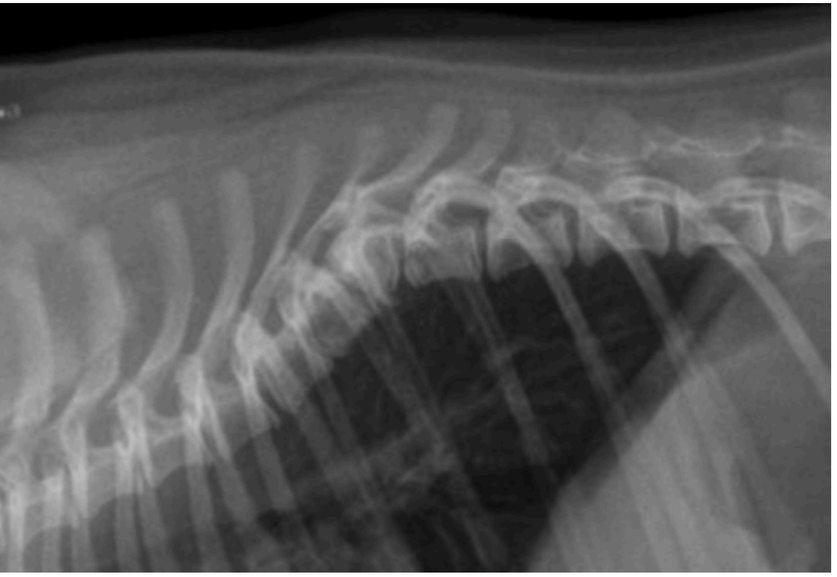
SPINAL dysostoses

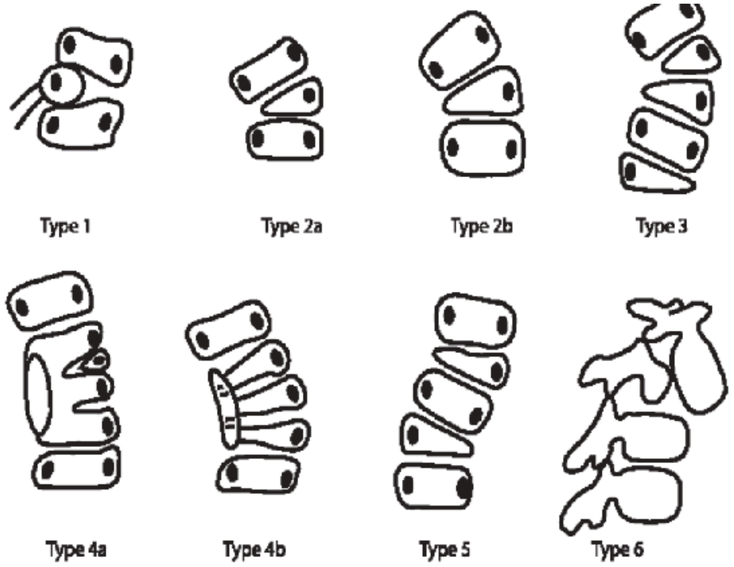
- Hemivertebra
- Block vertebra
- Butterfly vertebra
- Transitional vertebra
- Spina bifida
- Facet aplasia
- Dens malformation

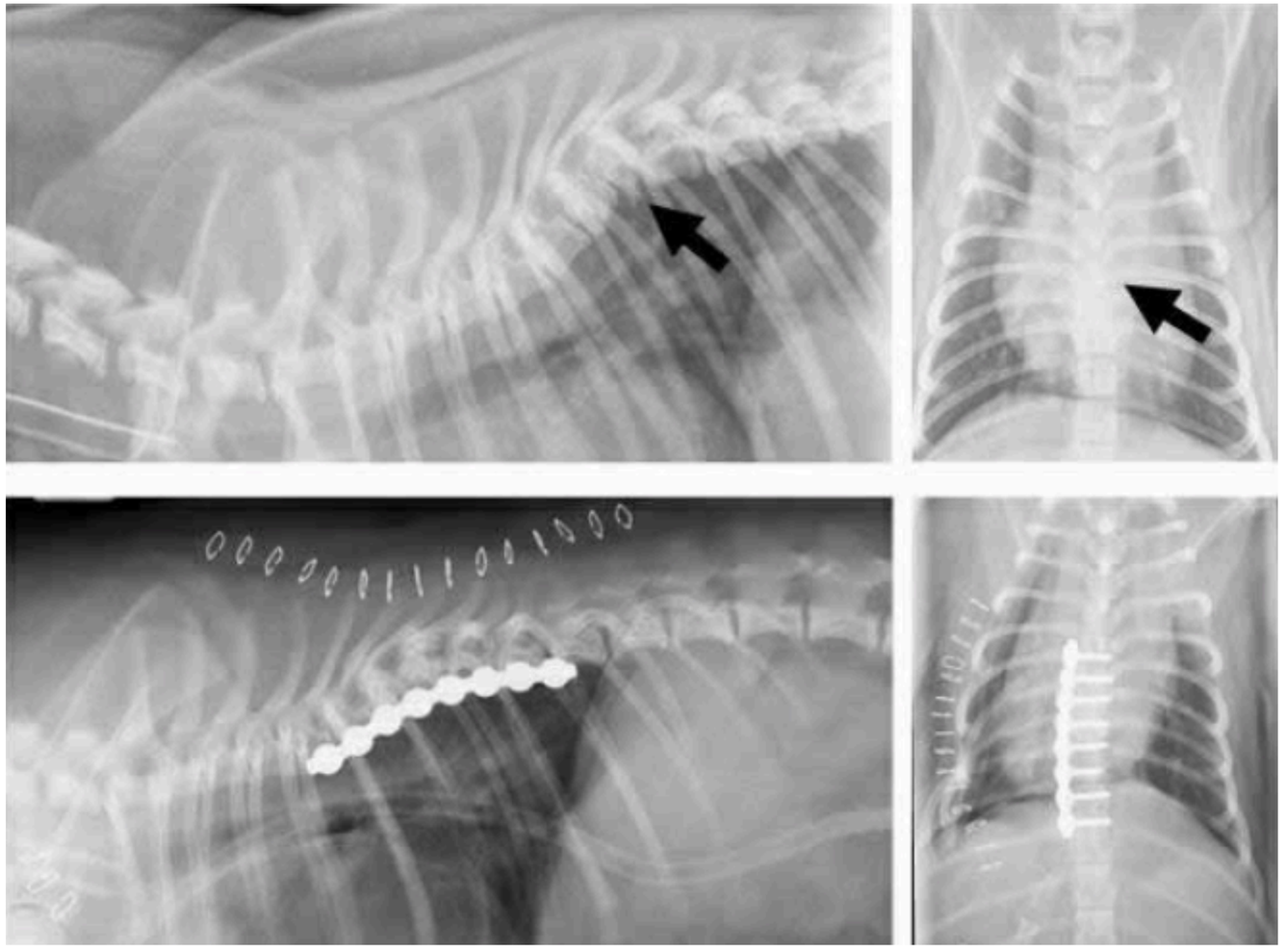


Consequence- varying degrees of spinal cord or nerve root compression

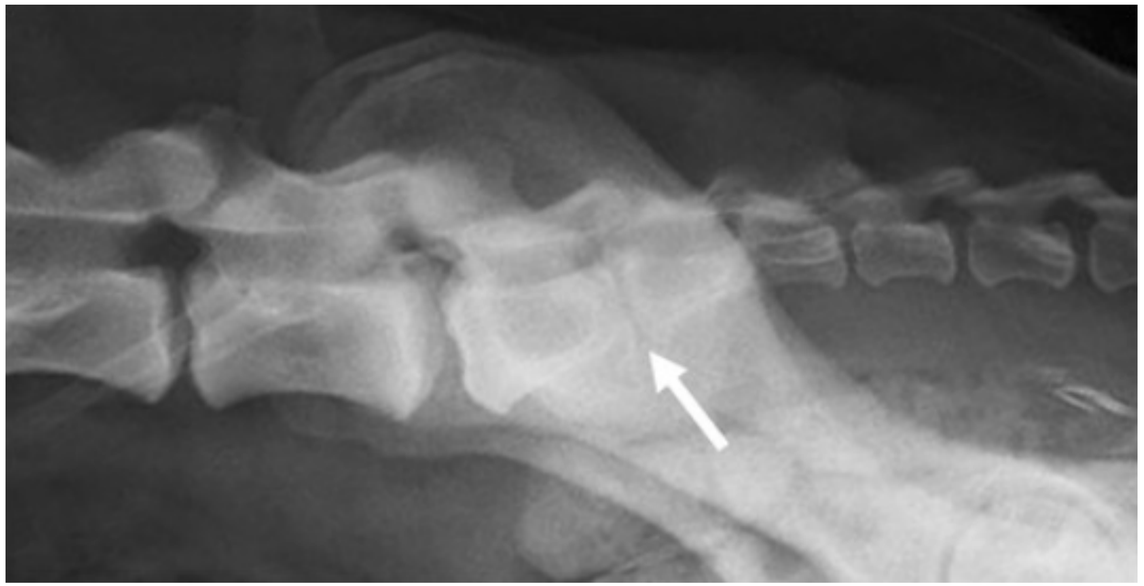
Hemivertebra







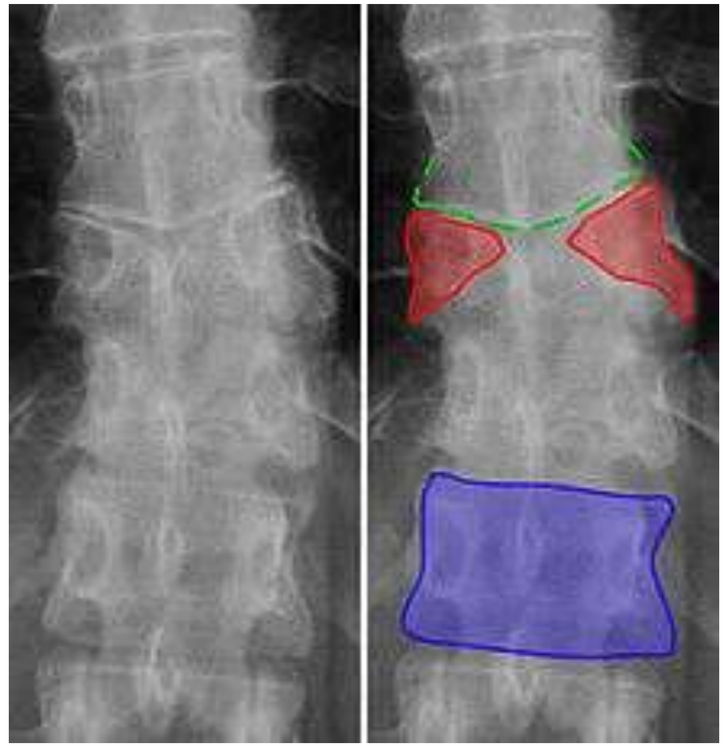
- **Transitional vertebra**



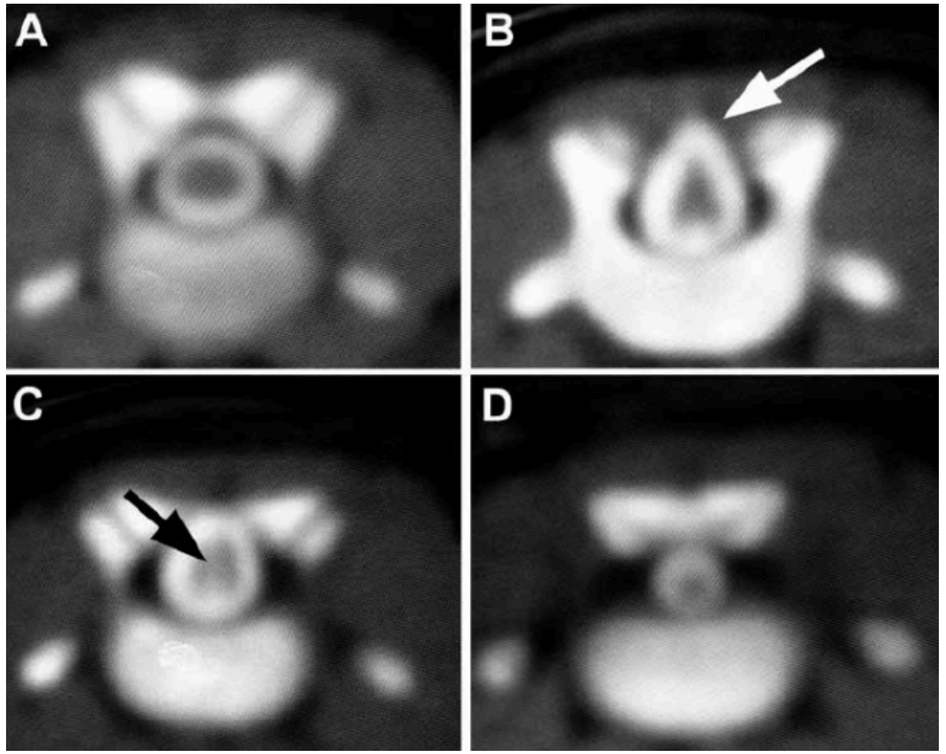
- Block vertebra

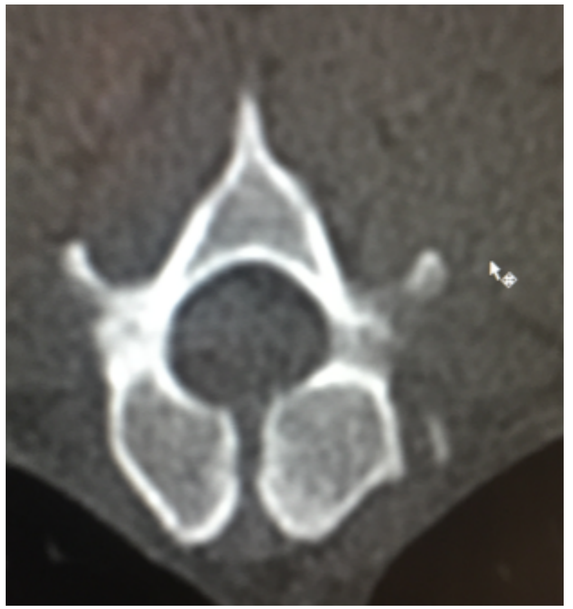
Klippel-Feil – like Syndrome (KFS)

- **Butterfly vertebra**



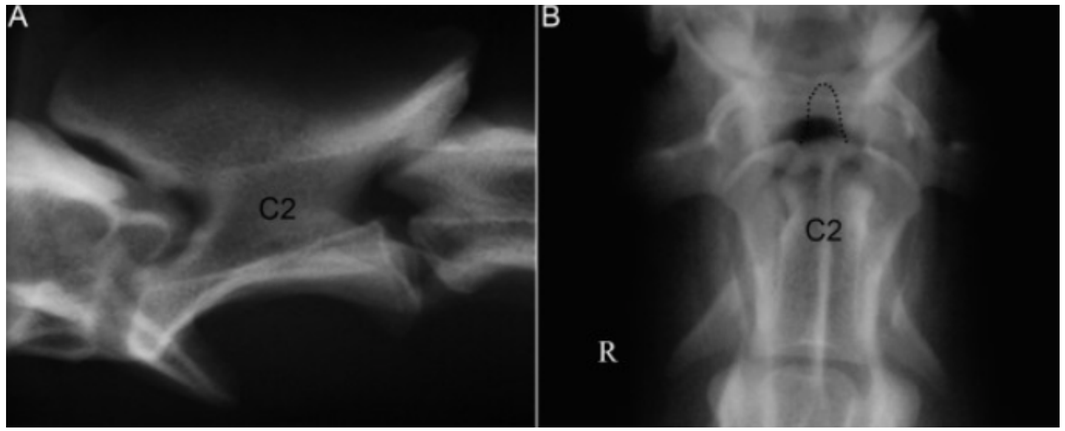
- **Spina bifida**

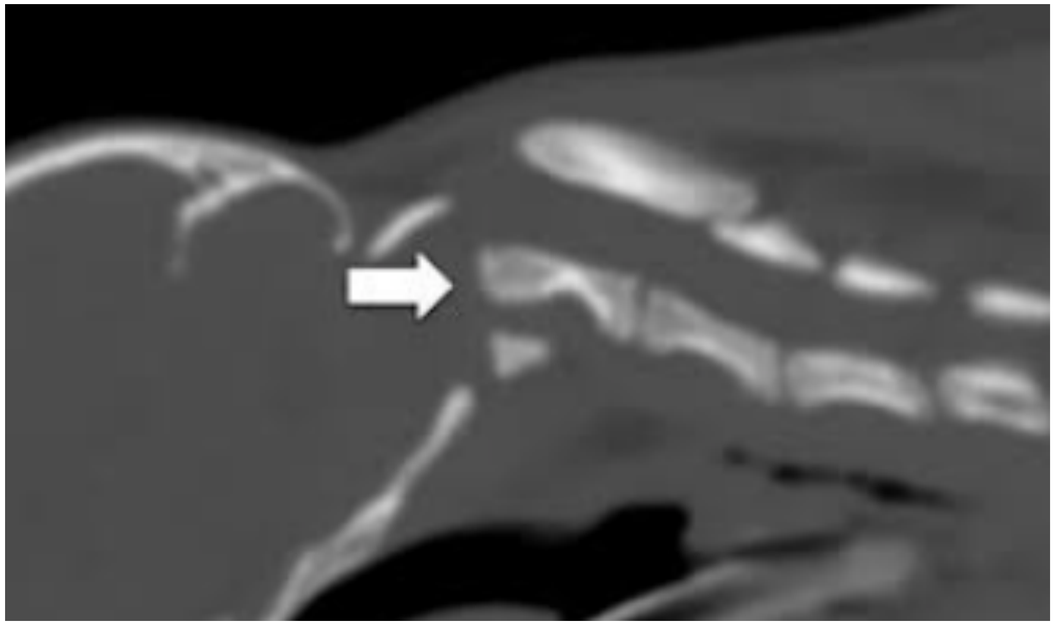




- Combination

- **Dens malformation**



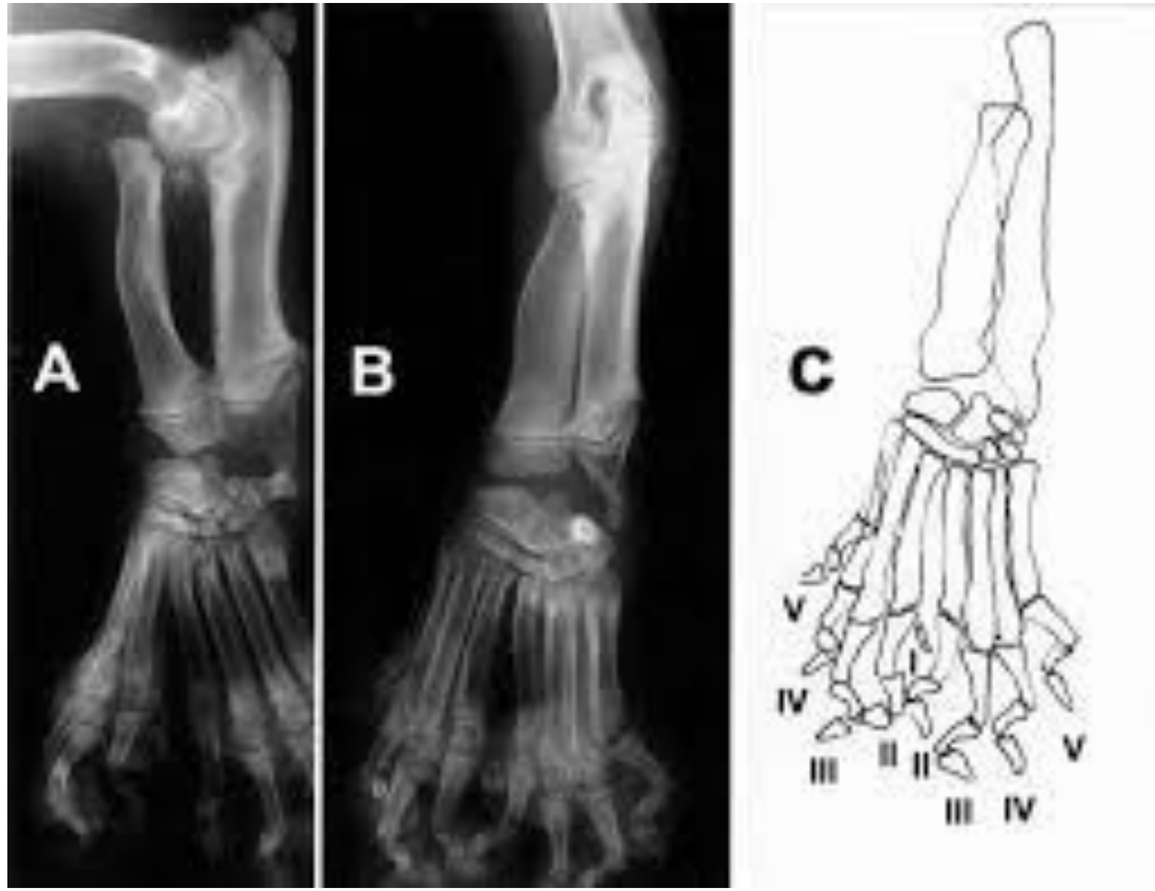


Appendicular dysostoses (Bone agenesis/duplication)

- Amelia
- Hemimelia
- Dimelia
- Ectrodactyly
- Polydactyly
- Syndactyly

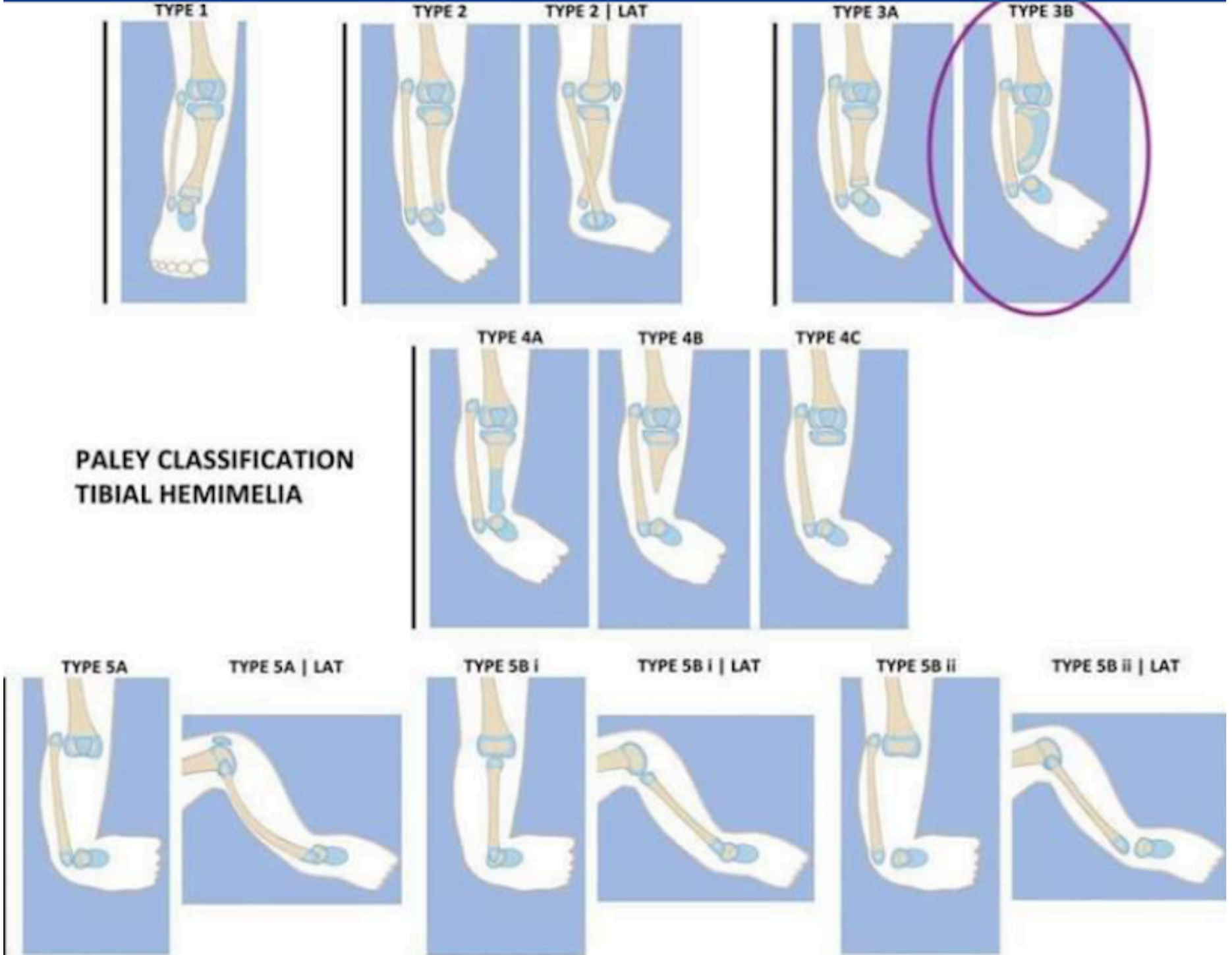
APPENDICULAR DYSOSTOSIS	DEFECT
Amelia	Absence of 1 or more limbs; frequently have concurrent life-threatening conditions
Hemimelia	Complete or partial absence of 1 or more bones
Dimelia	Duplication of entire, or part of, limb
Ectrodactyly	Digital cleft between metacarpal bones
Polydactyly	1 or more extra digits
Syndactyly	Lack of differentiation between 2 or more digits

