

Stickler syndrome



Pain Management for the Stickler Patient

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Summary



- Same-day clinic consultation offered alongside Mr Snead's team (Thursday afternoon)
- Whole family attendance encouraged

Medical management of osteoarthritis

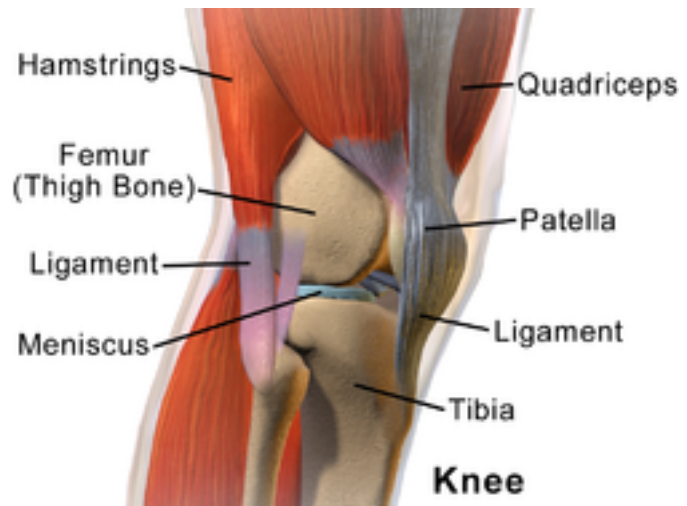
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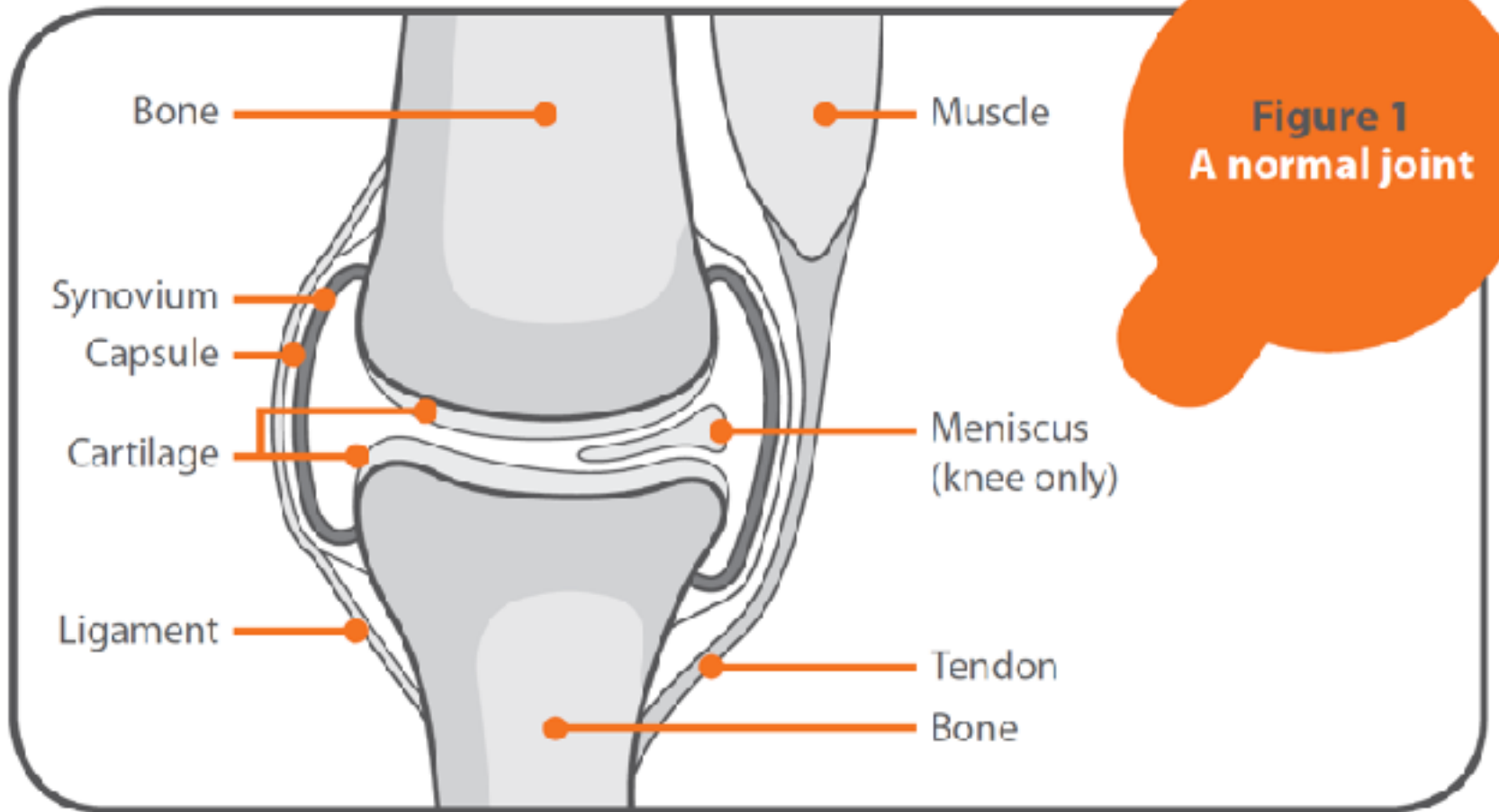
- Education
- Joint protection
- Pain management
- Physical therapies
- Liaison with surgeons

Stickler's Overview



- **Growing skeleton**
 - Foot and lower limb development
 - Writing
 - Exercise
- **Hypermobility**
- **Osteoarthritis**
- **Skeletal abnormalities**
- **Joint replacement**
- **Pain management**