- **Dimelia (duplication)**



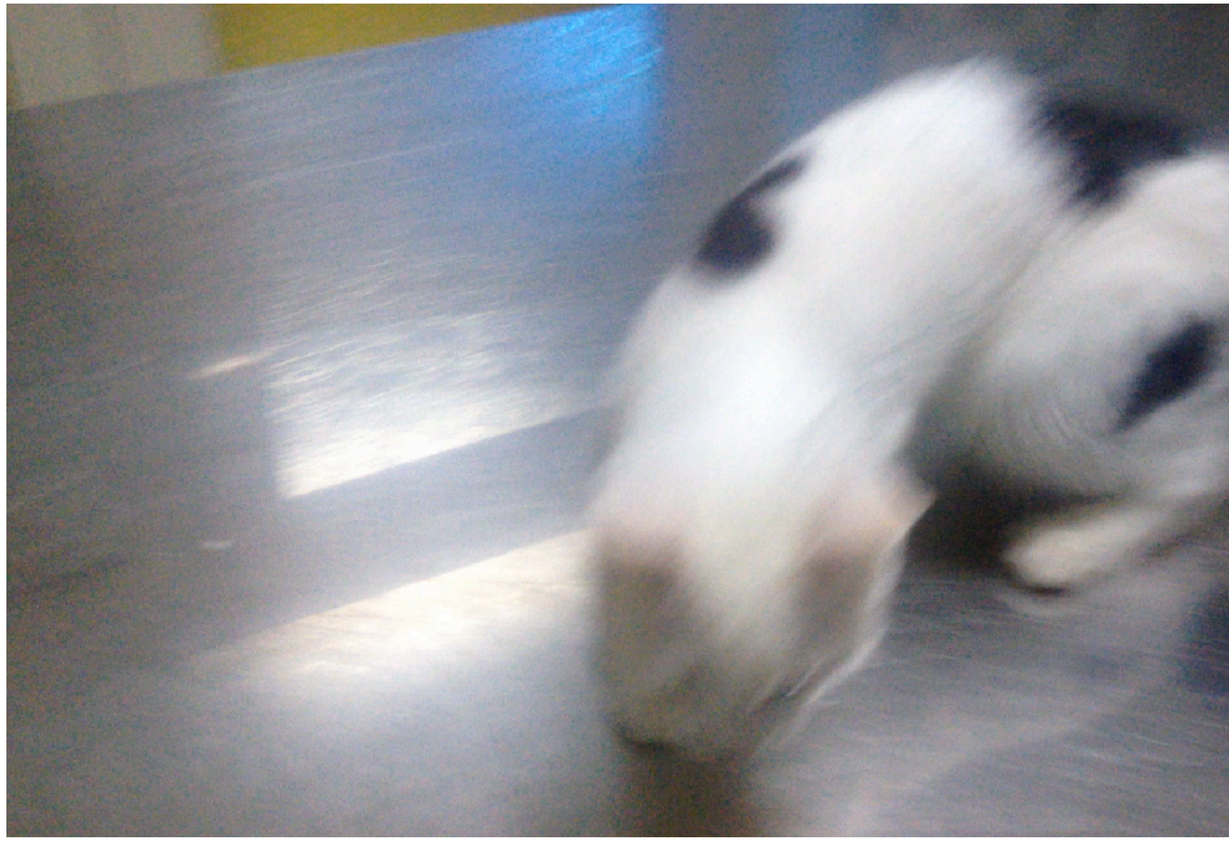
Hemimelia

- Complete radial agenesis


TYPE 1
TYPE 2
TYPE 2 | LAT
TYPE 3A
TYPE 3B
PALEY CLASSIFICATION
TIBIAL HEMIMELIA
TYPE 4A
TYPE 4B
TYPE 4C
TYPE 5A
TYPE 5A | LAT
TYPE 5B i
TYPE 5B i | LAT
TYPE 5B ii
TYPE 5B ii | LAT

Hemimelia

Complete radius agenesis ,
4 m old ESH kitten



Hemimelia

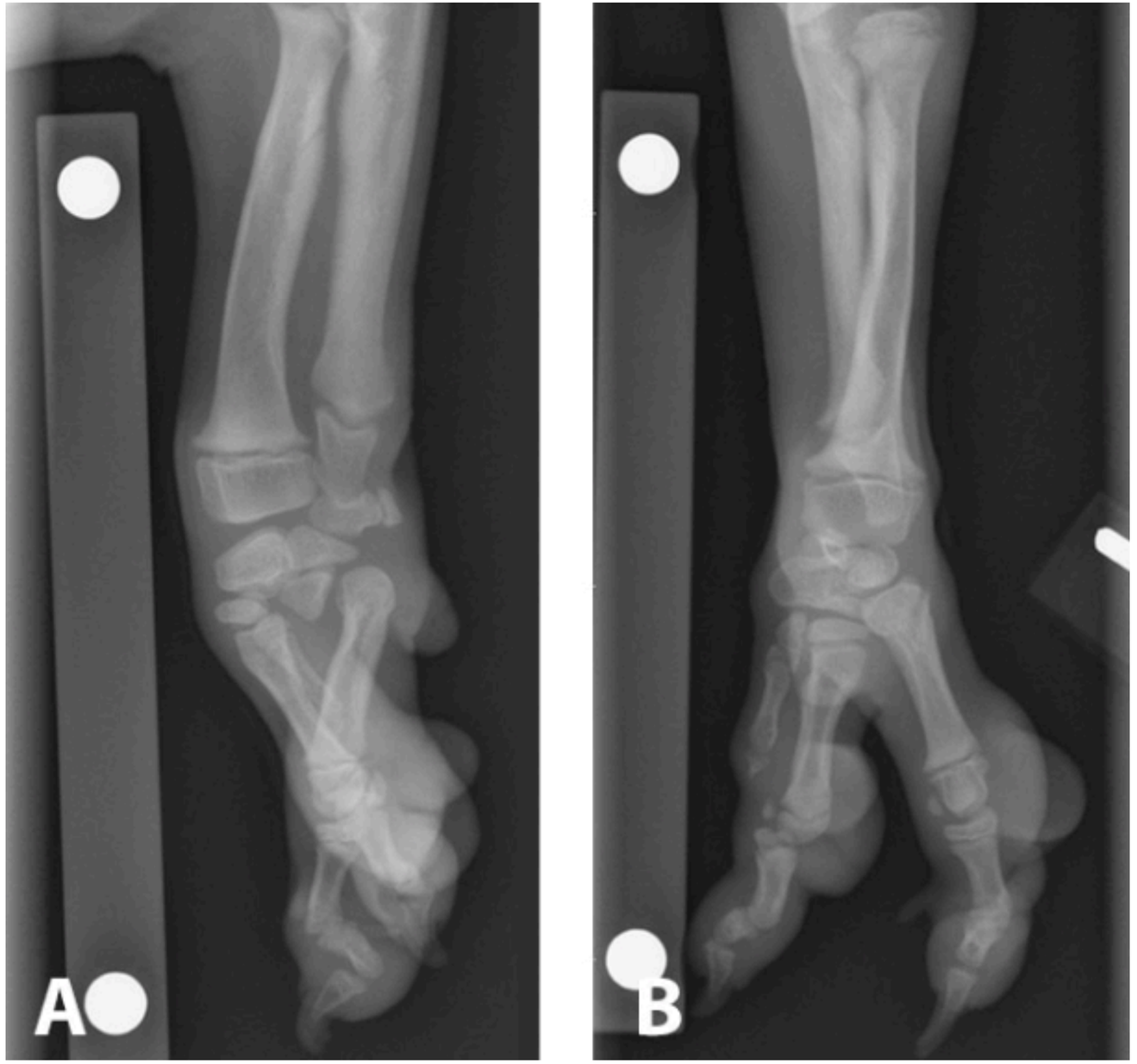
Partial Radius agenesis,
6 m old dachshund

Hemimelia

Calcaneus agenesis
1 y old ESH cat

Ectrodactyly

Terminal Intercalary hemimelia



Treatment

- **Medical**

-Physical rehabilitation- alleviate pain, build muscle, and regain function.
-Splinting -ectrodactyly or hemimelia

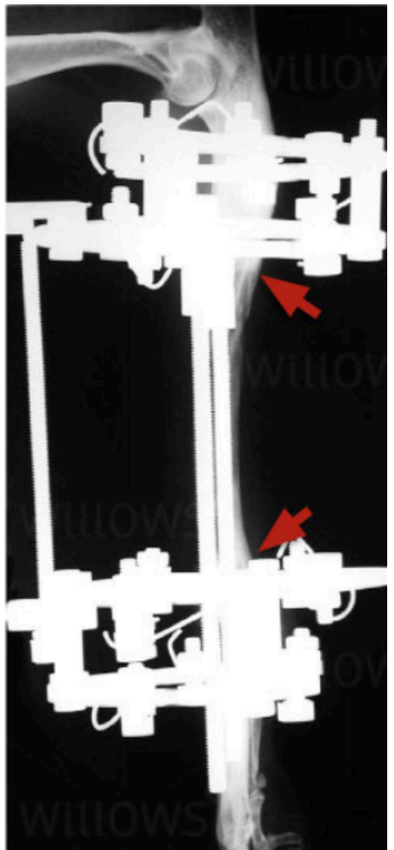
- **Surgery- palliative or reconstructive**

-Palliative- total or partial amputation

-Reconstruction- limb salvage, realignment, frequently, arthrodesis

- **Neutering!**

Specialist Services
- Arranging a Referral
- Pet Health Information
- Cardiology
- Dermatology
- Internal Medicine
- Joint Replacement
- Neurology
- Oncology



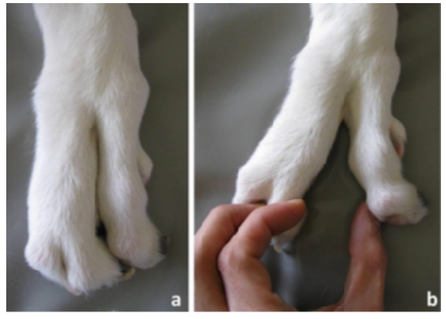
Ankara Üniv Vet Fak Derg, **64**, 229-233, 2017

Short Communication / *Kısa Bilimsel Çalışma*

Surgical management of ectrodactyly in a Turkish Kangal dog

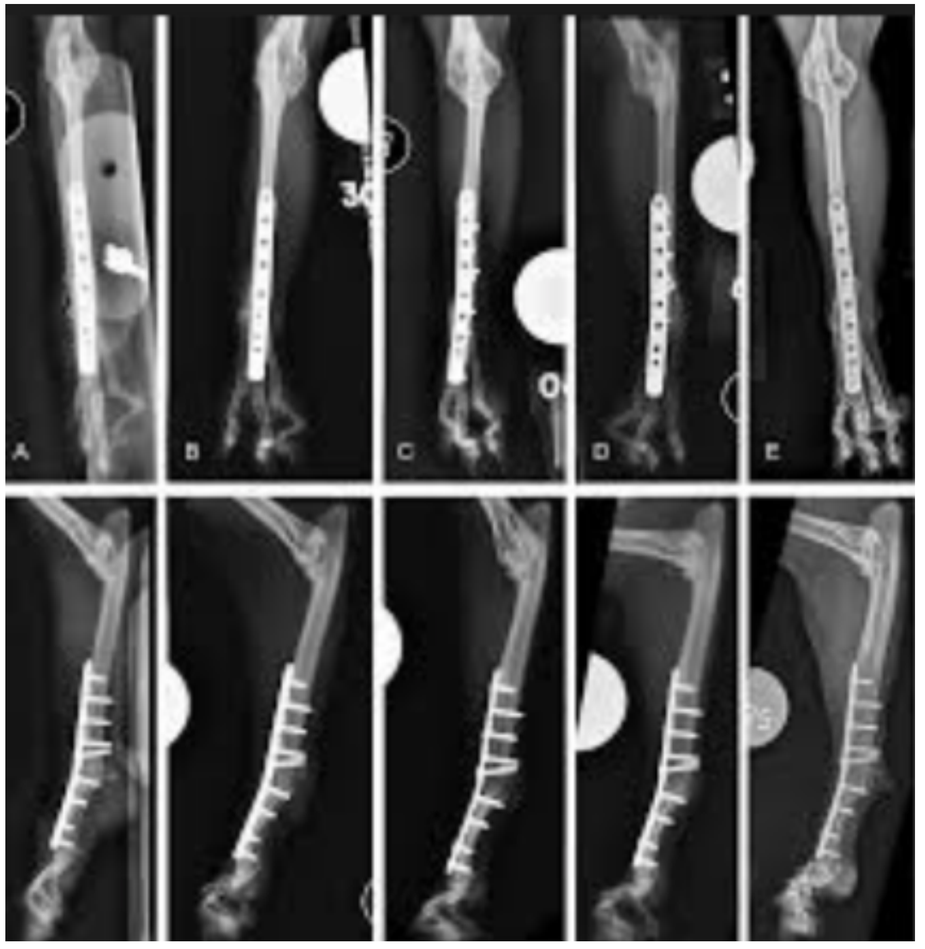
Cenk YARDIMCI, Taylan ÖNYAY, Kamil Serdar İNAL, Birsen Deniz ÖZBAKIR, Ahmet ÖZAK

Ondokuz Mayıs University, Faculty of Veterinary Medicine, Department of Surgery, Samsun, Turkey.

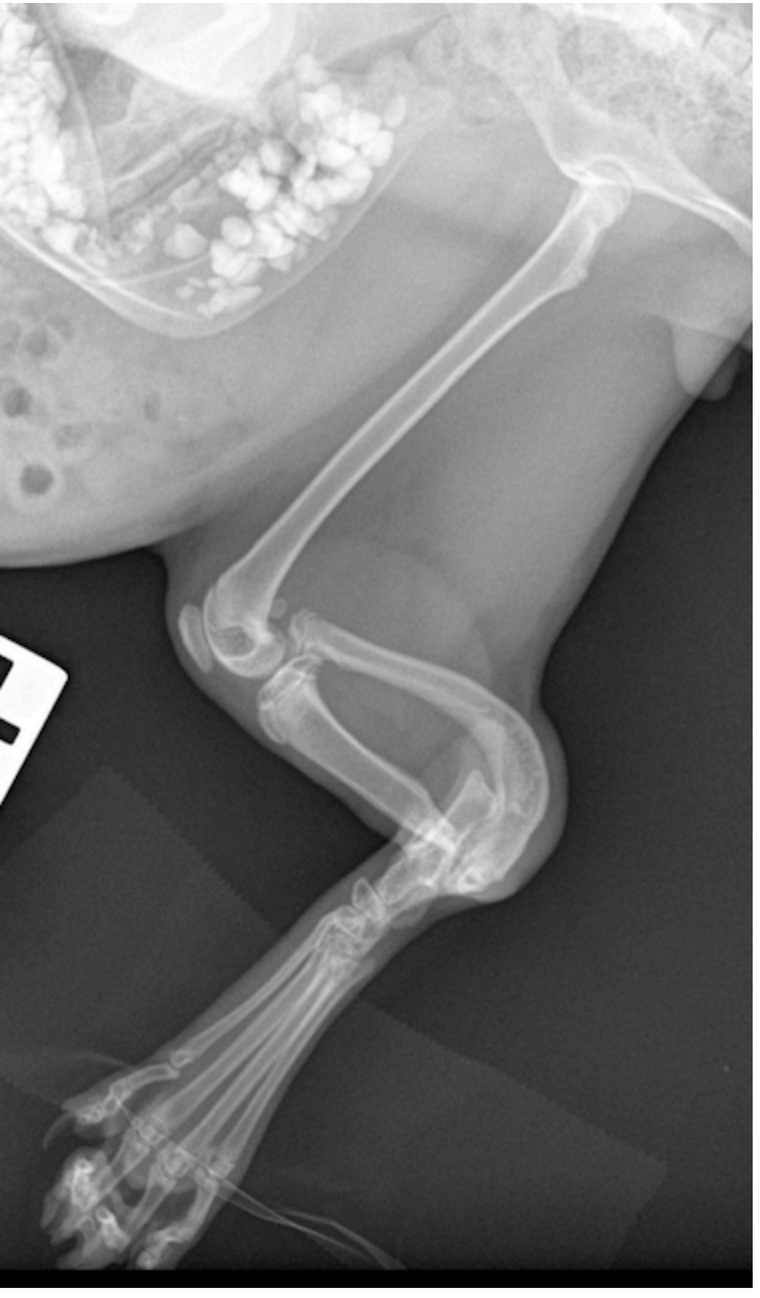


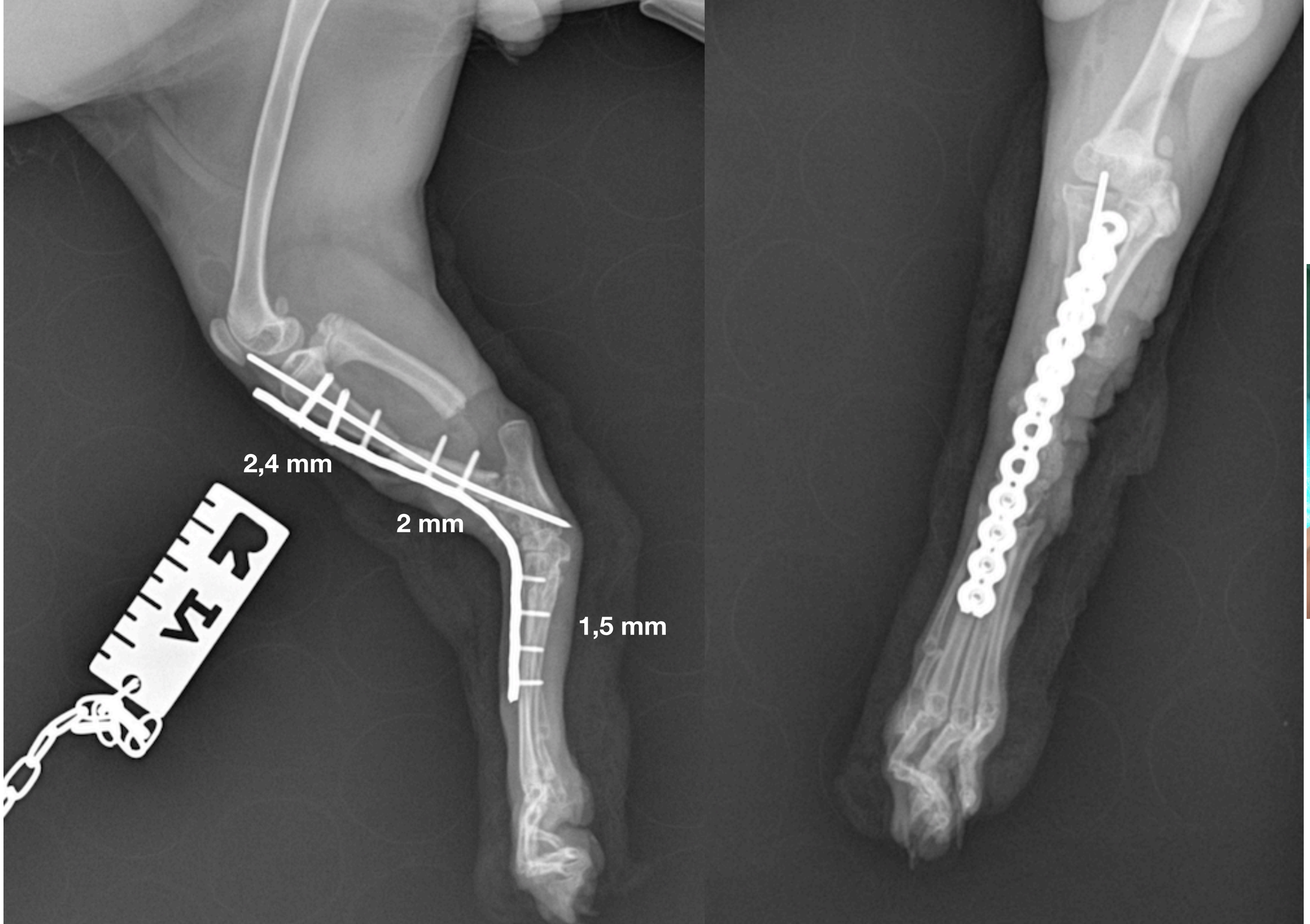
Bilateral Radial Agenesis in a Cat Treated with Bilateral Ulnocarpal Arthrodesis

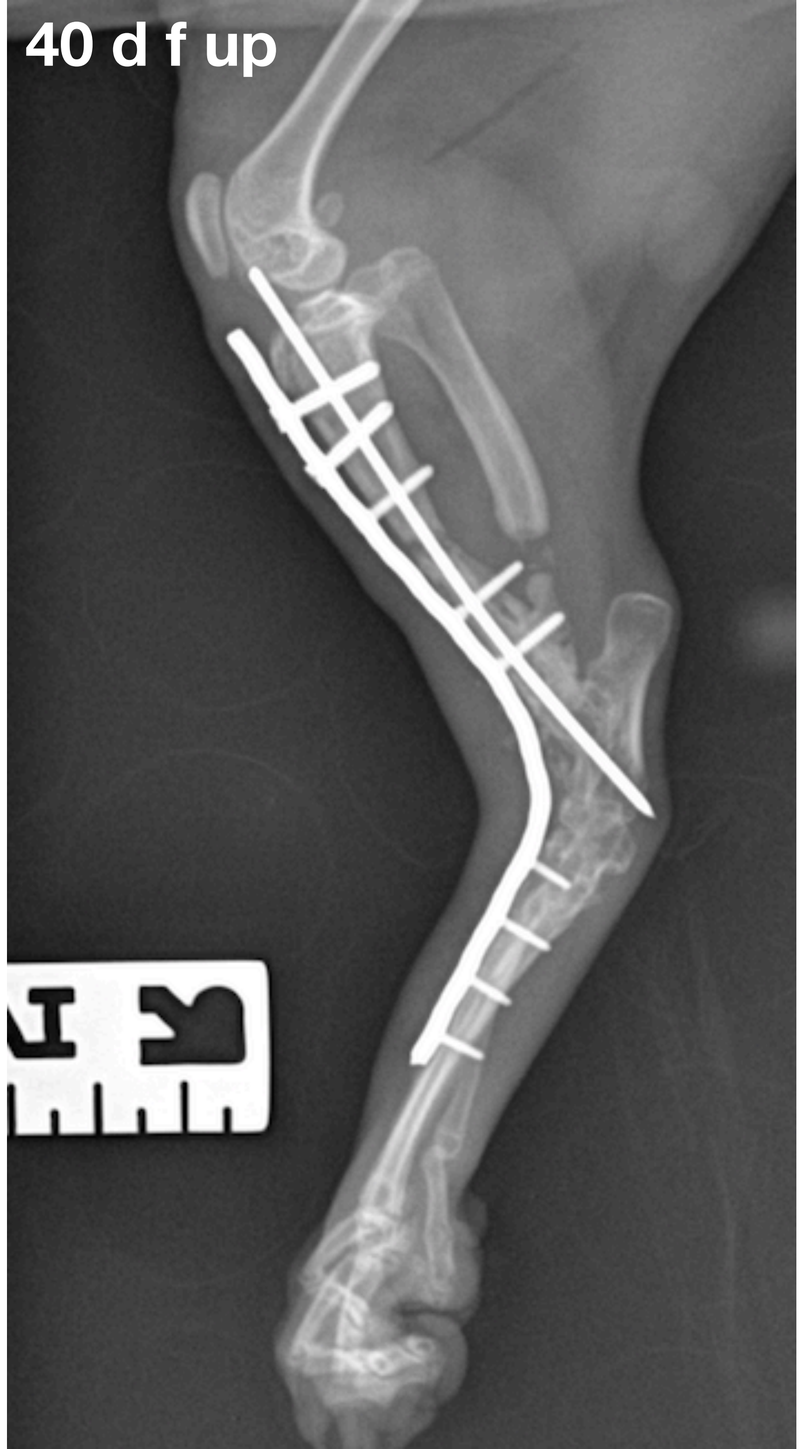
Alla Bezhentseva[1] Harpreet Singh[1] Randy J. Boudrieau[1]



Riko,
4 m old ESH
Deformed hind legs-
unable to walk on them at all.




2,4 mm
2 mm
1,5 mm


40 d f up


The other limb

60 days f up

Osteochondrodysplasias

- Group of cartilage and bone disorders => defective endochondral ossification.

Osteochondrodysplasias

- Group of cartilage and bone disorders => defective endochondral ossification.
- An autosomal recessive gene.
- The result- slowed growth and small stature.
- Dwarfism
  - proportionate
  - disproportionate

- Intentional osteochondrodysplasia - certain breeds.

Canine & Feline Osteochondrodysplasia[1,3]

BREED	TRAIT	MODE OF INHERITANCE
Akita	Achondrogenesis	Unknown
Alaskan malamute	Chondrodysplasia	Simple autosomal recessive
Beagle	Chondrodysplasia punctata	Unknown
	Multiple epiphyseal dysplasia	Simple autosomal recessive
	Osteogenesis imperfecta	
Bulldog	Osteochondrodysplasia	Unknown
Bull terrier	Osteochondrodysplasia	Unknown
Cocker spaniel	Hypochondrodysplasia	Unknown
Collie	Osteogenesis imperfecta	
Dunker	Multiple epiphyseal dysplasia	Unknown
Great Pyrenees	Chondrodysplasia	Simple autosomal recessive
Hygenhund	Multiple epiphyseal dysplasia	Unknown
Irish setter	Hypochondrodysplasia	Simple autosomal recessive
Labrador retriever	Oculoskeletal dysplasia	Simple autosomal recessive
Miniature poodle	Achondrodysplasia	Simple autosomal recessive
	Multiple epiphyseal dysplasia	Unknown
	Pseudoachondrodysplasia	Unknown

Diagnosis

Radiographic assessment:

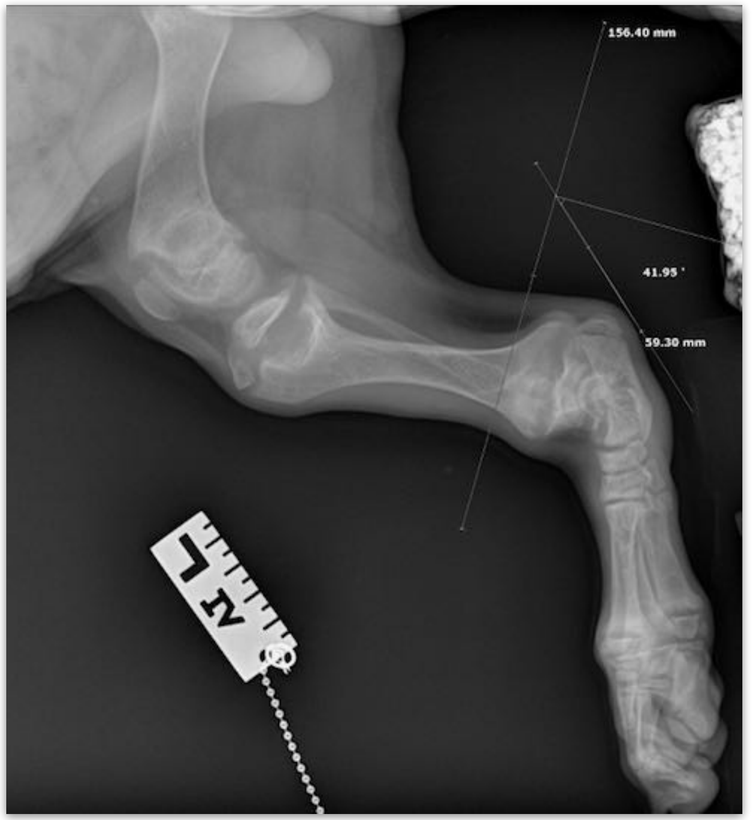
- abnormal growth plate morphology, reduced length of the axial/appendicular skeleton.
- lumbar spine
- radius/ulna

If endocrinopathy is suspected:

Serum T4
TSH
Growth hormone (stimulation test)
Insulin-like growth factor-1 serum concentrations.

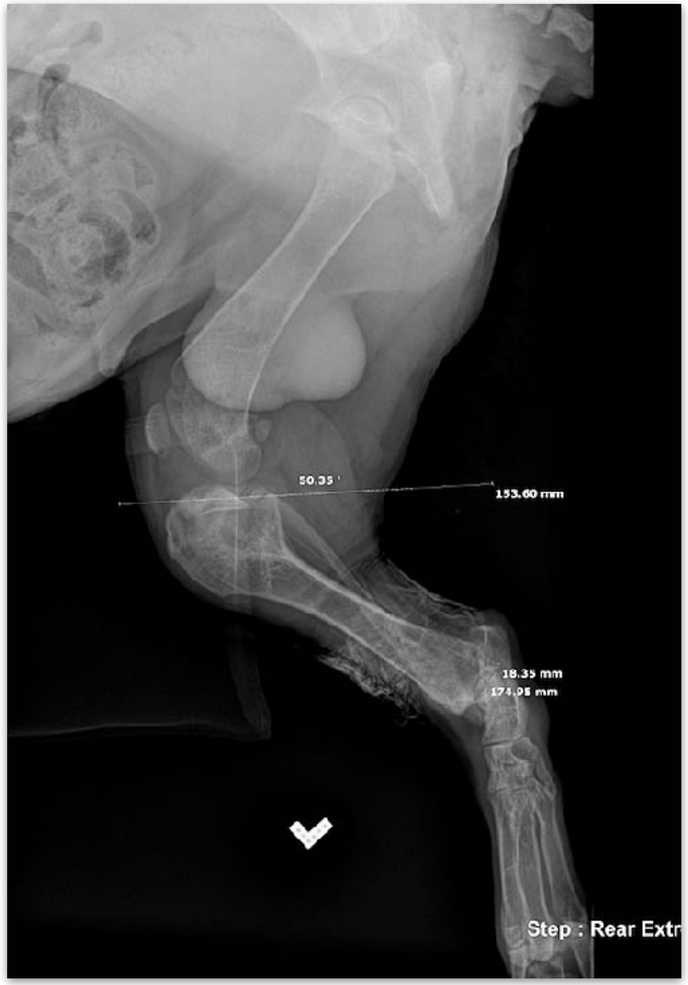
Chondrodysplasias

4 m lab



Osteochondrodysplasias

11 month of age, partially torn cruciate, excessive tibial slope



Treatment

- Paliation of clinical signs.
- Neutering

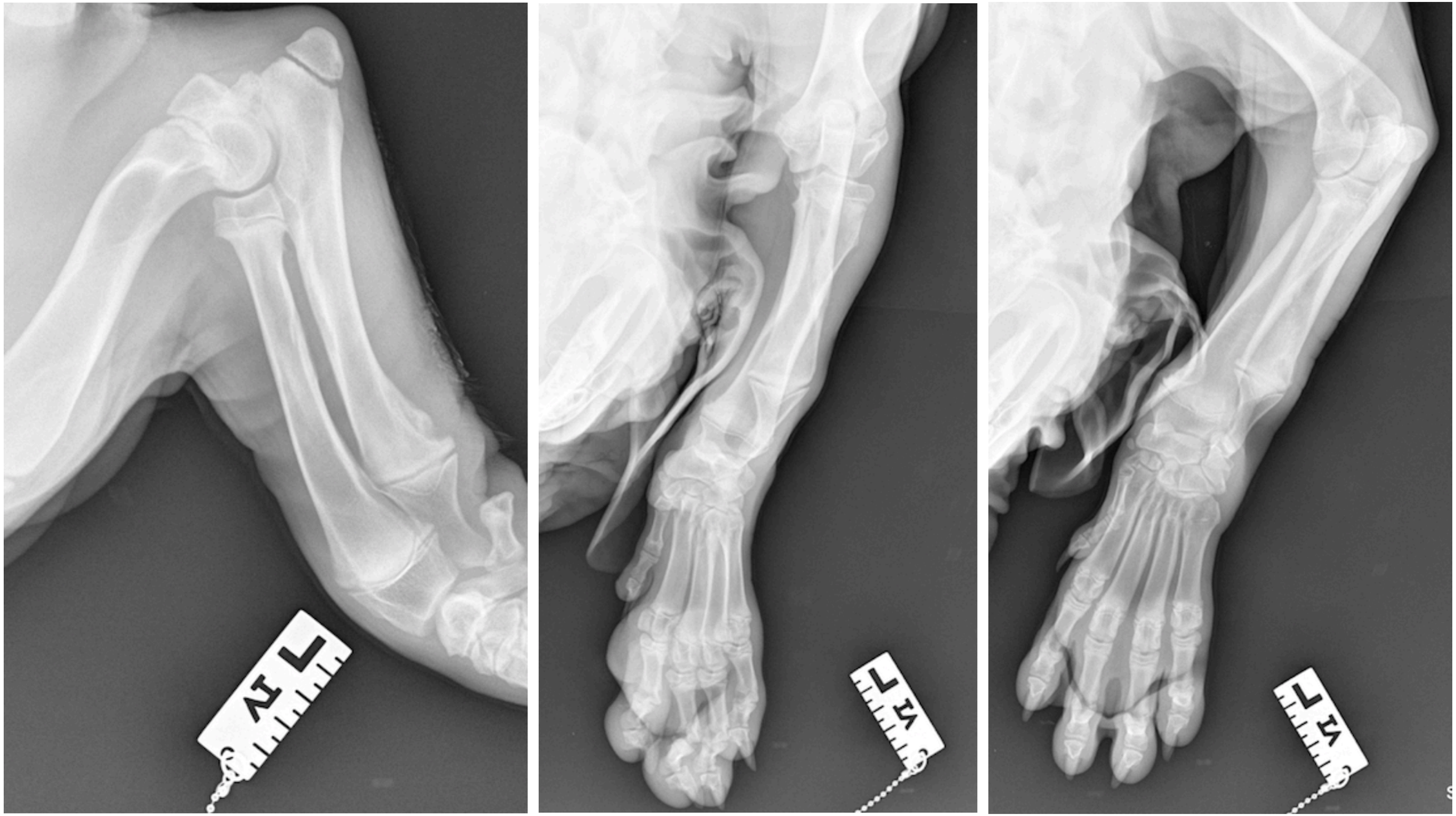
Archi

4 m old male cane corso
27 kg

“Crooked front legs”

Chondrodysplstic?

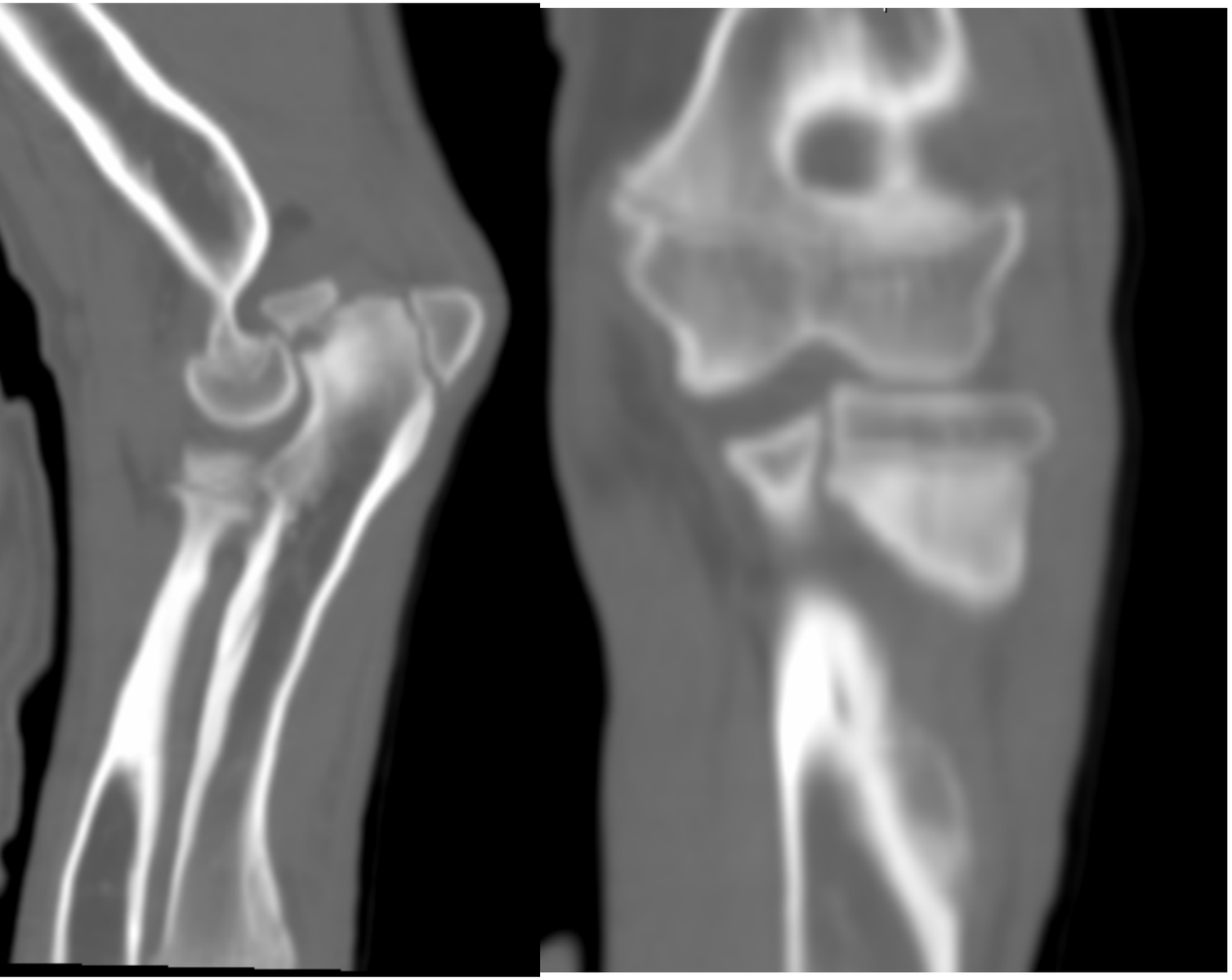
Retained ulna growth?



UAP

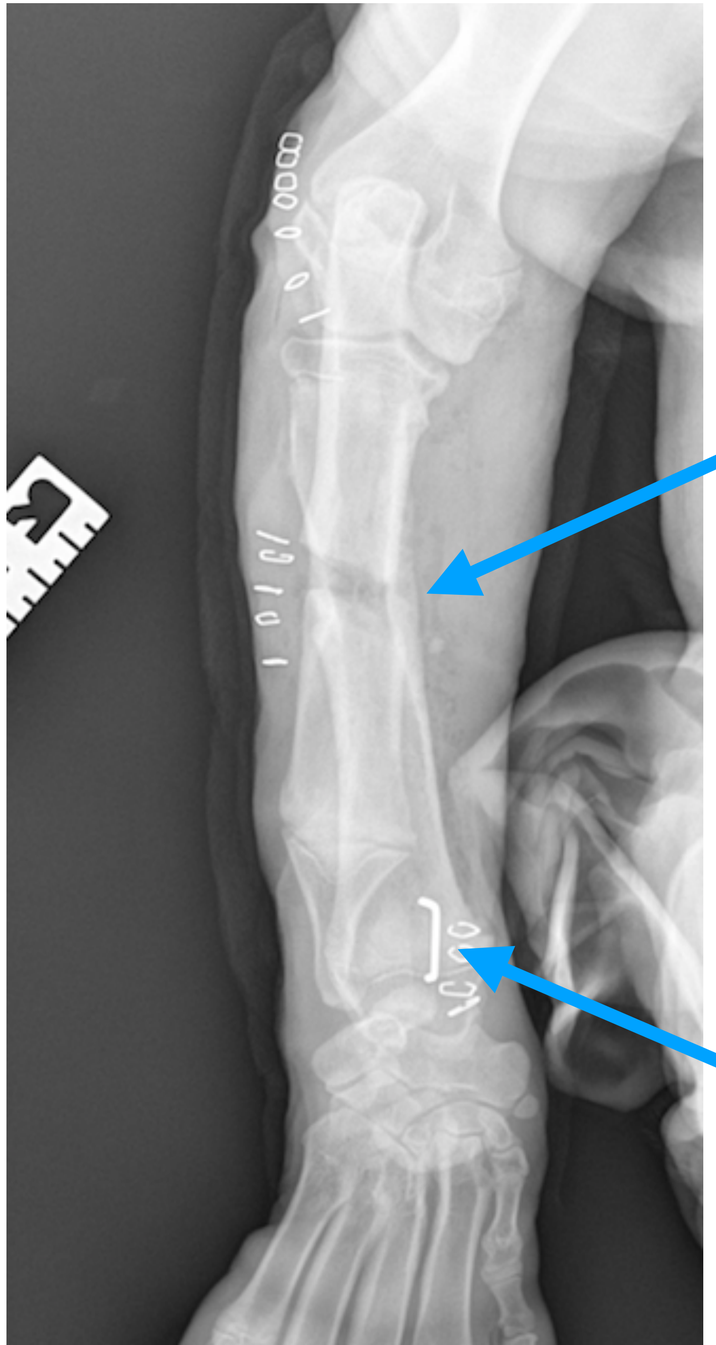
Normal hind legs

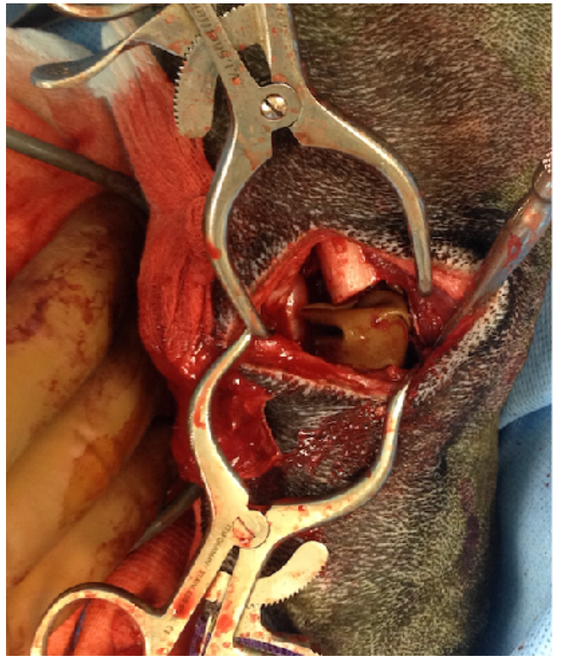
CT

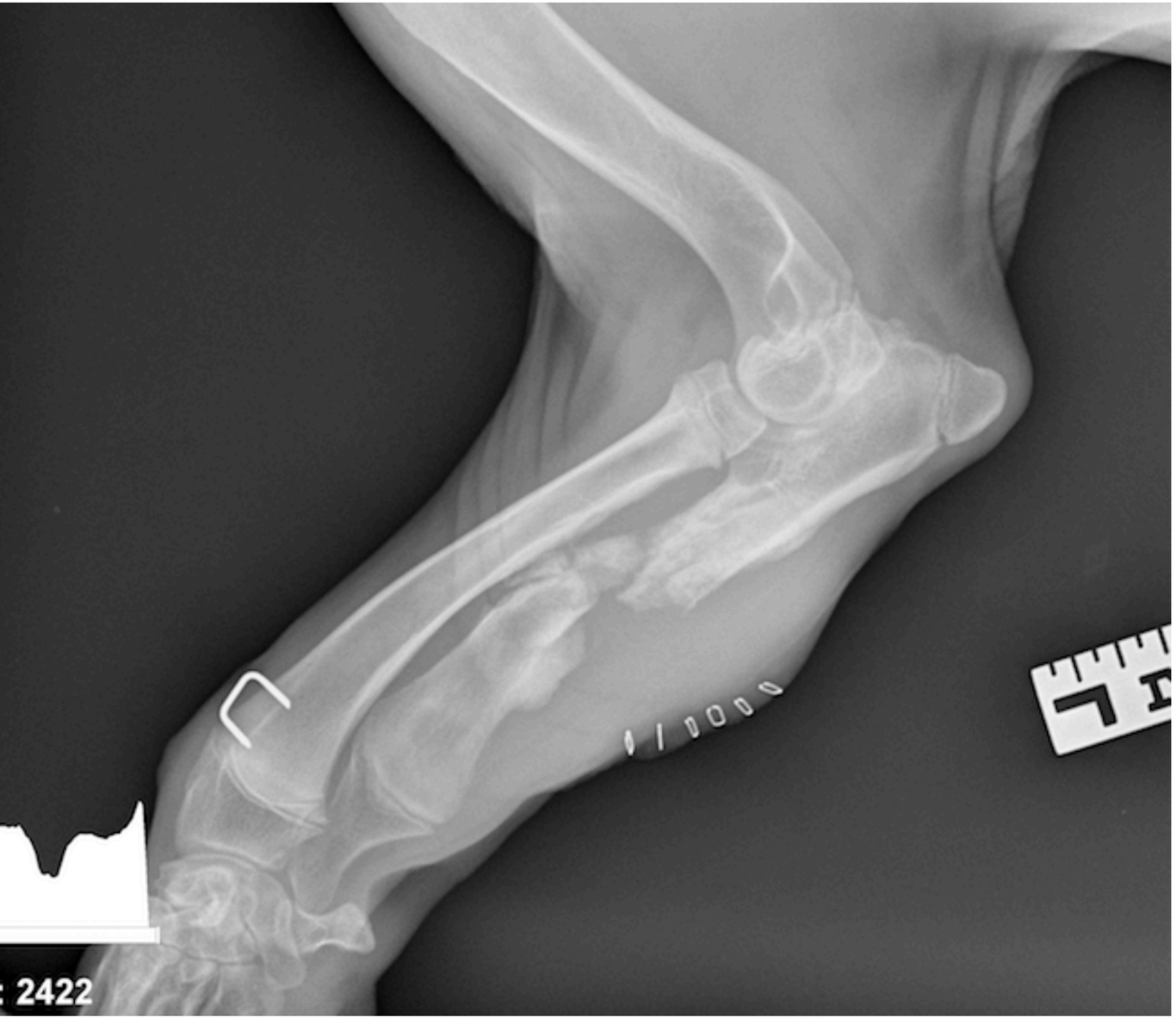


Retained ulna growth
4 months age?


W: 218
I
S



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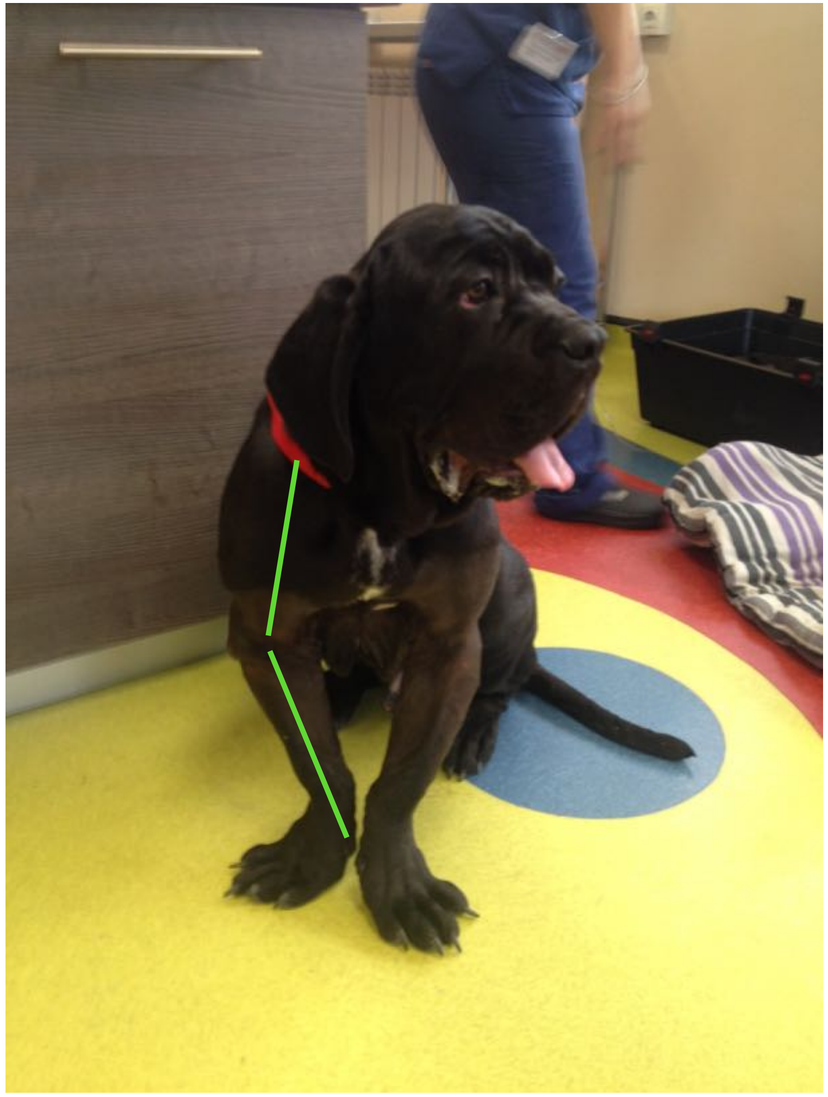
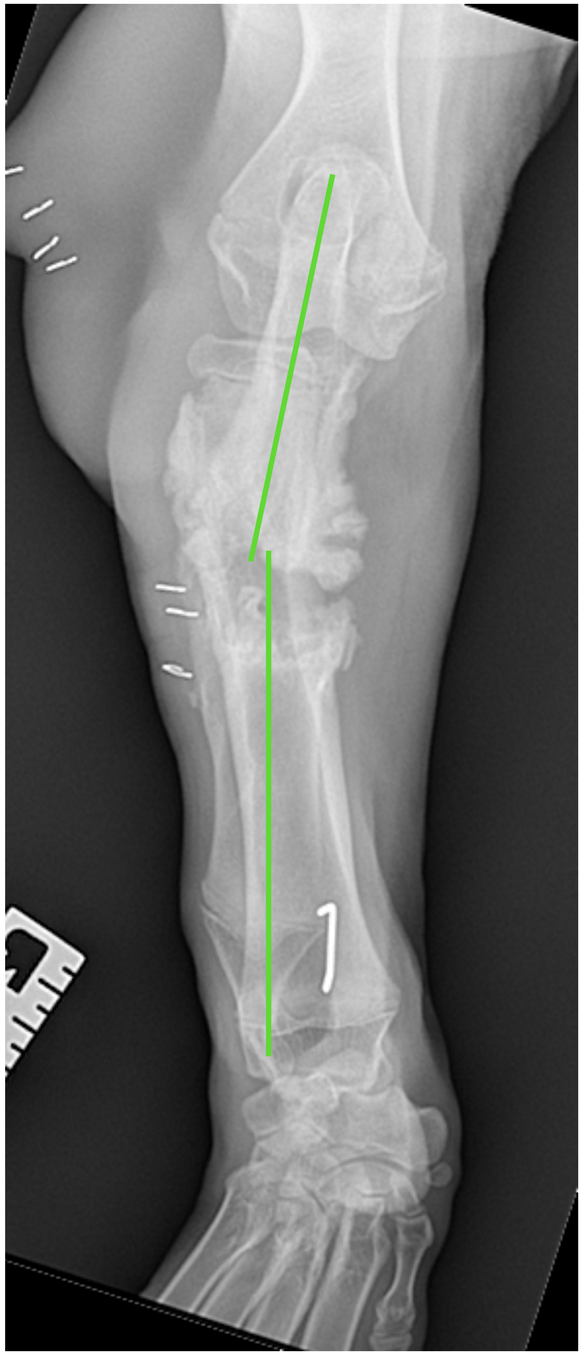
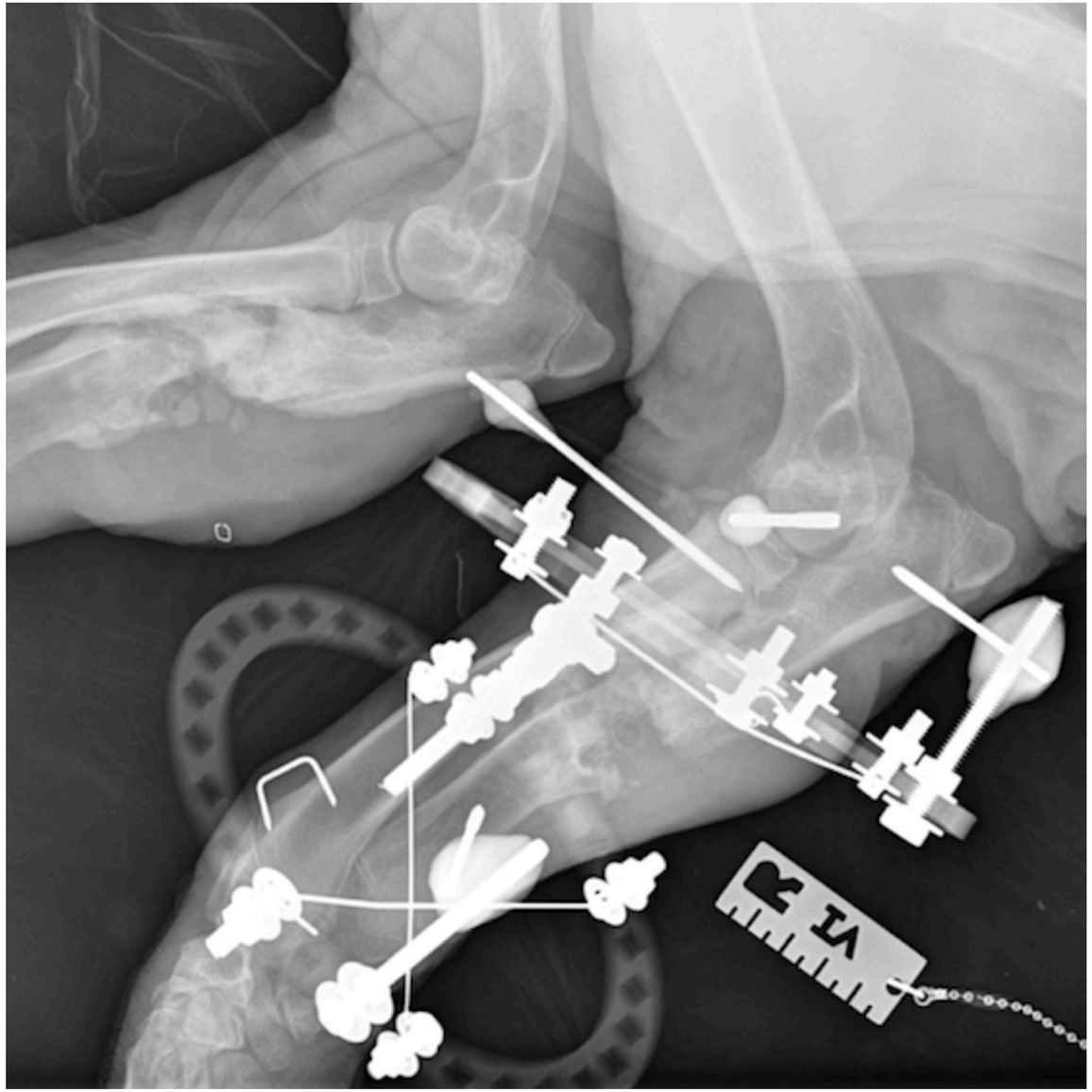
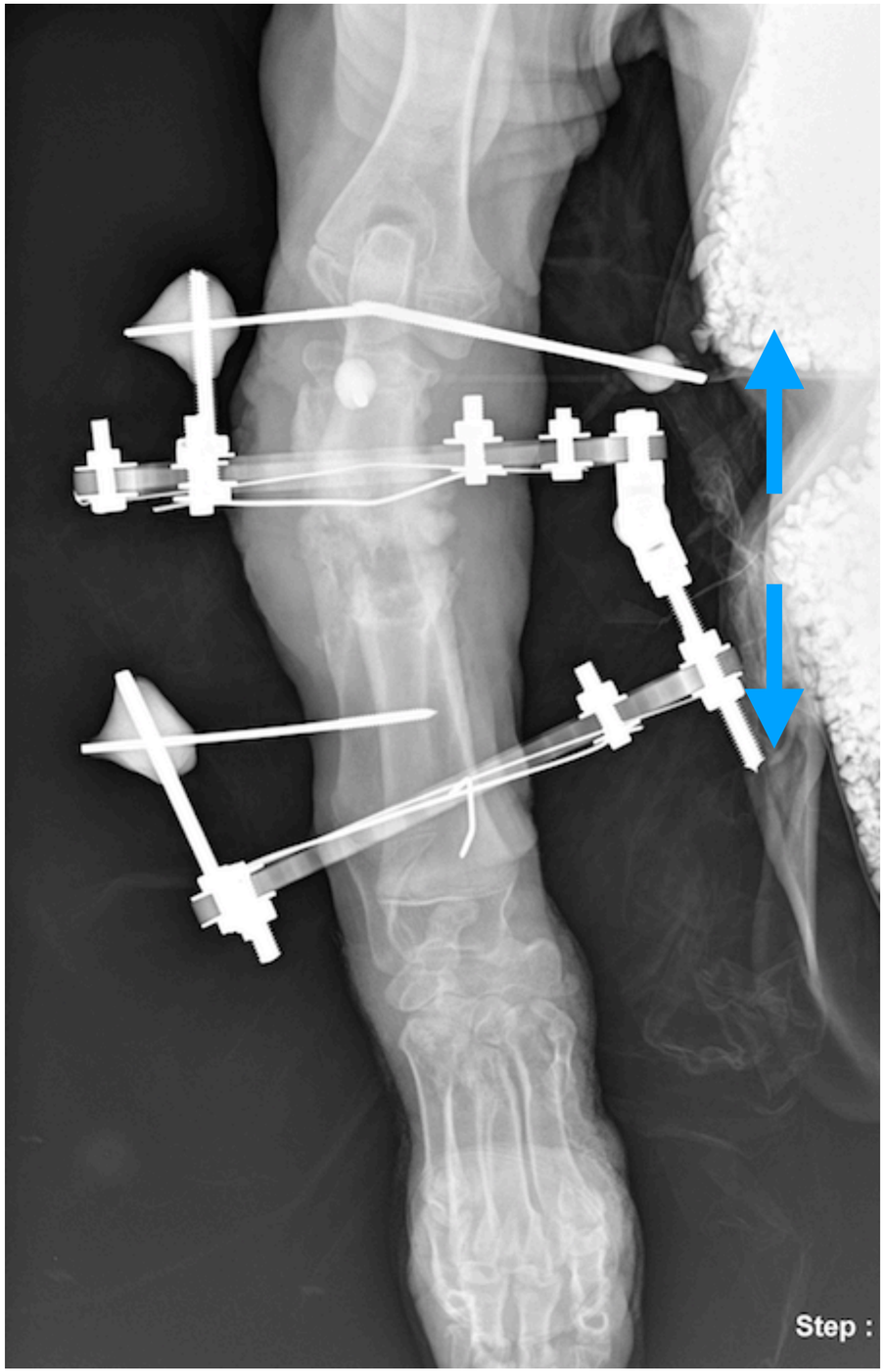
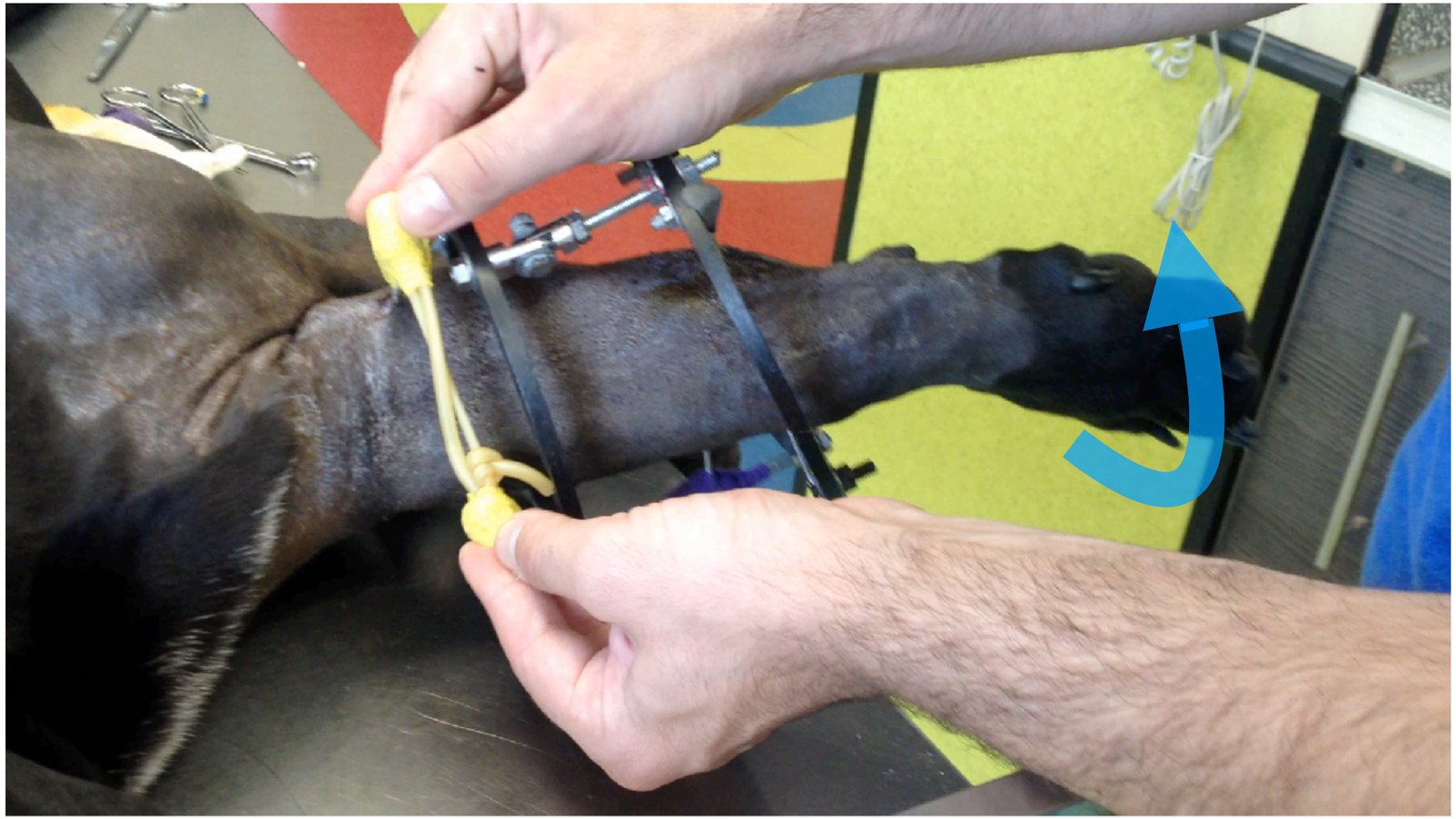
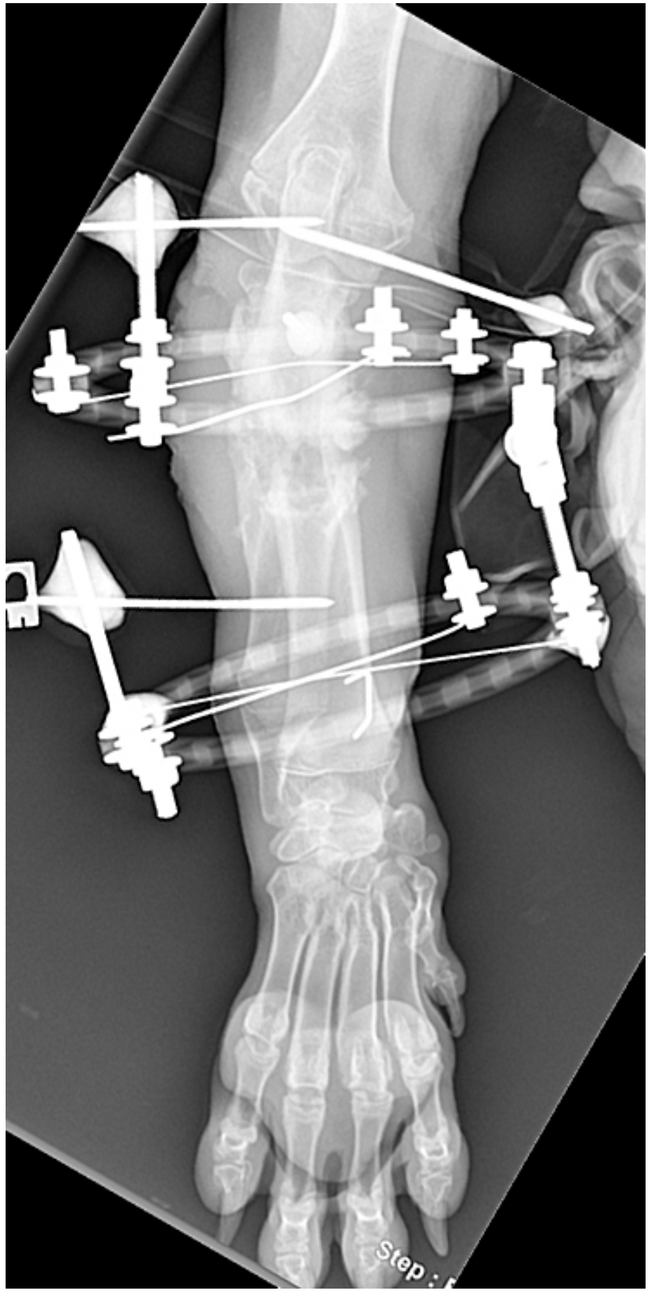


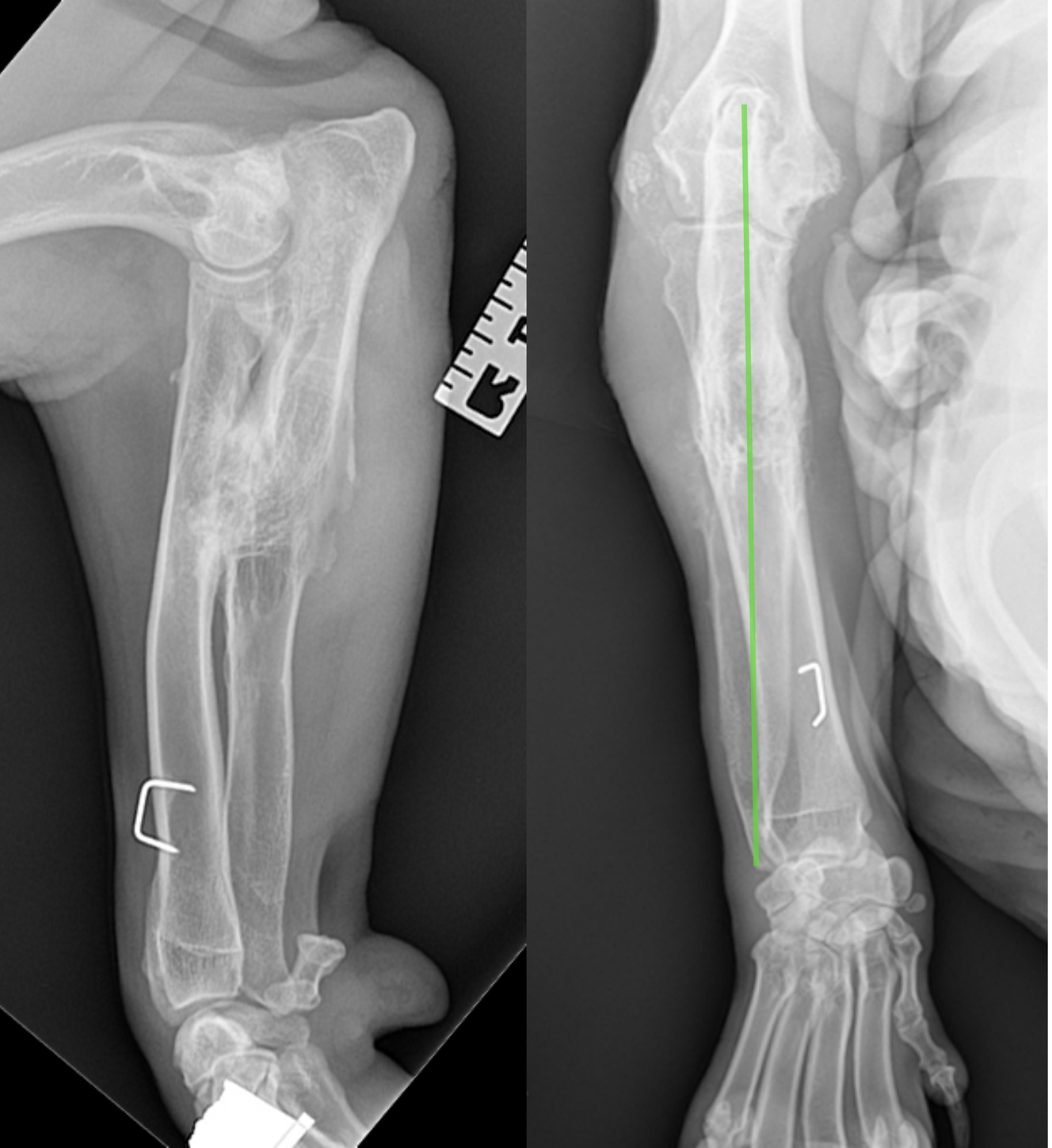





Step :

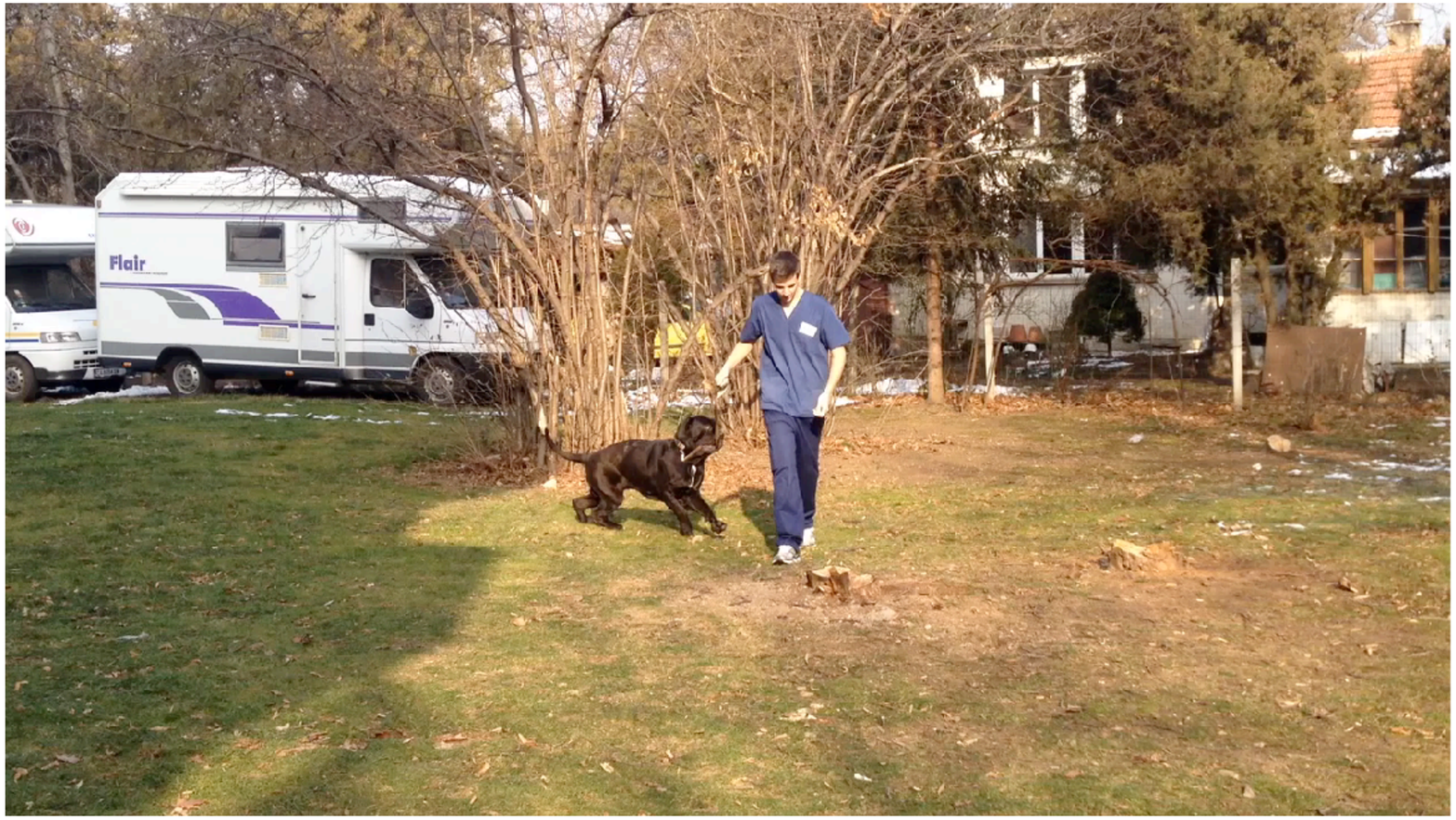









1 year of age,
55 kg



Osteochondrodysplasia in scottish fold

- Generalized defective cartilage formation
- Affects mostly development of the ears, tail and distal limbs
- Autosomal-dominant inherited trait
- Progressive osteoarthritis

Osteochondrodysplasia in scottish fold

- Irregularity- tarsal, carpal, metatarsal and metacarpal bones, phalanges, and caudal vertebrae

- Progressive new bone formation

- Plantar exostosis caudal to the calcaneus in advanced cases

- More spectacular in the hind limbs

Treatment

- Conservative- NSAIDs..
- Surgical approaches- ostectomy and pantarsal arthrodeses
- Palliative irradiation

Mucopolysaccharidosis

- Rare an autosomal recessive disease
- ("lysosomal storage desease" = delayed or missing degradation of glucose-aminoglycans)=> multisystemic accumulation

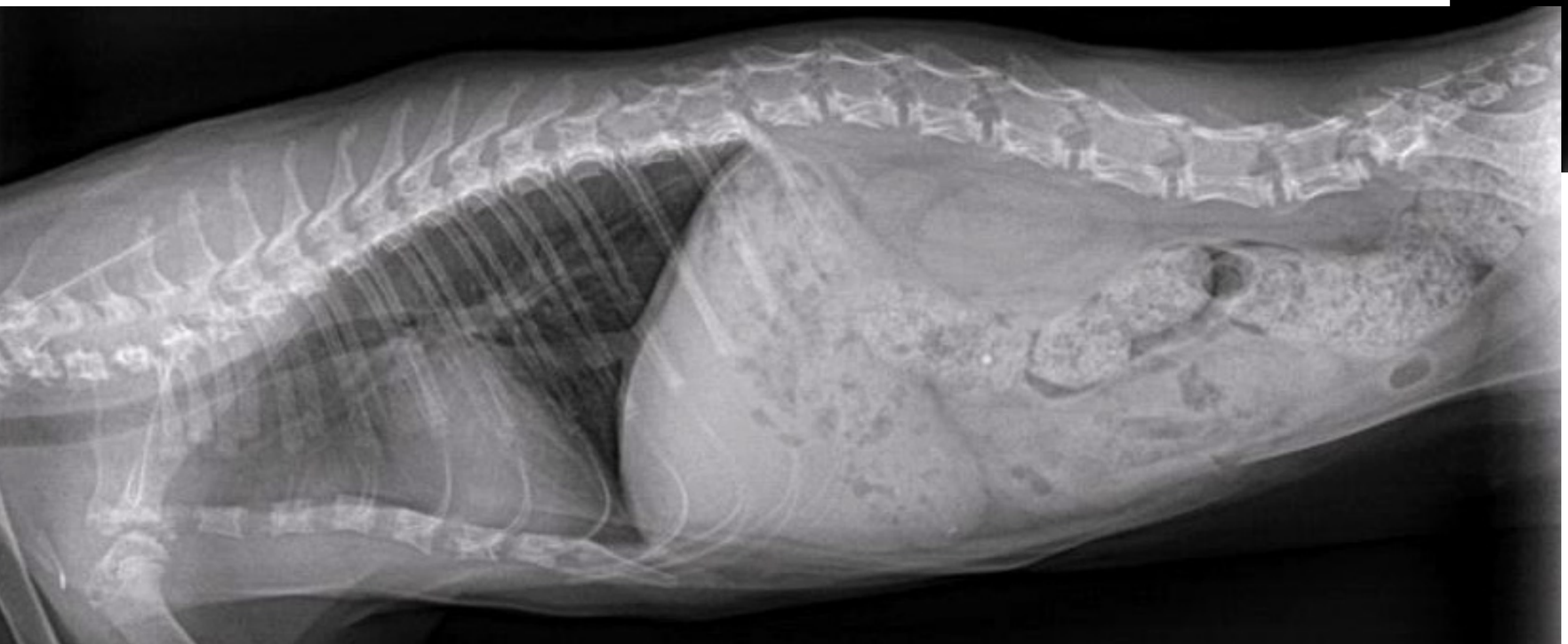
- Several forms- MPS I to VII
- Some breeds over-presented

Common clinical signs

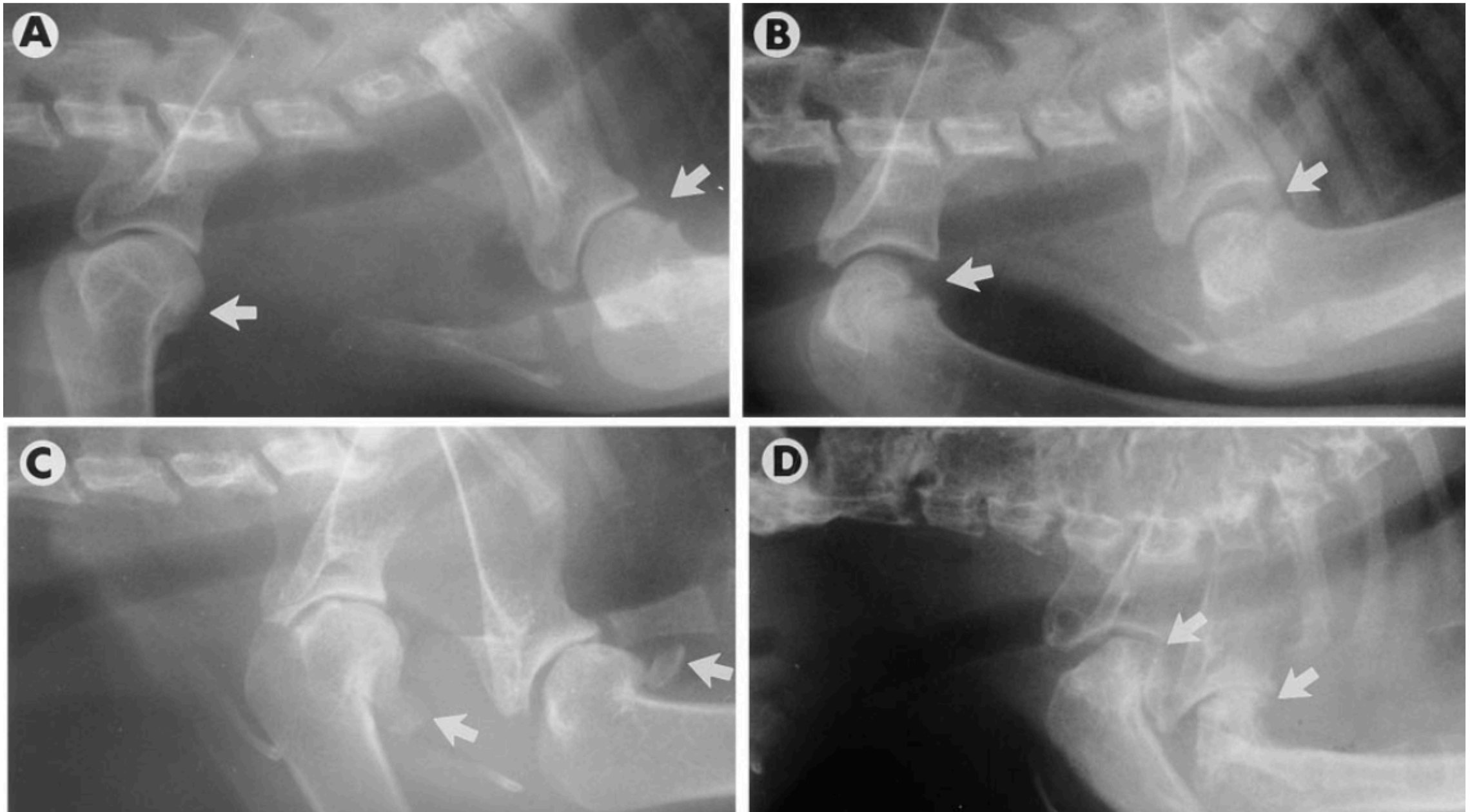
- Facial deformity – flattened face with widely spaced eyes and small ears
- Stunted growth and skeletal abnormalities
- Enlarged liver
- Cloudy discolouration of the corneas
- Mental retardation
- Joints laxity
- Dwarfism

Radiogrpahic signs

- Generlised epyphiseal dysplasia
- Lumbar vertebral deformation
- Joint deformation with bone proliferation.
- Lax hip joints, shallow acetabulums

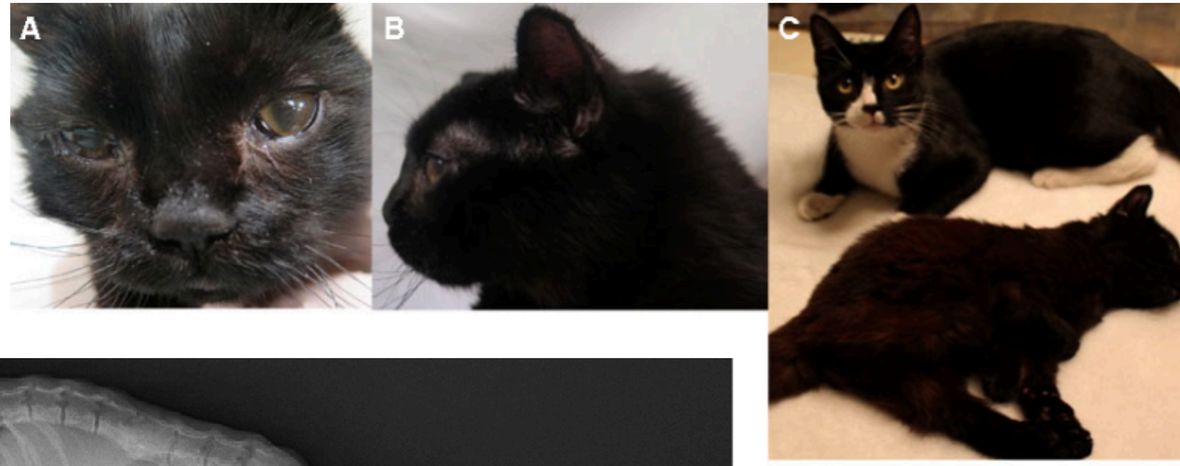


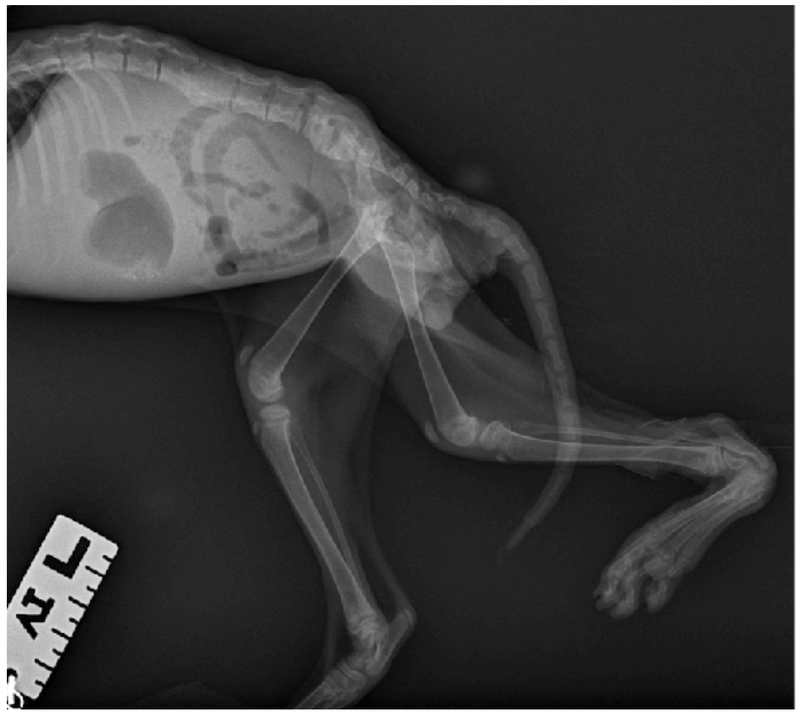
Mucopolysaccharidosis

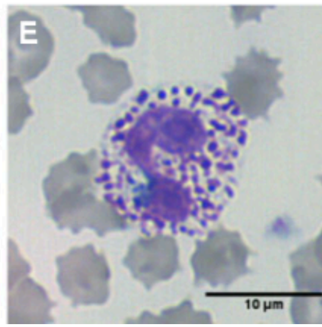


Diagnosis

- Radiographic changes
- Granules and vacuoles in peripheral white blood cells
- Positive toludine blue ‘spot’ test in urine.
- Genetic tests







Treatment

Mol Genet Metab. 2001 Mar;72(3):199-208.

Enzyme replacement therapy in feline mucopolysaccharidosis I.

Kakkis ED[1], Schuchman E, He X, Wan Q, Kania S, Wiemelt S, Hasson CW, O'Malley T, Weil MA, Aguirre GA, Brown DE, Haskins ME.

Enzyme replacement therapy was effective in reversing storage in some tissues at the biochemical and histologic level in MPS I cats…

Radiographic evaluation of bones and joints in mucopolysaccharidosis I and VII dogs after neonatal gene therapy

Ramin Sedaghat Herati [a], Van W. Knox [b], Patricia O'Donnell [c], Marina D'Angelo [d], Mark E. Haskins [c], Katherine P. Ponder [a, e]

"intravenous injection of a retroviral vector (RV) with the appropriate gene resulted in expression in liver and blood cells, and high serum enzyme activity."

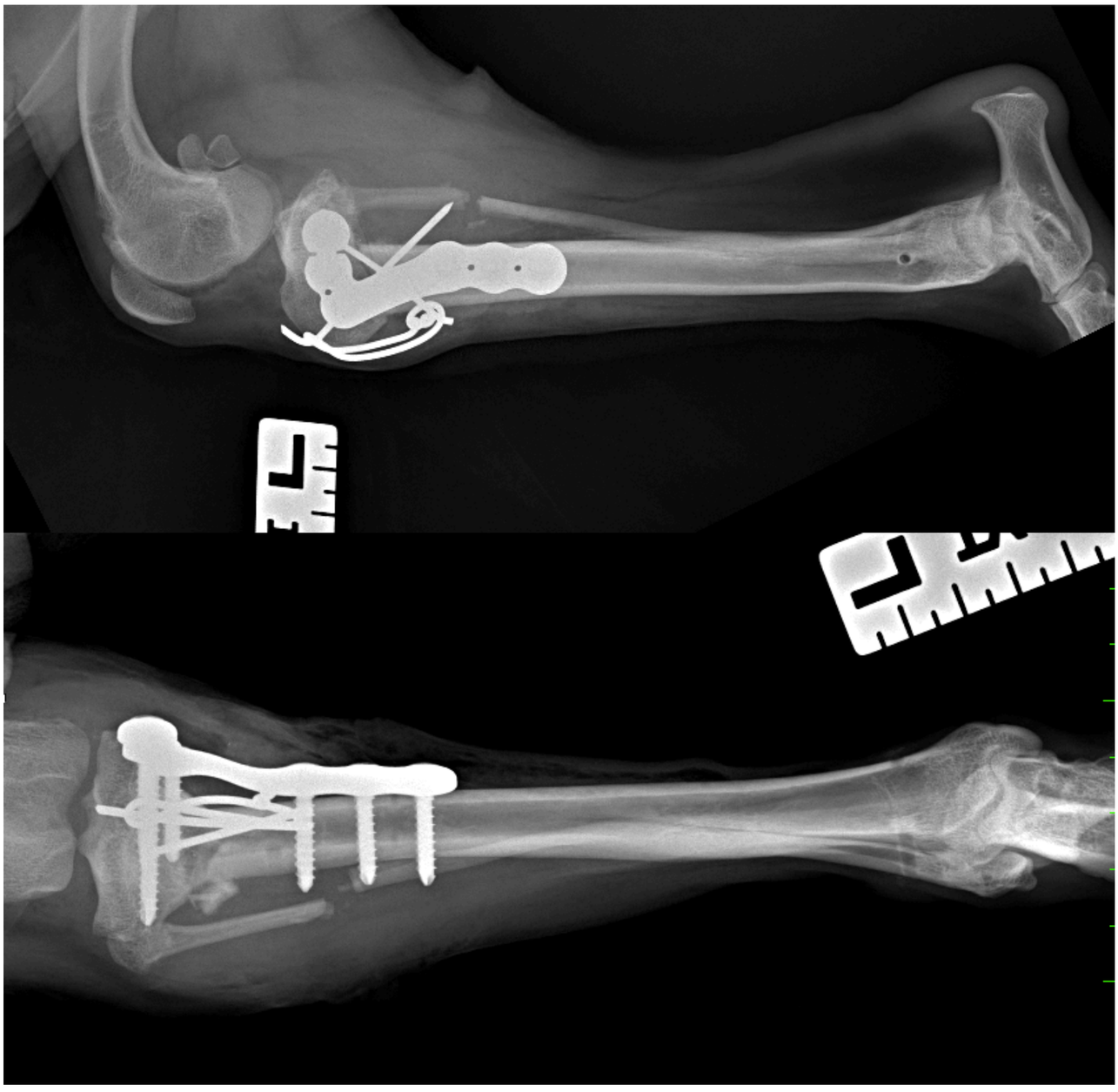
Zahari

- 3 m old puppy, mix, found on the street
- Early signs of limbs deformation
- Corrective surgery attempts

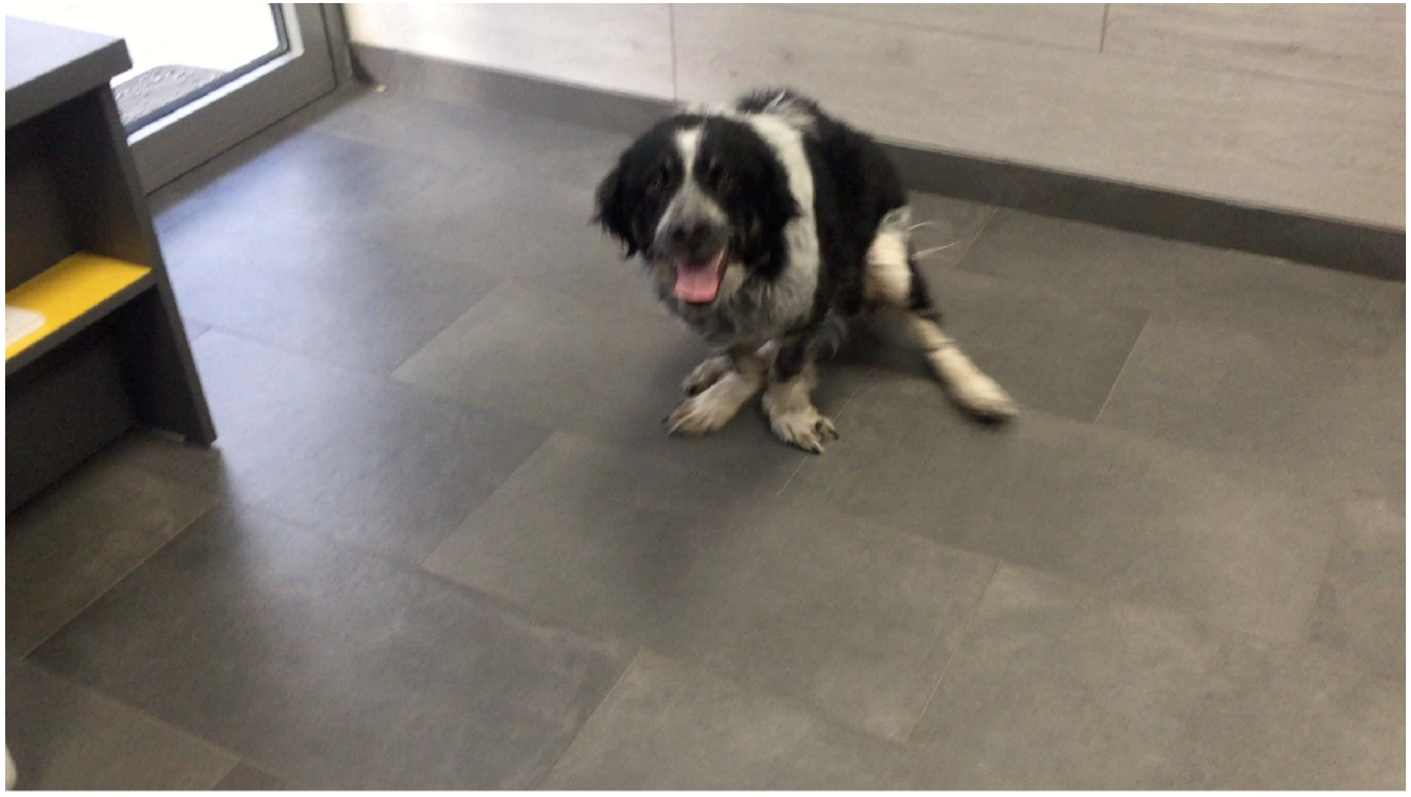
- Four limbs deformation

-tibial valgus
-carpal laxity
-hip laxity

- Short, disproportional face



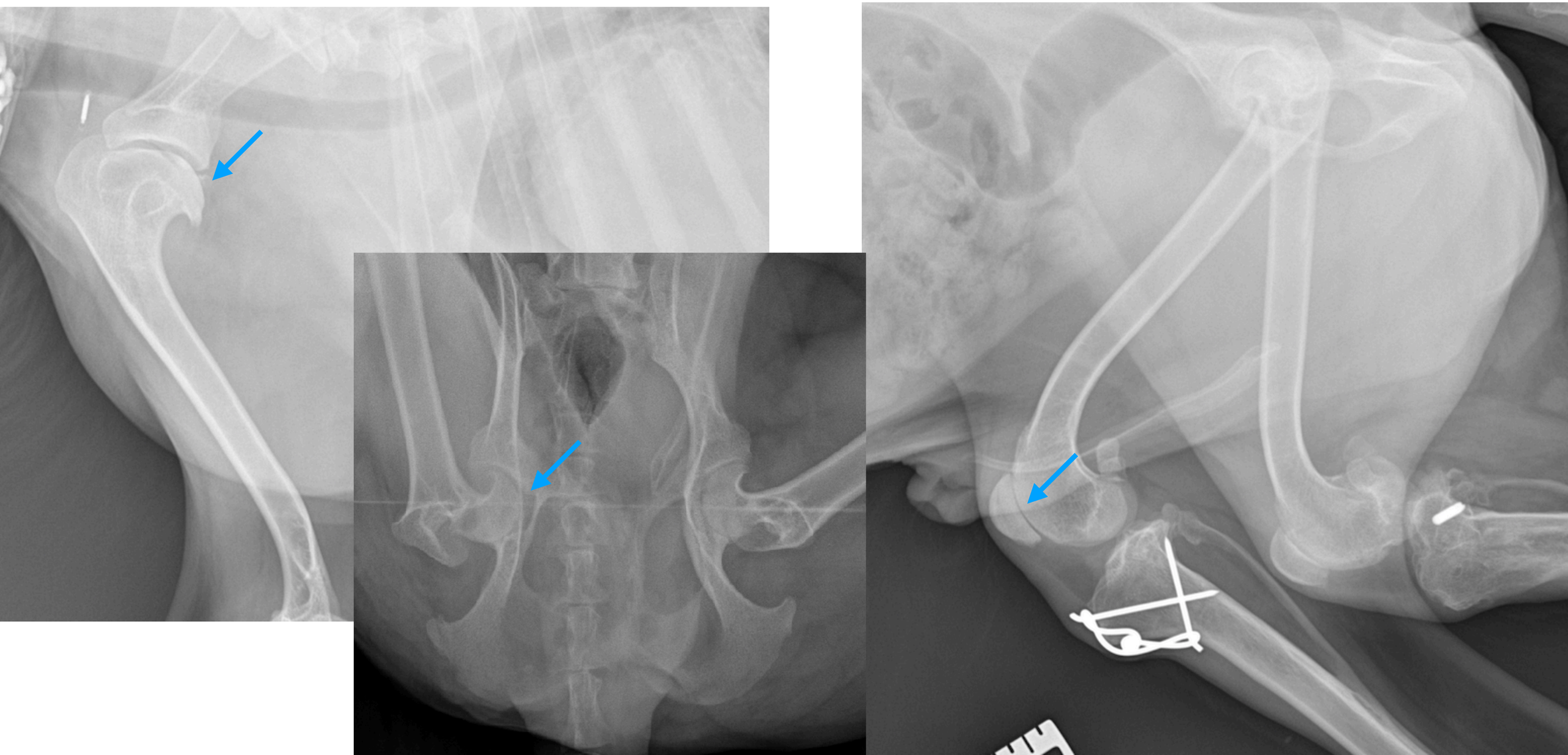

41.75 °
44.85 mm
276.35 mm
19.45 °
9.20 mm




La
Step : Front Ext

6m f up, stiff gait

6m f up- multiple joint affected by DJD- cartilage thinning

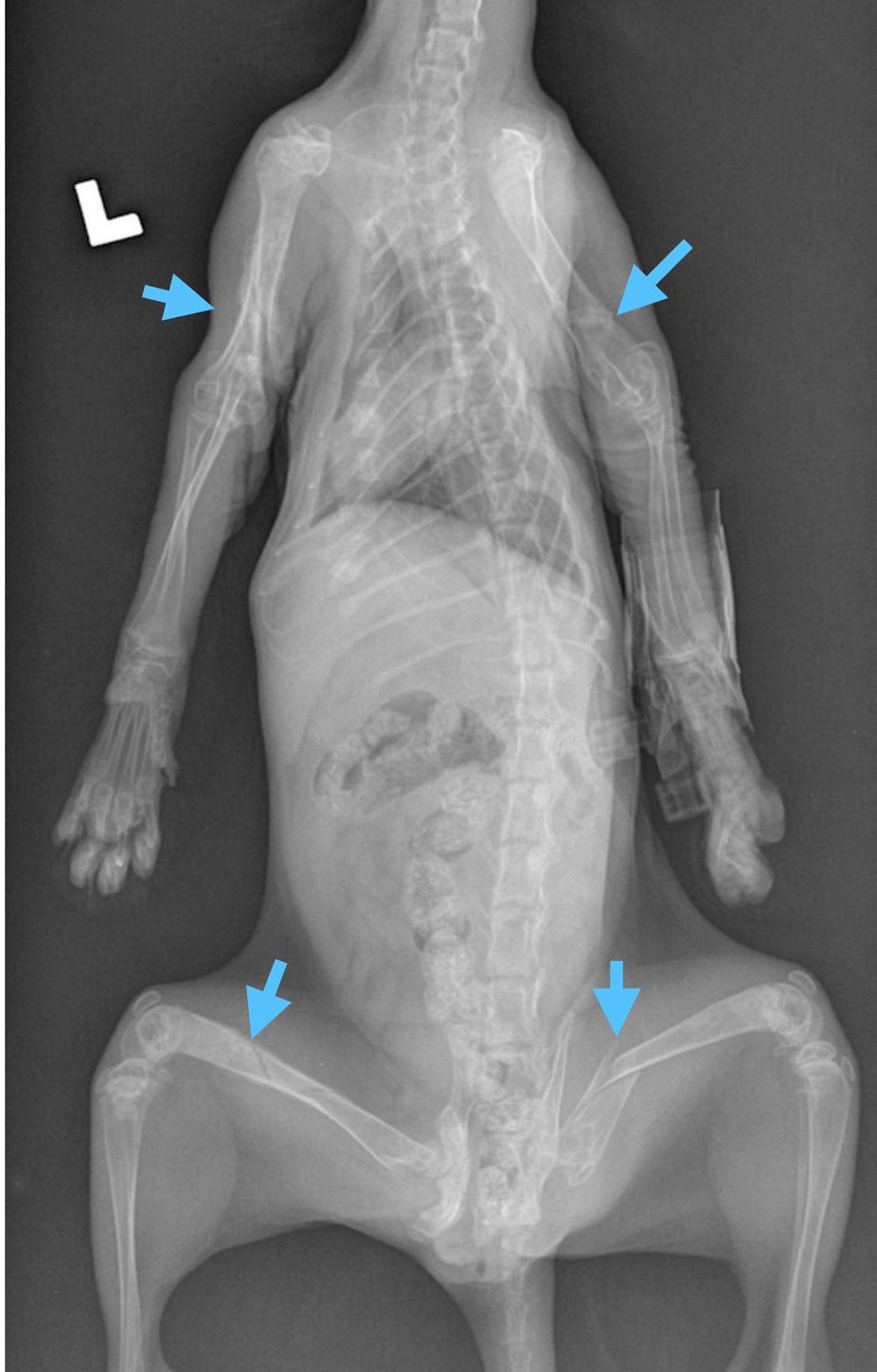


Osteogenesis imperfecta

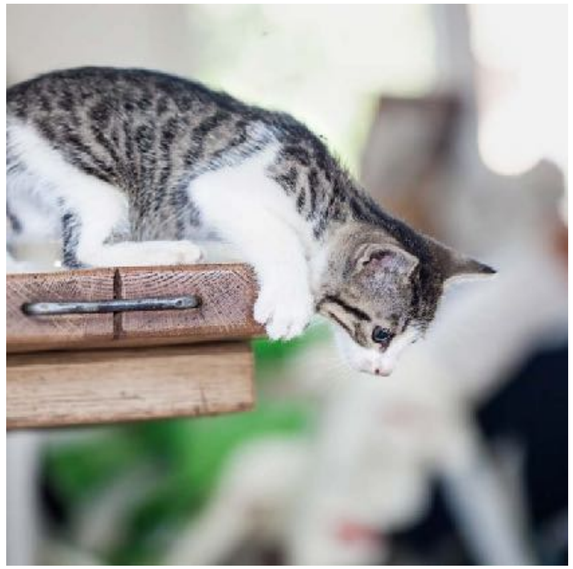
- Mutation of a gene coding for **type I collagen**.
- Fragile bones=> pathologic fractures with minimal trauma.
- Dogs- Golden retriever, Collie, Puddle, Beagle.
- Infrequently in cats.
- Usually-10 and 18 weeks of age.

Diagnosis

- History of multiple fractures with minimal trauma
- “Folding fractures”
- Old healed fractures.
- Thin long bone cortices
- Biopsy
- Teeth fractures, discoloration



Prognosis and treatment



- **<u>Trauma prevention</u>**

- Vitamin C
- Bisphosphonate therapy-**alendronate** at 3 mg/ kg/12 h p.o.

- The prognosis- guarded to poor
- Owners compliance with recurrent fractures

Steven

6 m old , ESH, male

Multiple fracture history


60 d
R


75 d

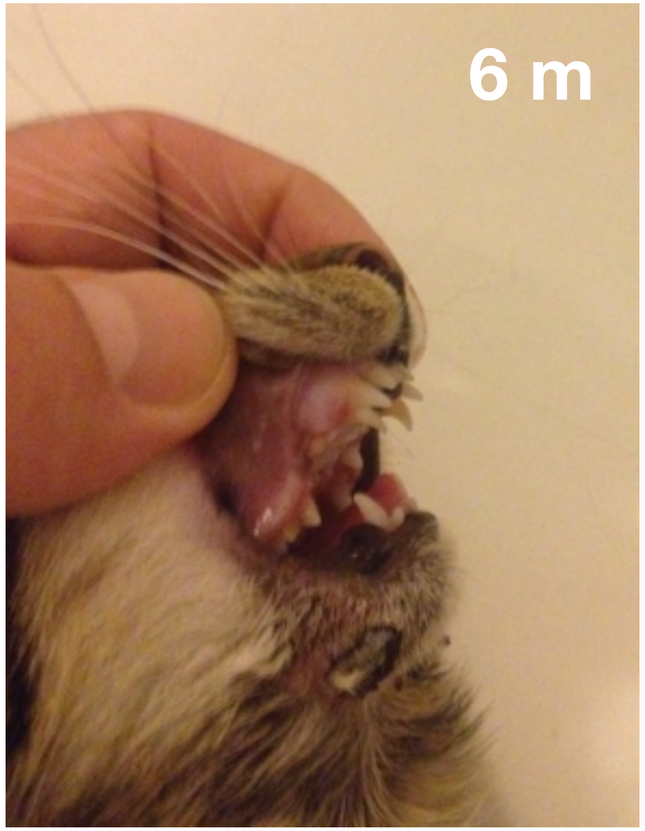

85 d


75 d


90 d

Radius refracture

Spontaneos femoral fracture



Telscoping rodding techniques in children with OI

J Med Life. 2015 Oct-Dec; 8(4): 544–547.

PMCID: PMC4656969

PMID: 26664487

Early telescopic rod osteosynthesis for Osteogenesis Imperfecta patients

A Sterian,[*,**] R Balanescu,[*,**] A Barbilian,[**,***] I Tevanov,[*] M Carp,[*] C Nahoi,[*] M Barbu,[*] and A Ulici[*,**]

J Child Orthop. 2018 Feb 1; 12(1): 97–103.

doi: 10.1302/1863-2548.12.170009

PMCID: PMC5813132

PMID: 29456761

Effects of a telescopic intramedullary rod for treating patients with osteogenesis imperfecta of the femur

D. L. Rosemberg,[a,1] E. O. Goiano,[2] M. Akkari,[2] and C. Santili[2]

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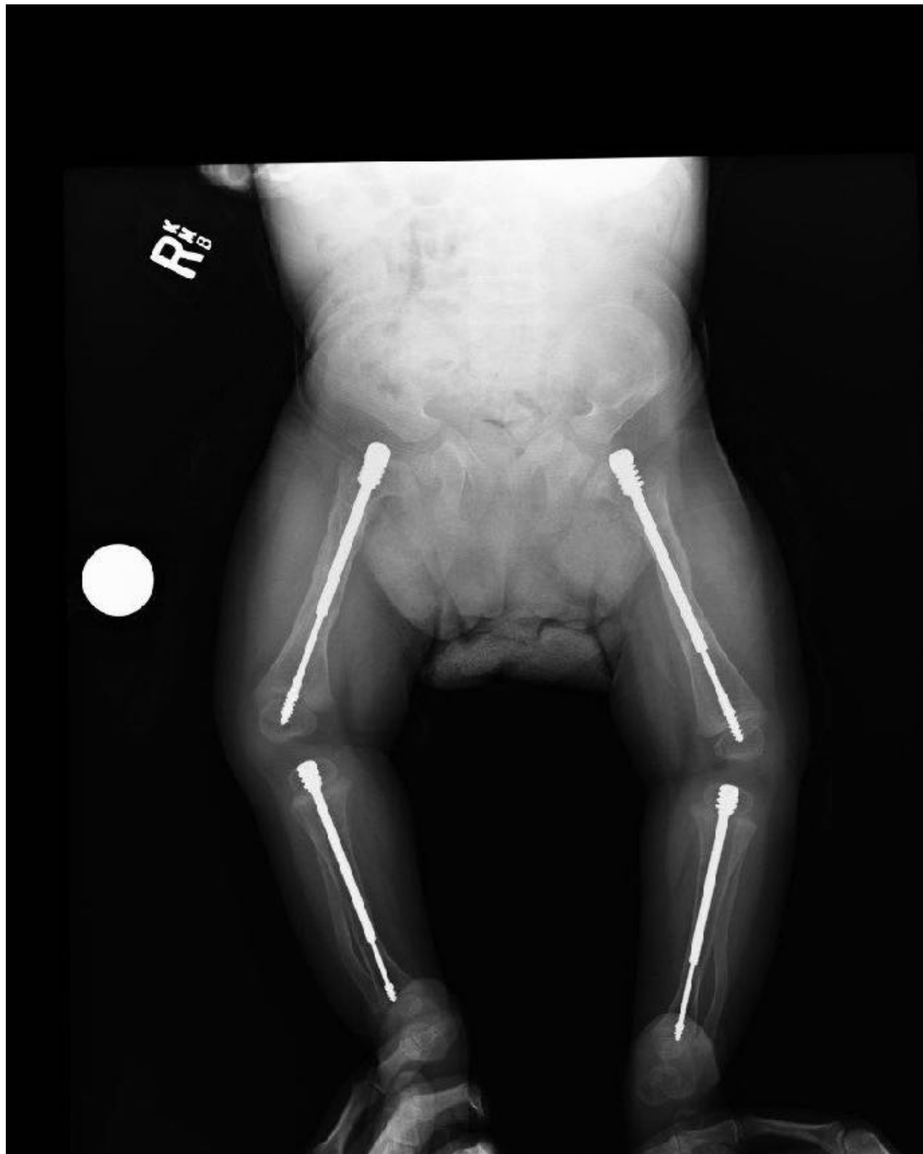

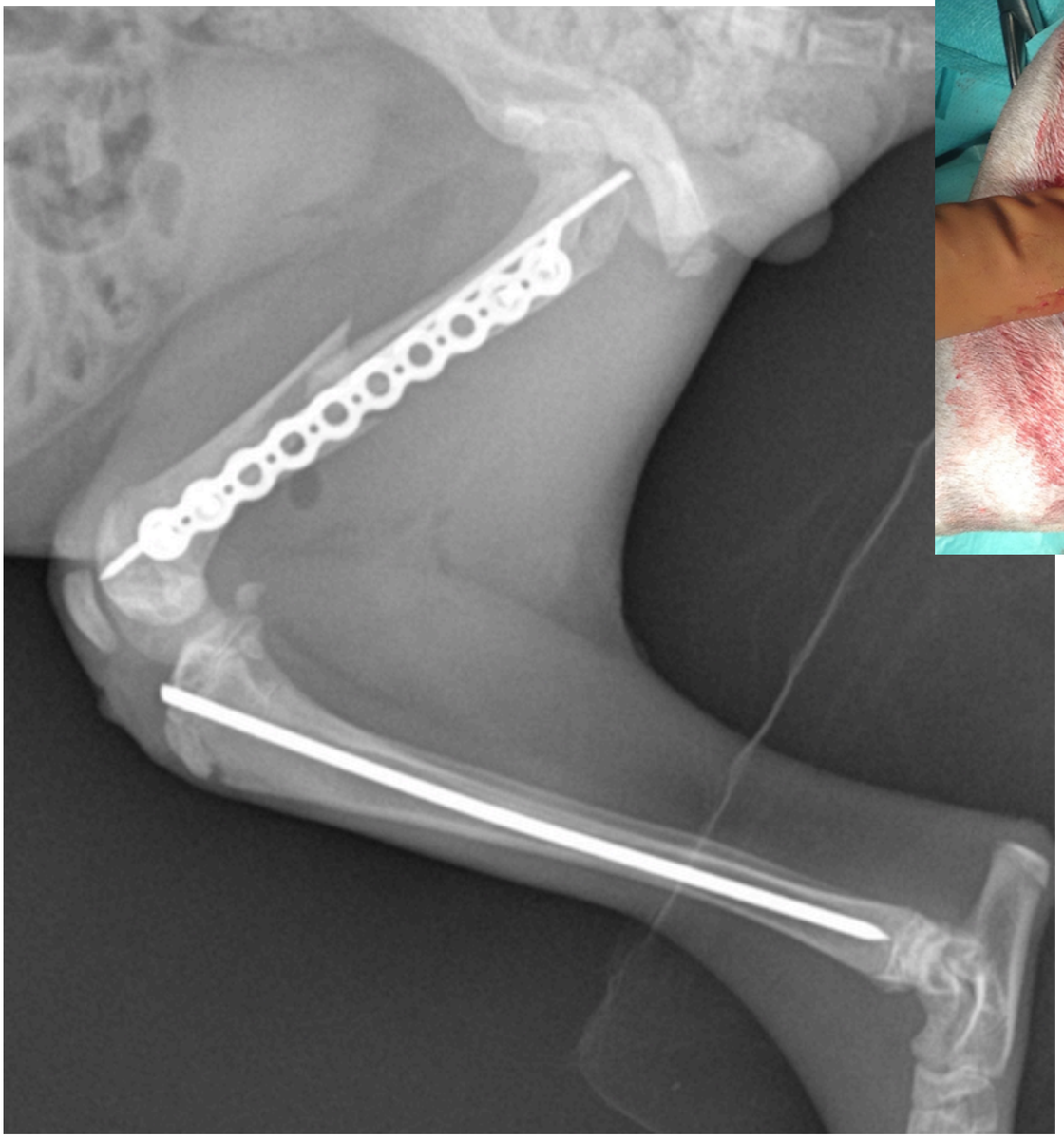

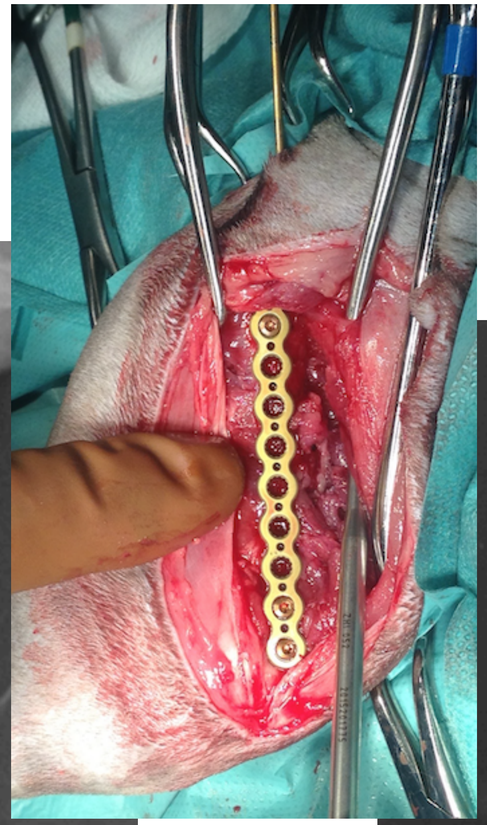

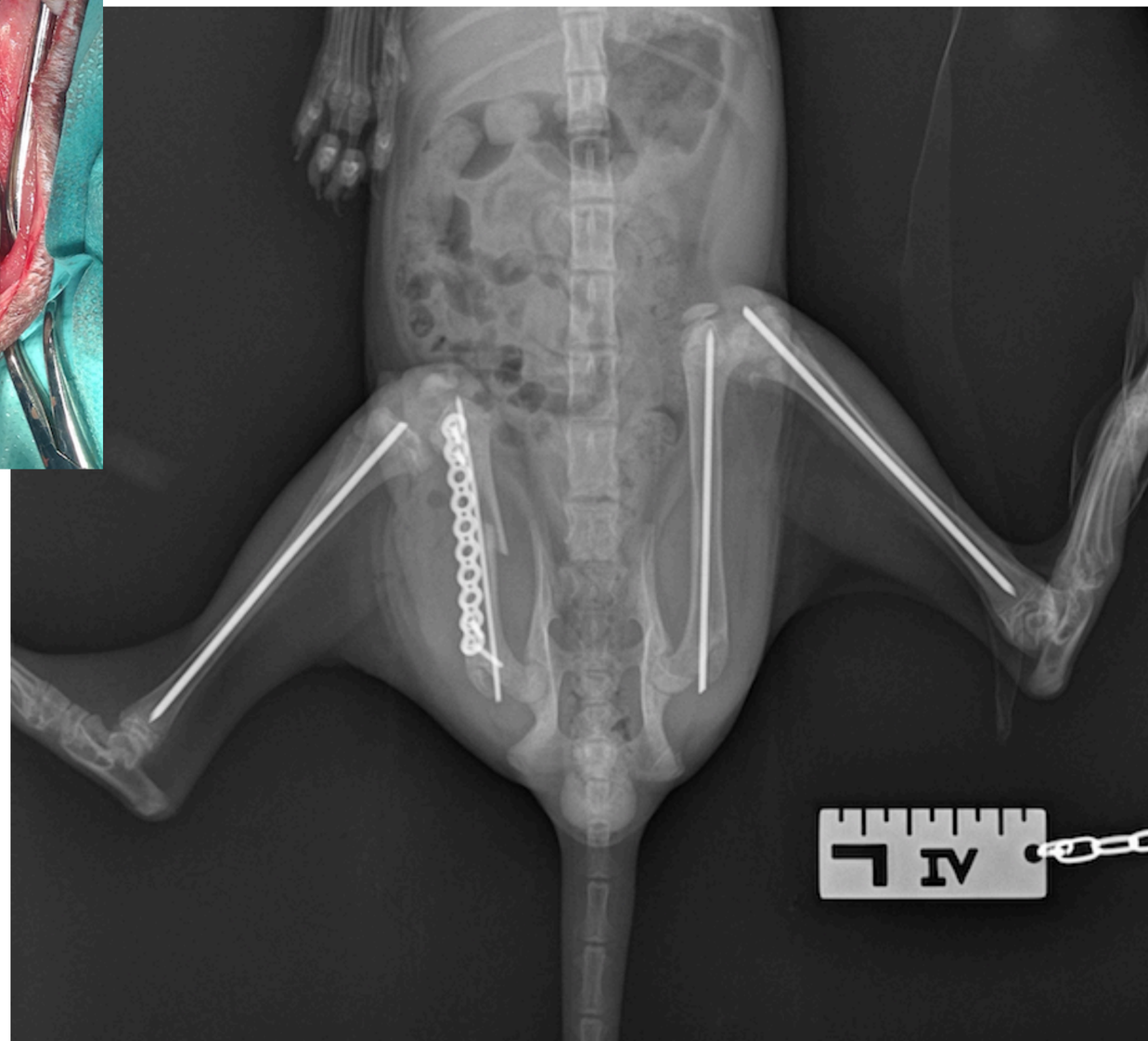

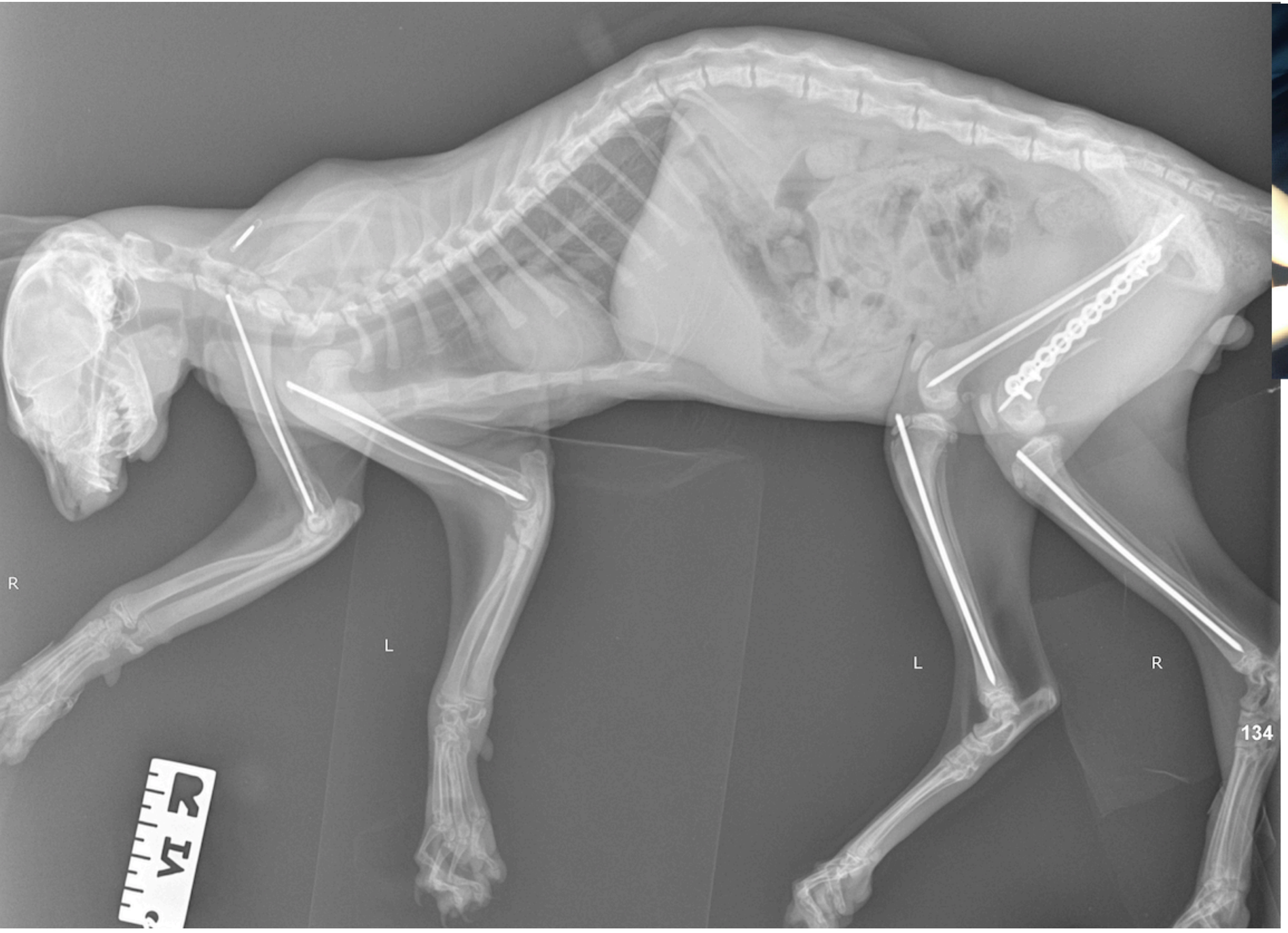
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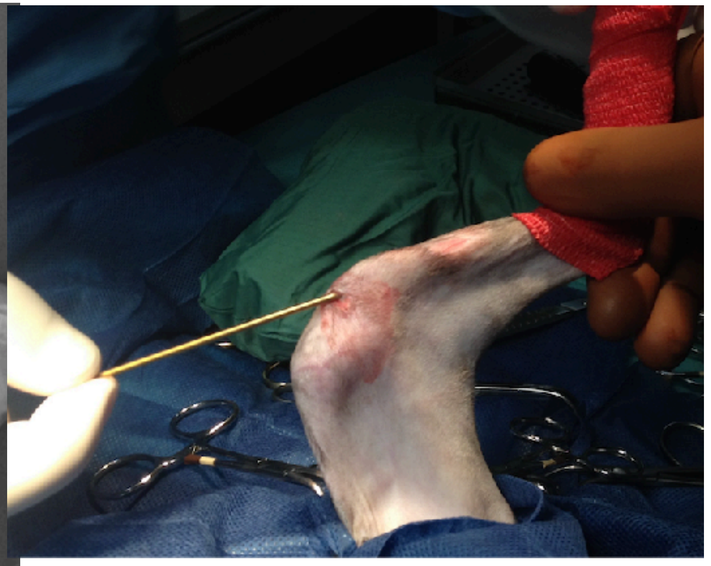
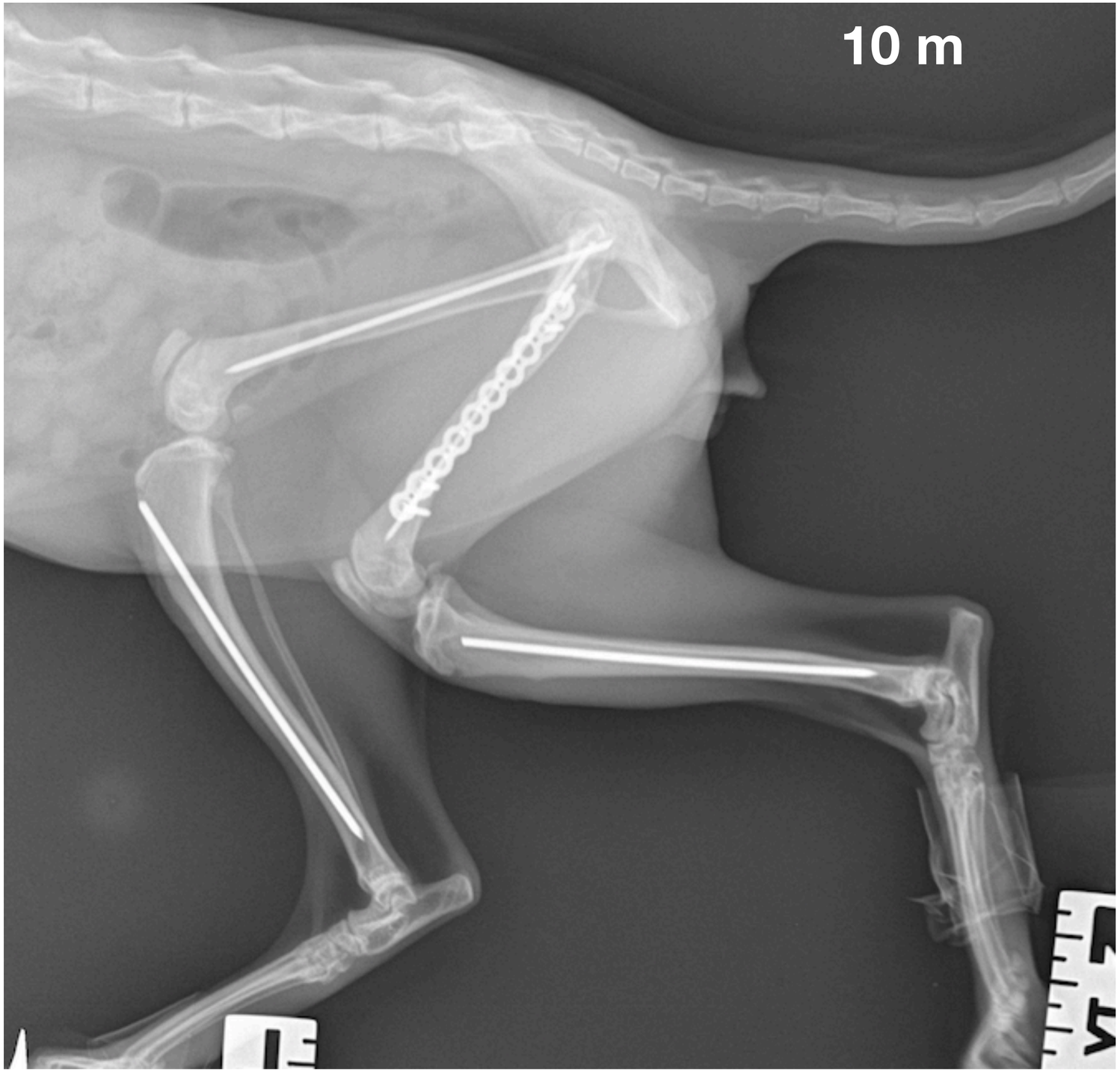
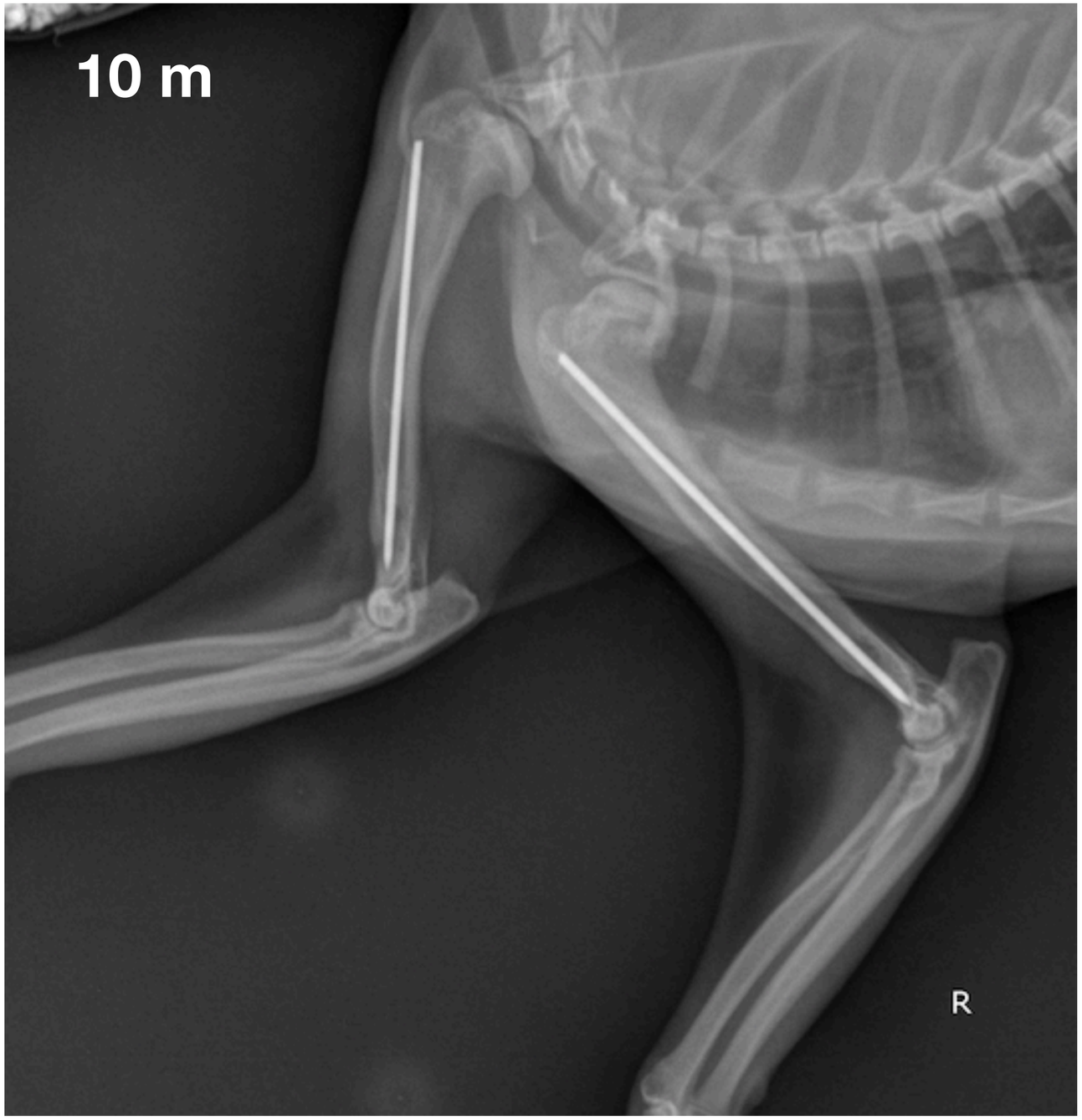
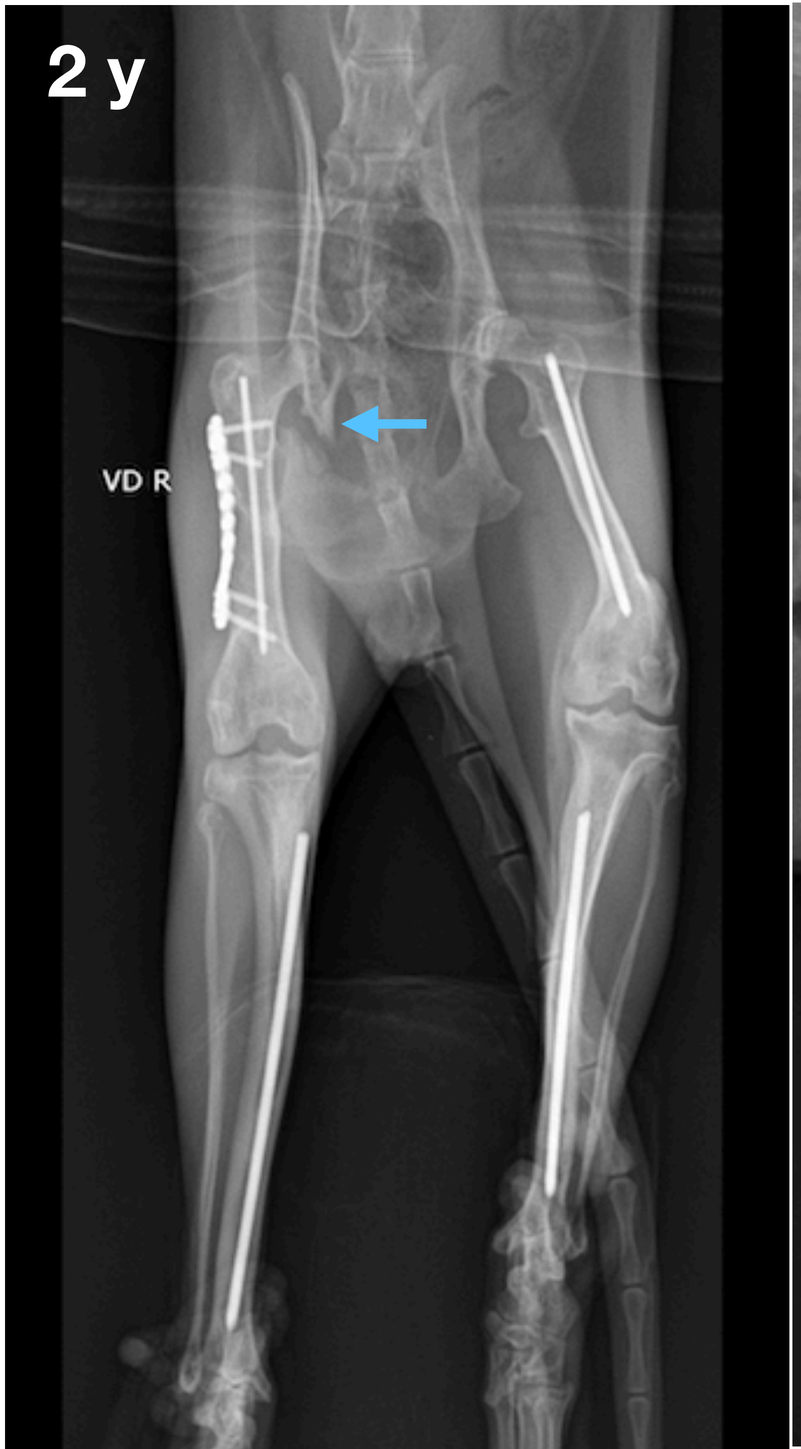
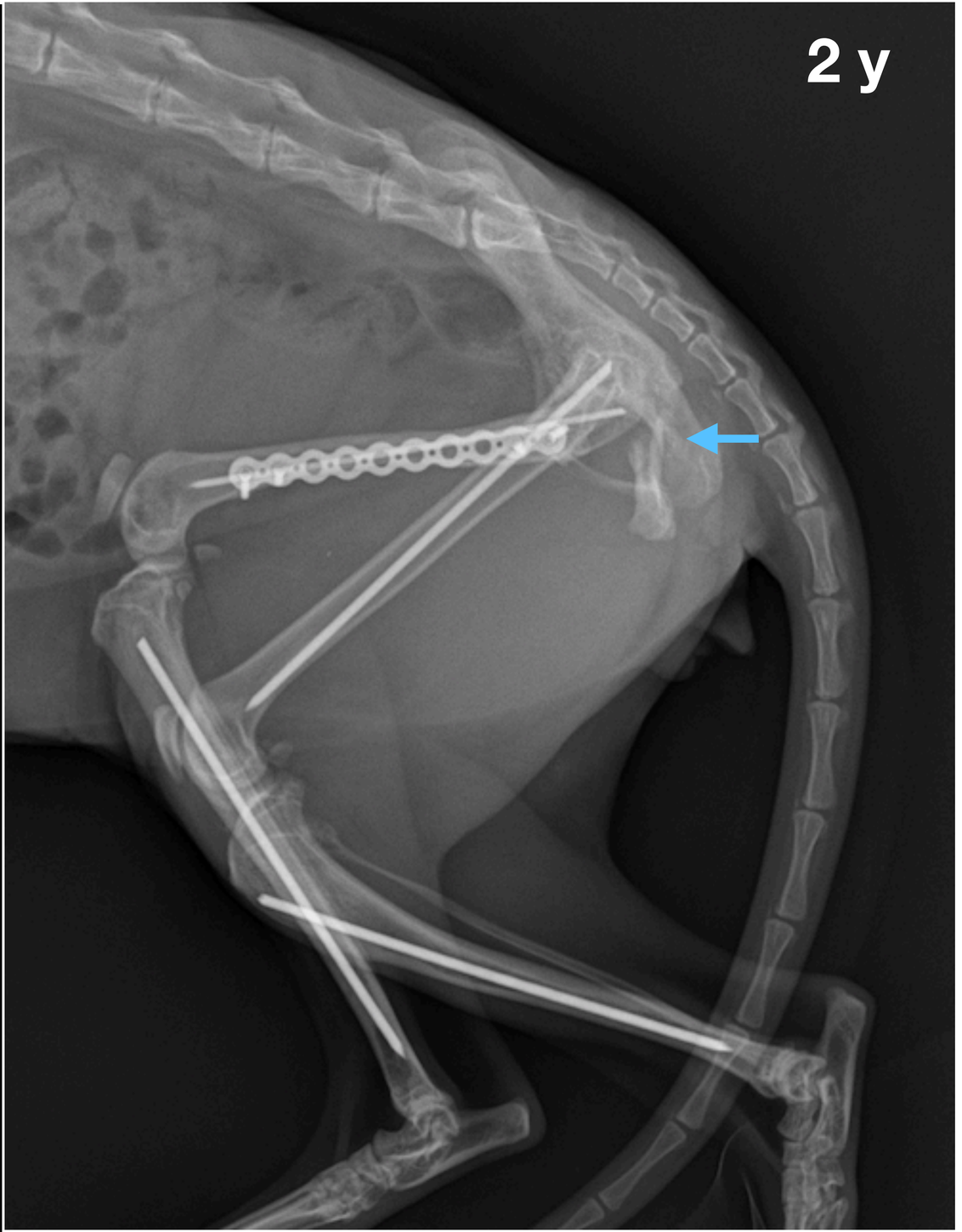


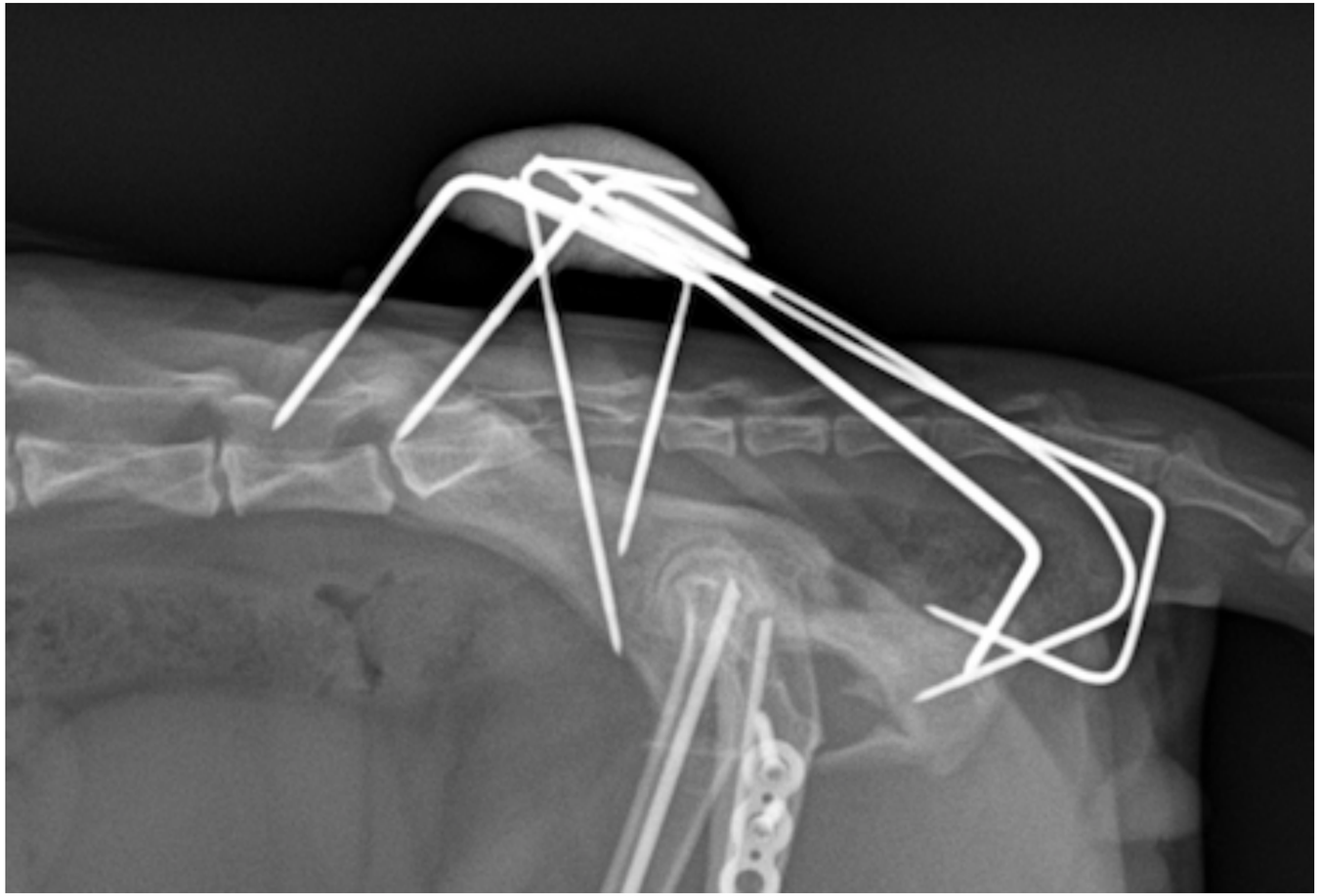

10 m

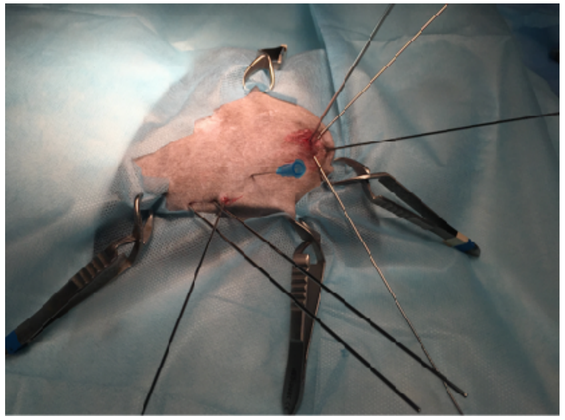

10 m
R


VD R

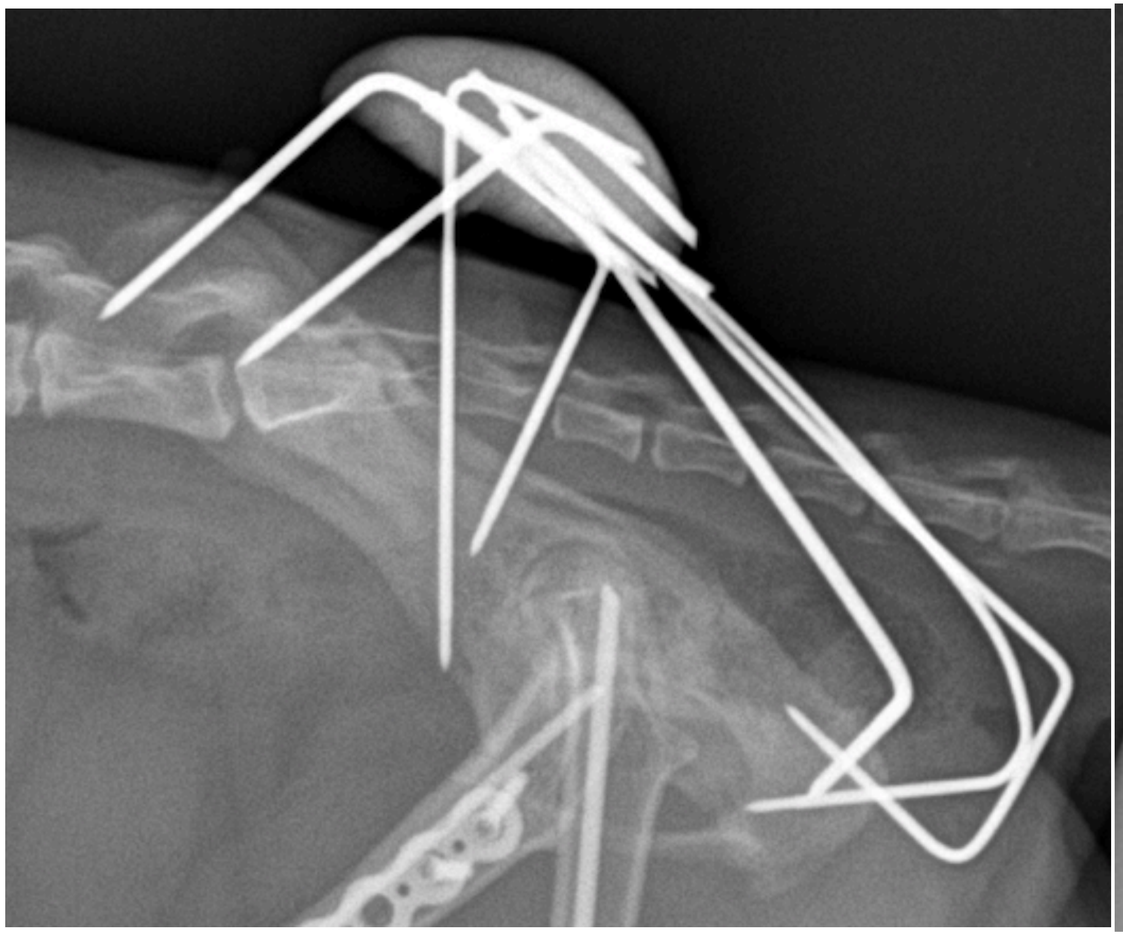

2 y
VD R


2 y





30 day post op



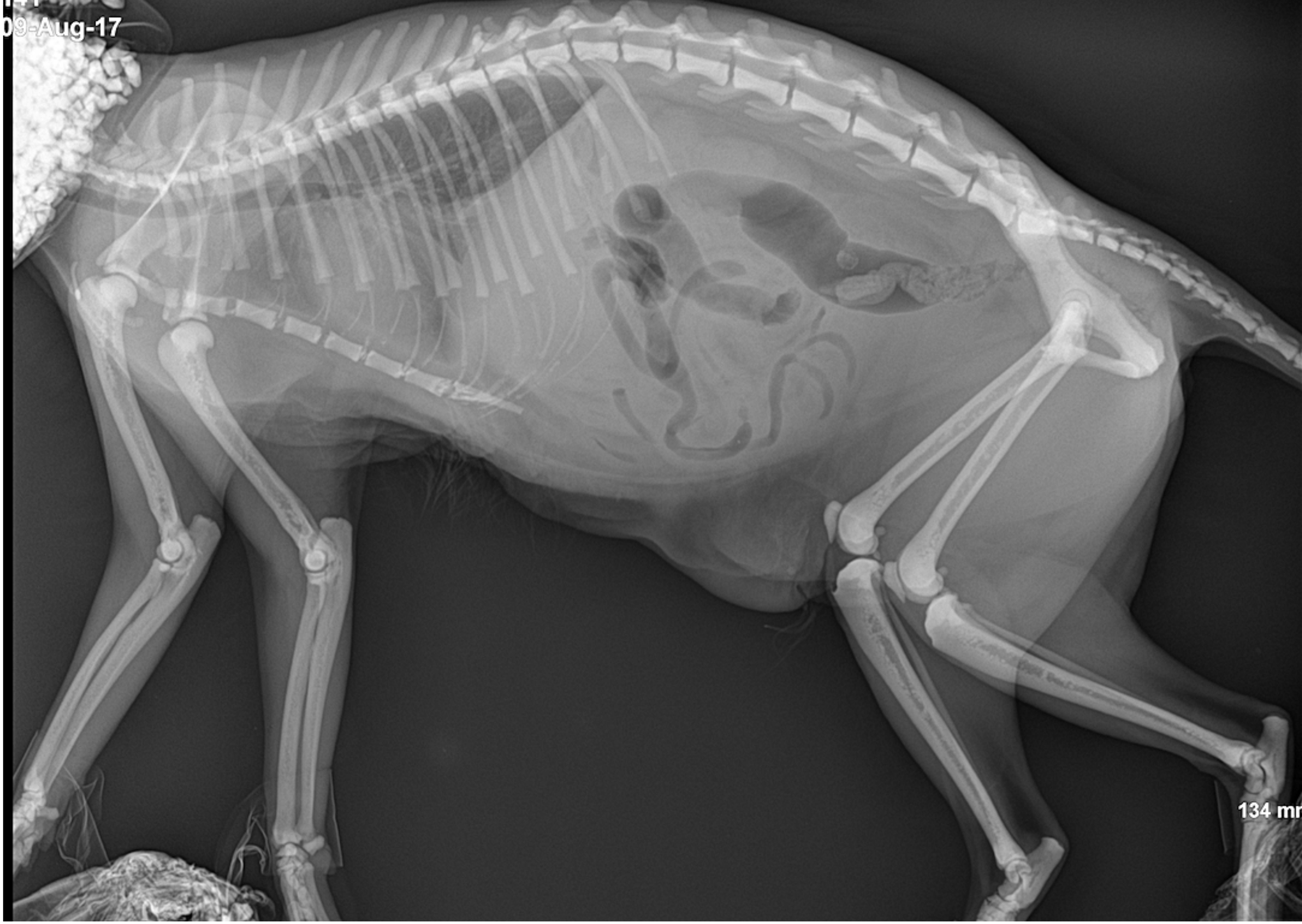
Osteopetrosis

- Extremely rare
- Malfunctioning osteoclasts, inability to resorb bone
- The exact molecular defect unknown.
- Bone marrow cavities, replaced by primary spongiosa.
- Increased bone fragility.
- Myeloplastic anemia

Diagnosis

- Radiographic- “marble view”.
- Australian Shepherds, Pekingese and Dachshunds.
- Only few cases reported in cats

14 y old ESH, cardiac pathology



Congenital tarsal hyperextension/ Tarsal arthrogryphosis

- Recent, uknown ethilogy
- Very rare
- Usually bilateral
- Only cats?

Treatment

J Am Vet Med Assoc. 2006 Apr 15;228(8):1200-3.

Congenital tarsal hyperextension in three cats.

Buote NJ[1], Reese CJ.

“Conservative management involving external coaptation and physical therapy led to favorable results in all 3 cats.”

Treatment

JSAP JOURNAL OF SMALL ANIMAL PRACTICE
BSAVA BRITISH SMALL ANIMAL VETERINARY ASSOCIATION

Partial tarsal arthrodesis and botulinum toxin A injection for correction of tarsal arthrogryposis in a cat

S. R. Bright, S. L. Girling, T. O'Neill, J. F. Innes

Botulinum toxin A intramuscular injections directly into the gastrocnemius muscle and passive physiotherapy marginally improved the range of motion in the talocrural joint.

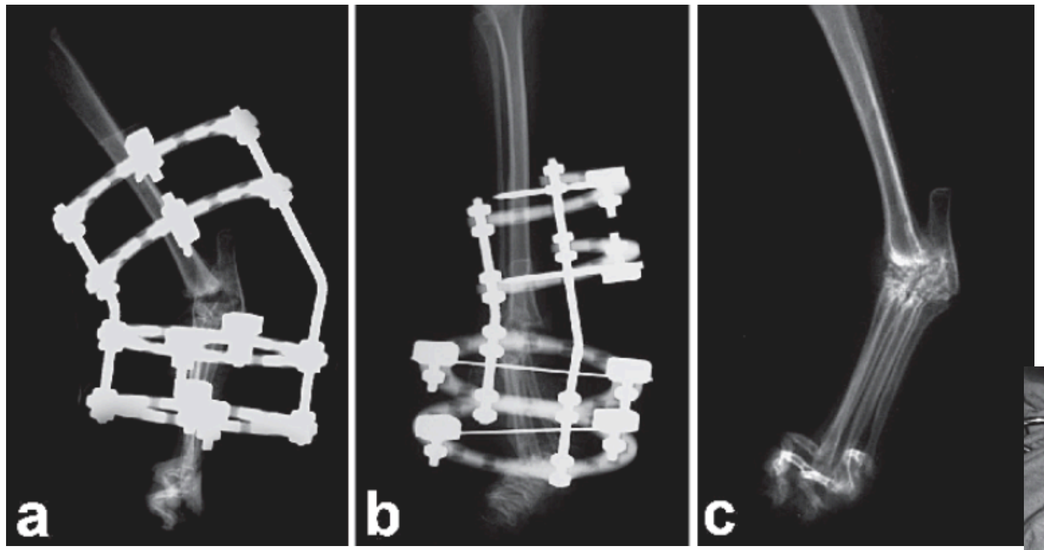
Definitive surgical treatment with partial tarsal arthodesis produced an excellent clinical outcome.

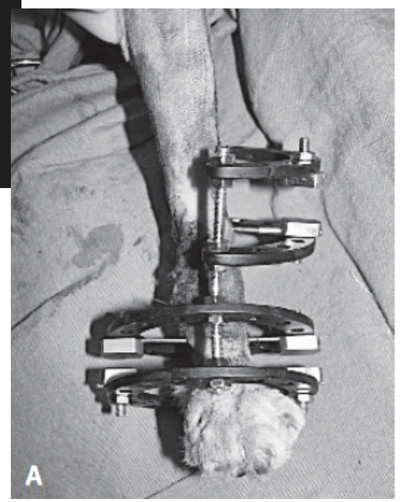
Treatment

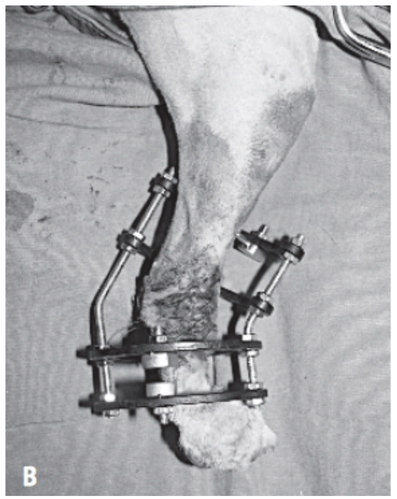
Surgical correction of a bilateral congenital tarsal hyperextension deformity in a cat

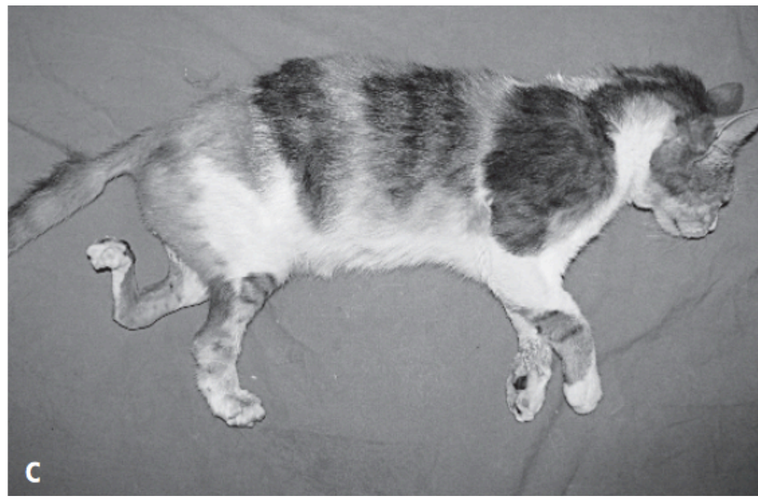
C. Yardımcı; A. Özak; H. Özlem Nisbet

Department of Surgery, Faculty of Veterinary Medicine, Ondokuz Mayıs University, Kurupelit, Samsun, Turkey



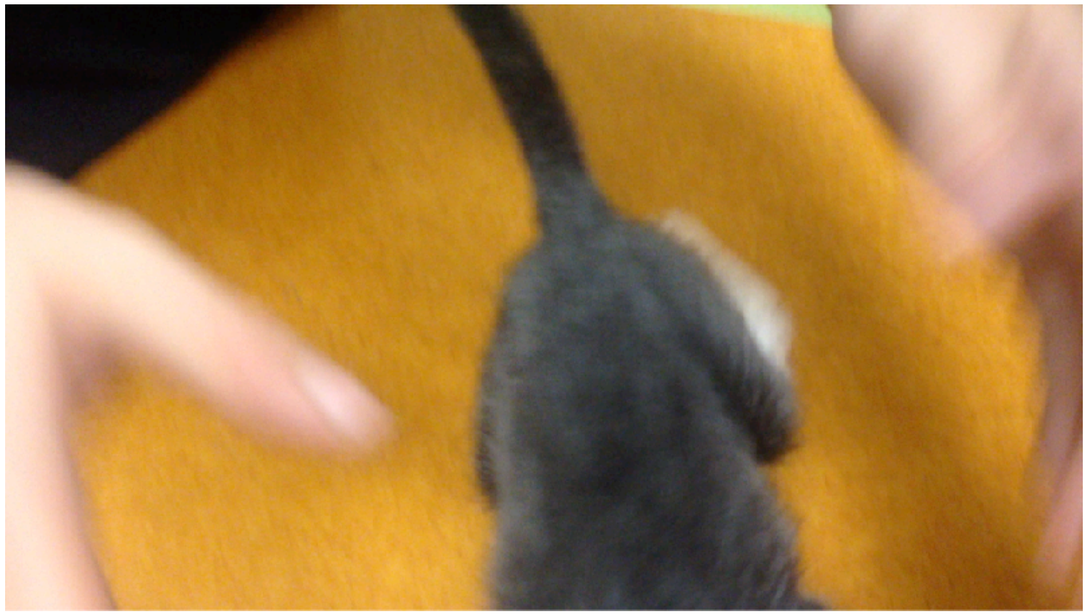




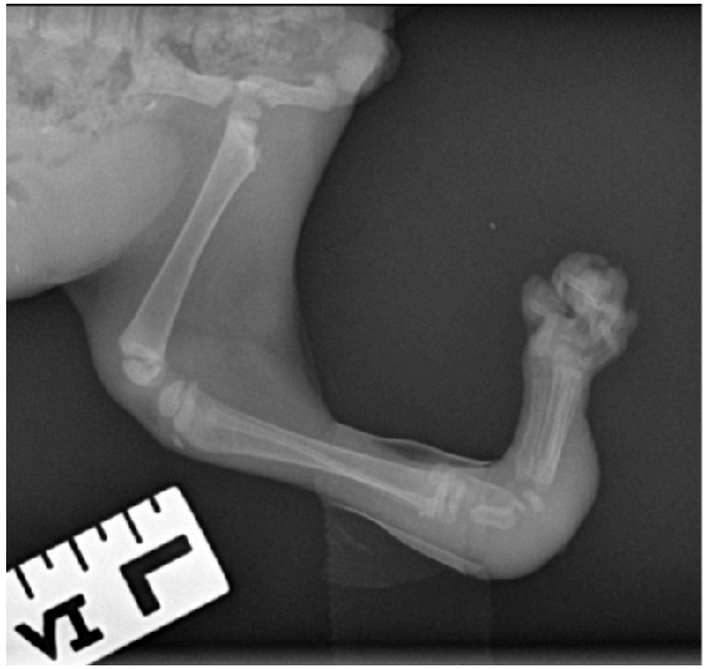


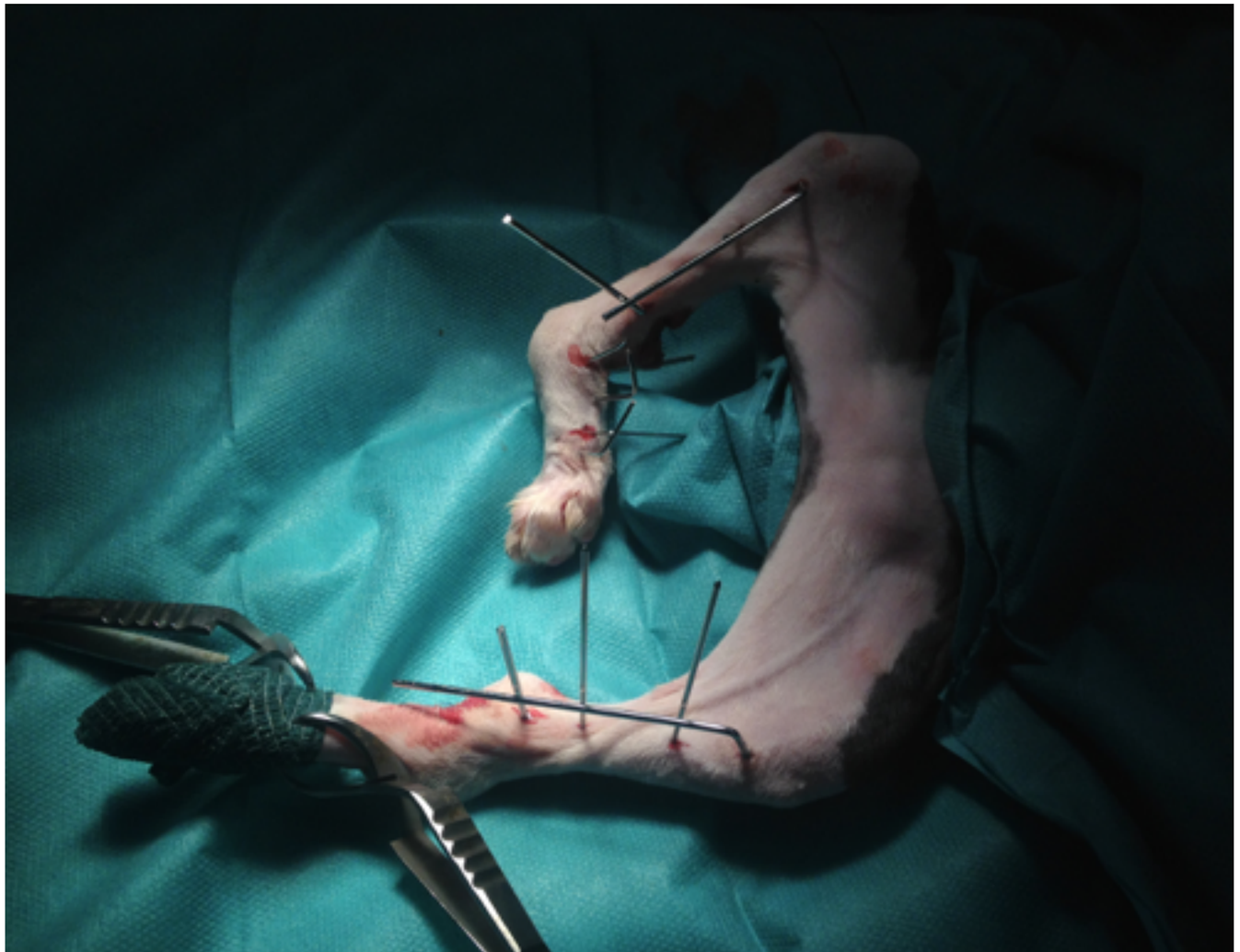
Amigo

2 m old, male, ESH, 900g



Bilateral tarsal hyperextension

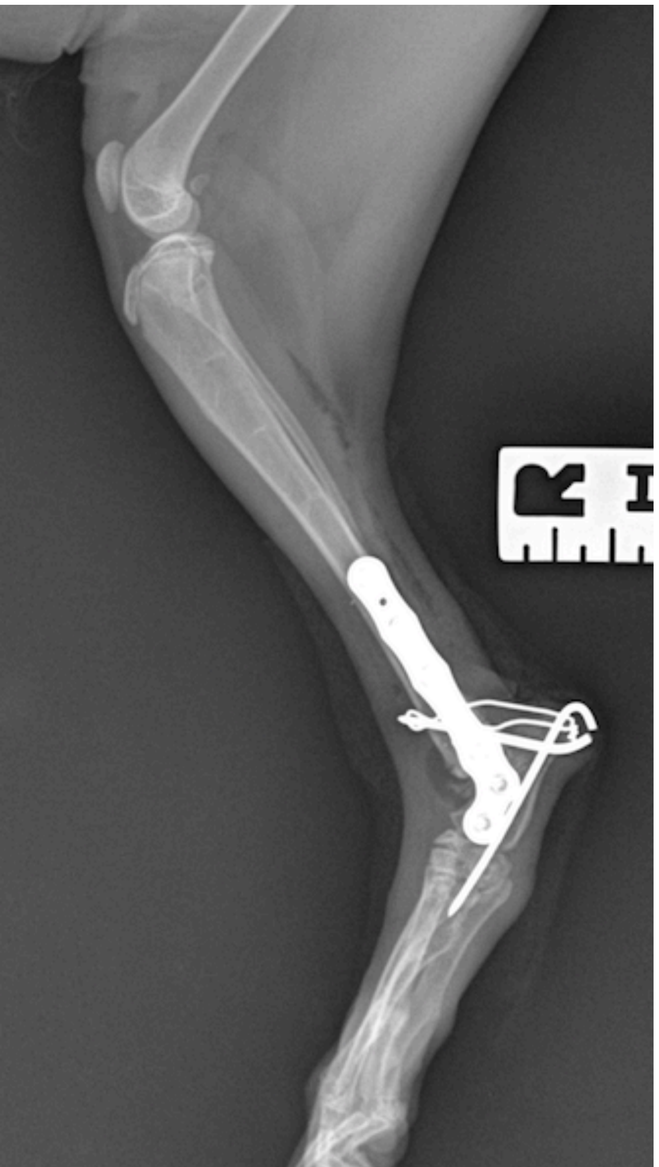


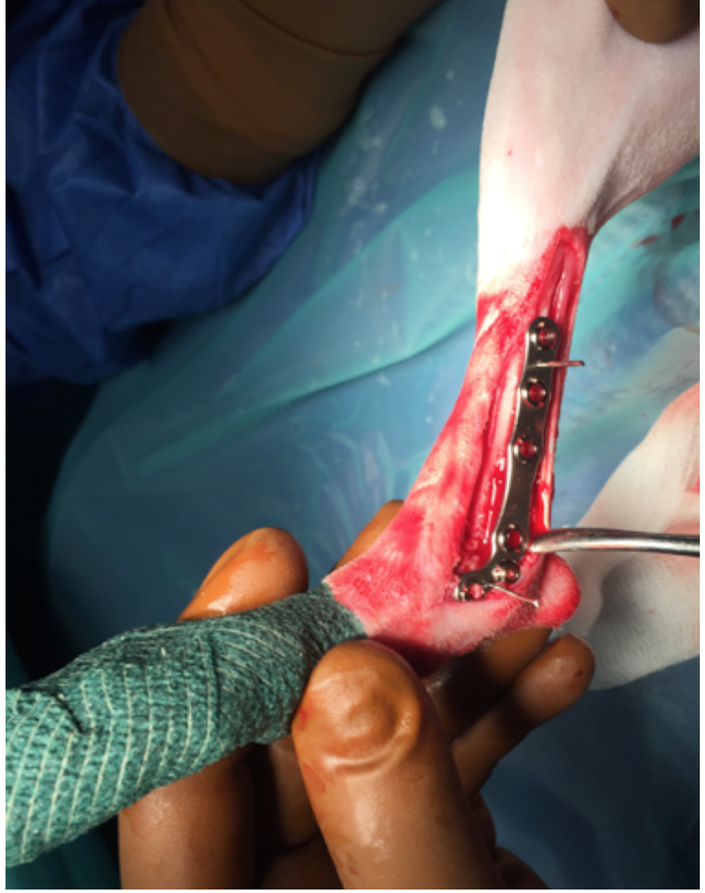


6 m of age, ESF removal

Tarsal hyperflexion

Bilateral partial tarsal arthrodesis





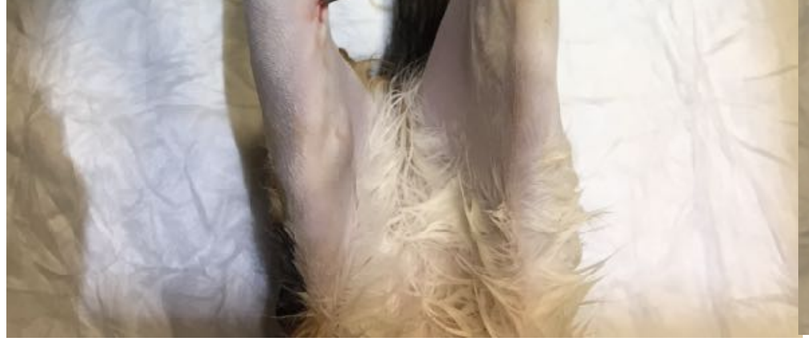
10 m age

Franki

7 m old, female, ESH,1,9 kg

Found on the street

Bilateral tarsal hyperextension

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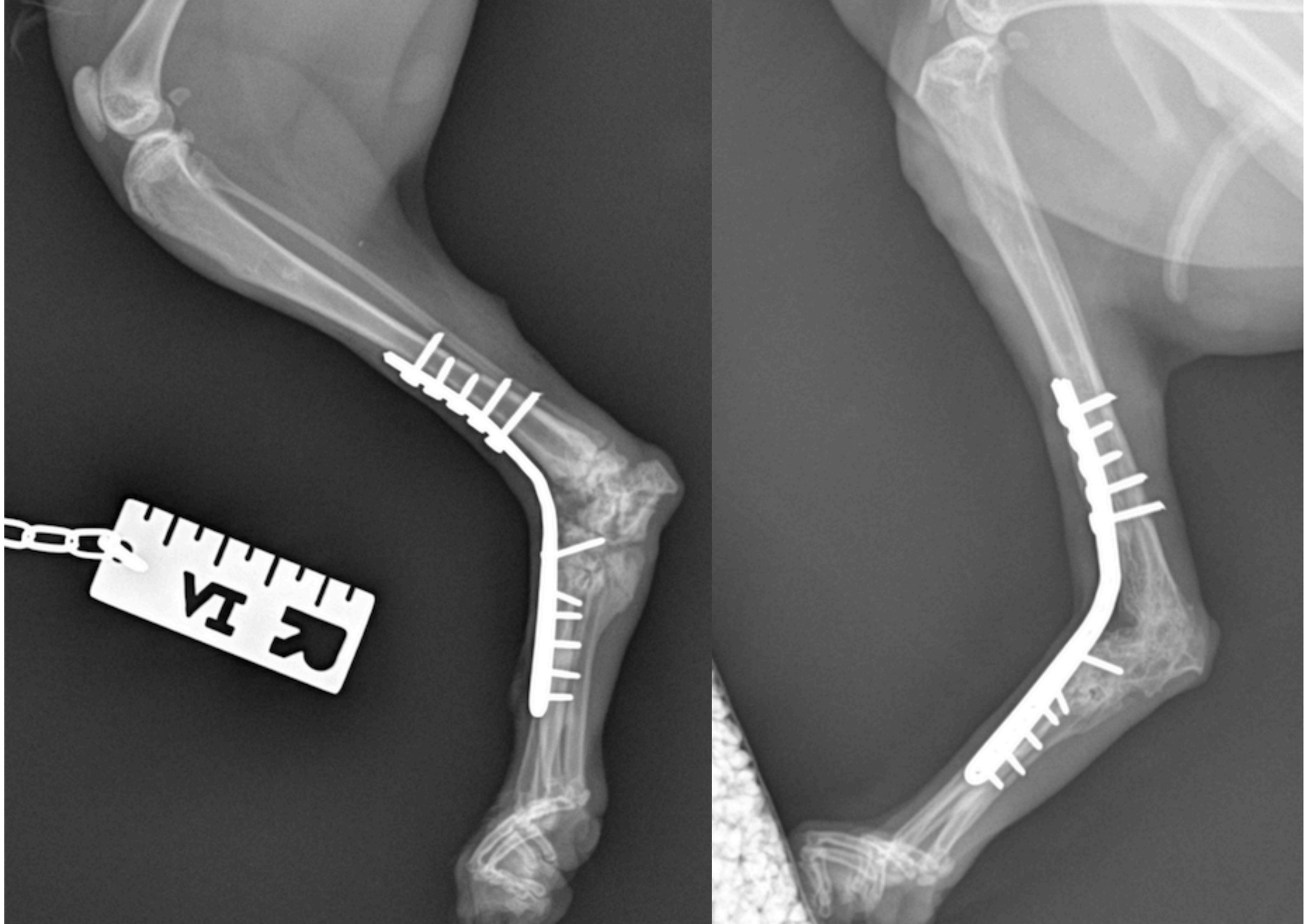
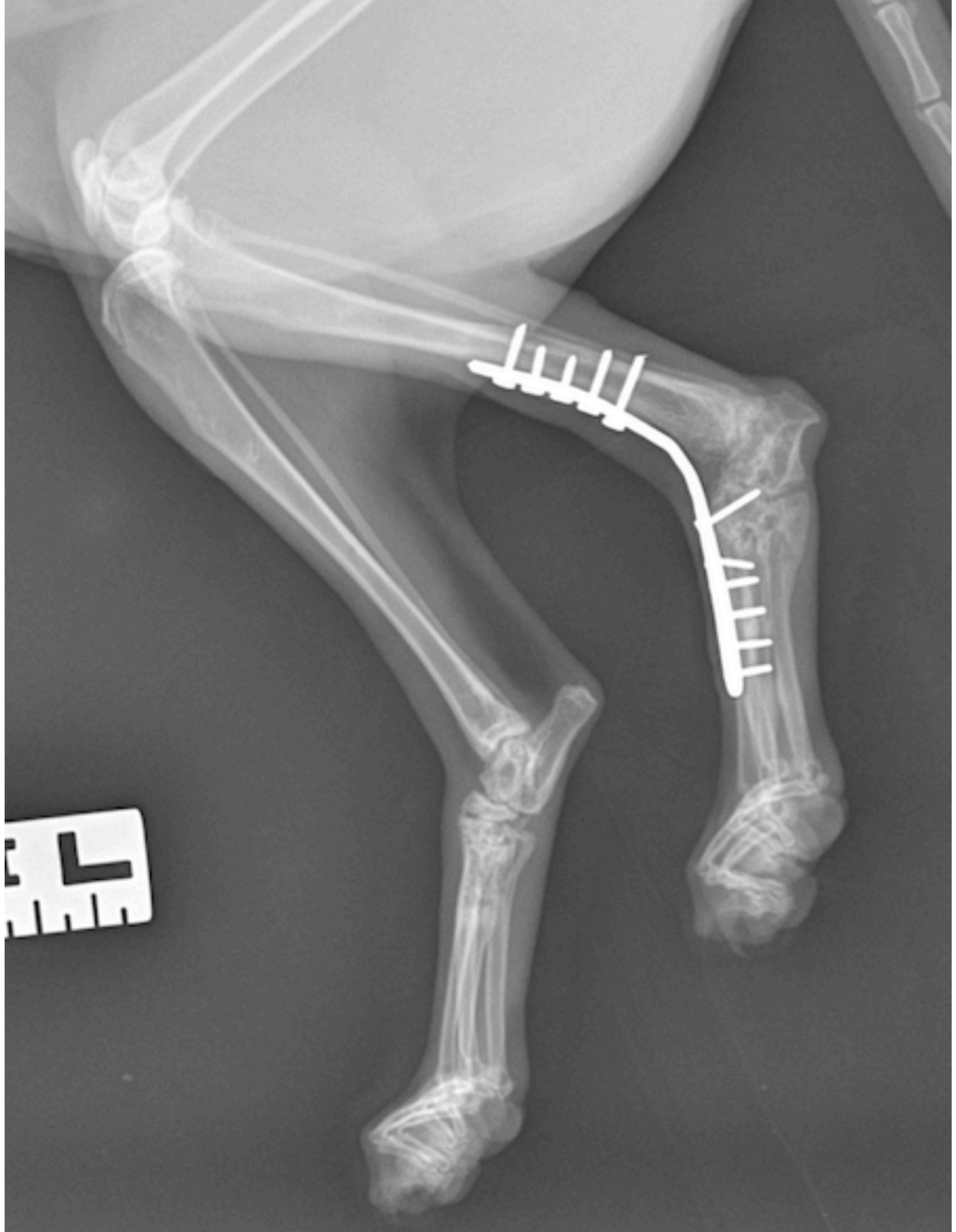
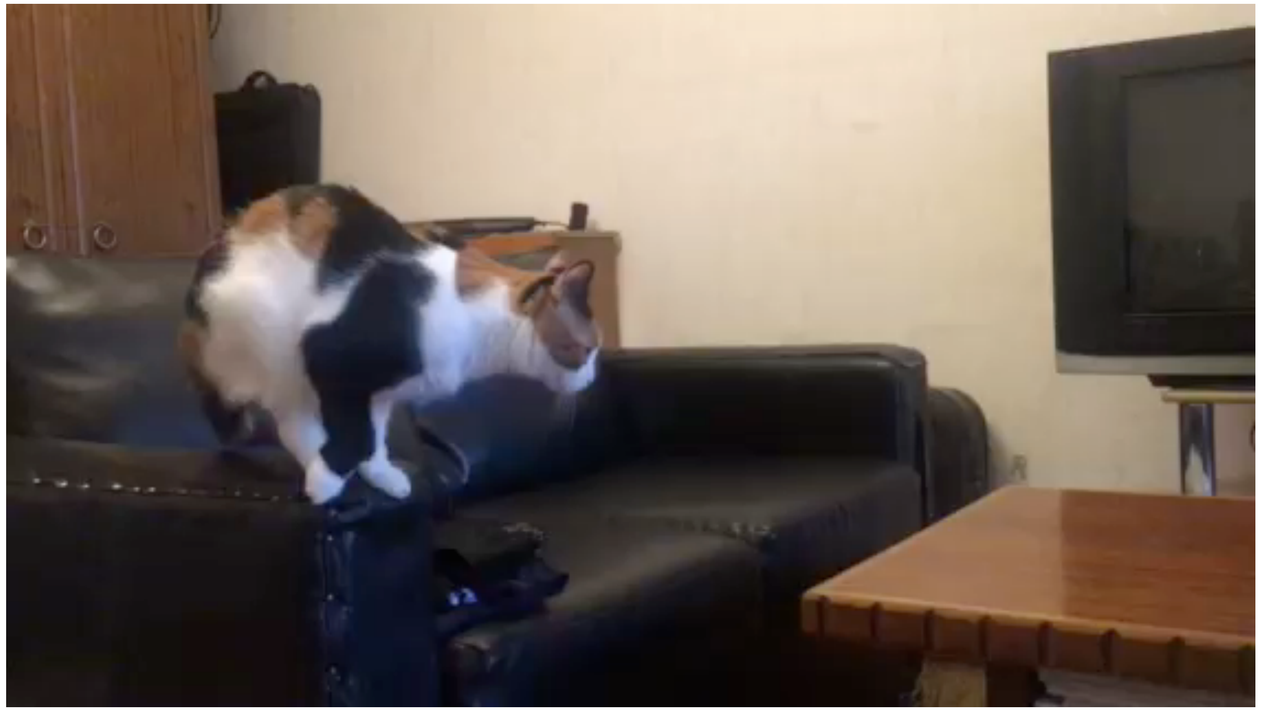


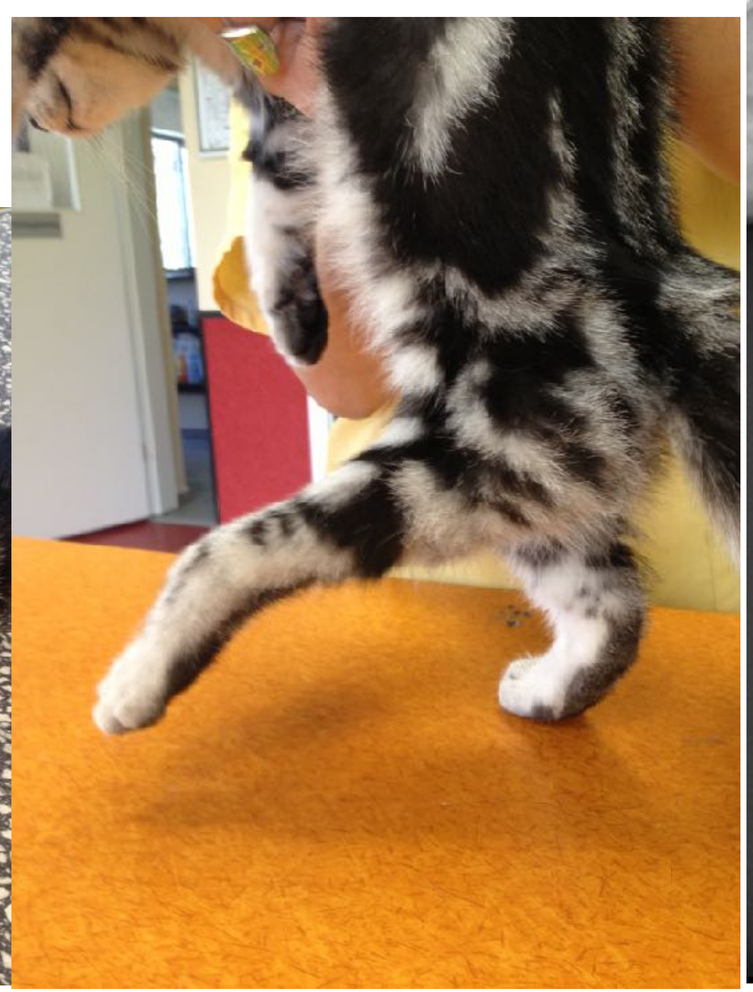




Congenital stifle hyperextension/ Genu recurvatum

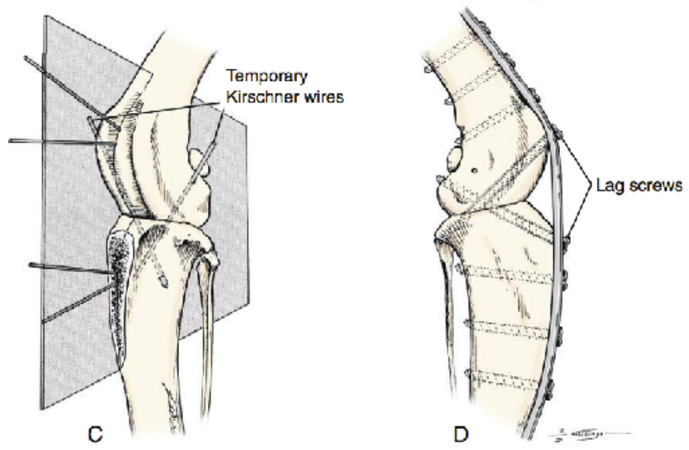
- Uncommon, Not clear etiology
- Embryonic myelitis affecting the lumbar motor center?
- **Neospora sp. associated**
- Unilateral or bilateral
- Dogs and cats

“Muscles dys-synergisim”



Treatment

- 3 m old kitten
- Unilaterally affected
- Stifle arthrodesis at the age of 6,5m



Functional result

Swimming puppy/kitten syndrome

- Rare, usually puppies
- Unknown etiology-congenital adductor weakness
- Inability for quadrupedal position primarily-pelvic limbs.
- Rarely- thoracic limbs, pact excavatum
- Usually 2-3 weeks after birth

Treatment

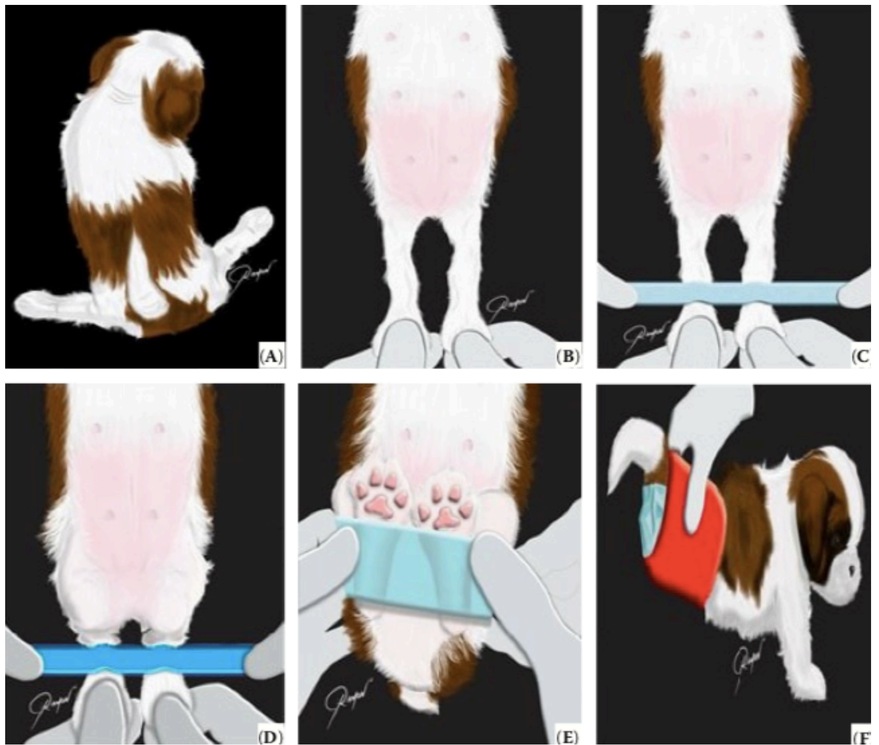
- Generally good prognosis
- Nutritional- restricted calories, Vitamin E, Selenium
- Physical therapy
- Hobble bandages

A modified technique for treating swimmer puppy syndrome

D.E. Karcher[1], R.C. Costa[1], T.C. Prada[1], P.C. Moraes[1], L.A. Ramon[2], B.W. Minto[1], L.G.G.G. Dias[1]*

[1]Faculty of Agricultural and Veterinary Sciences, State University "Julio de Mesquita Filho"/ UNESP, Jaboticabal, Brazil

[2]Faculty of Veterinary Medicine, Franca University, Franca, Brazil



Take home messages

- Congenital skeletal deformities have huge effect over the animal's quality of life.
- Curative treatment is not always possible.
- Theoretical knowledge is needed for recognition.
- Breeding prevention is the best possible solution.


Thank you!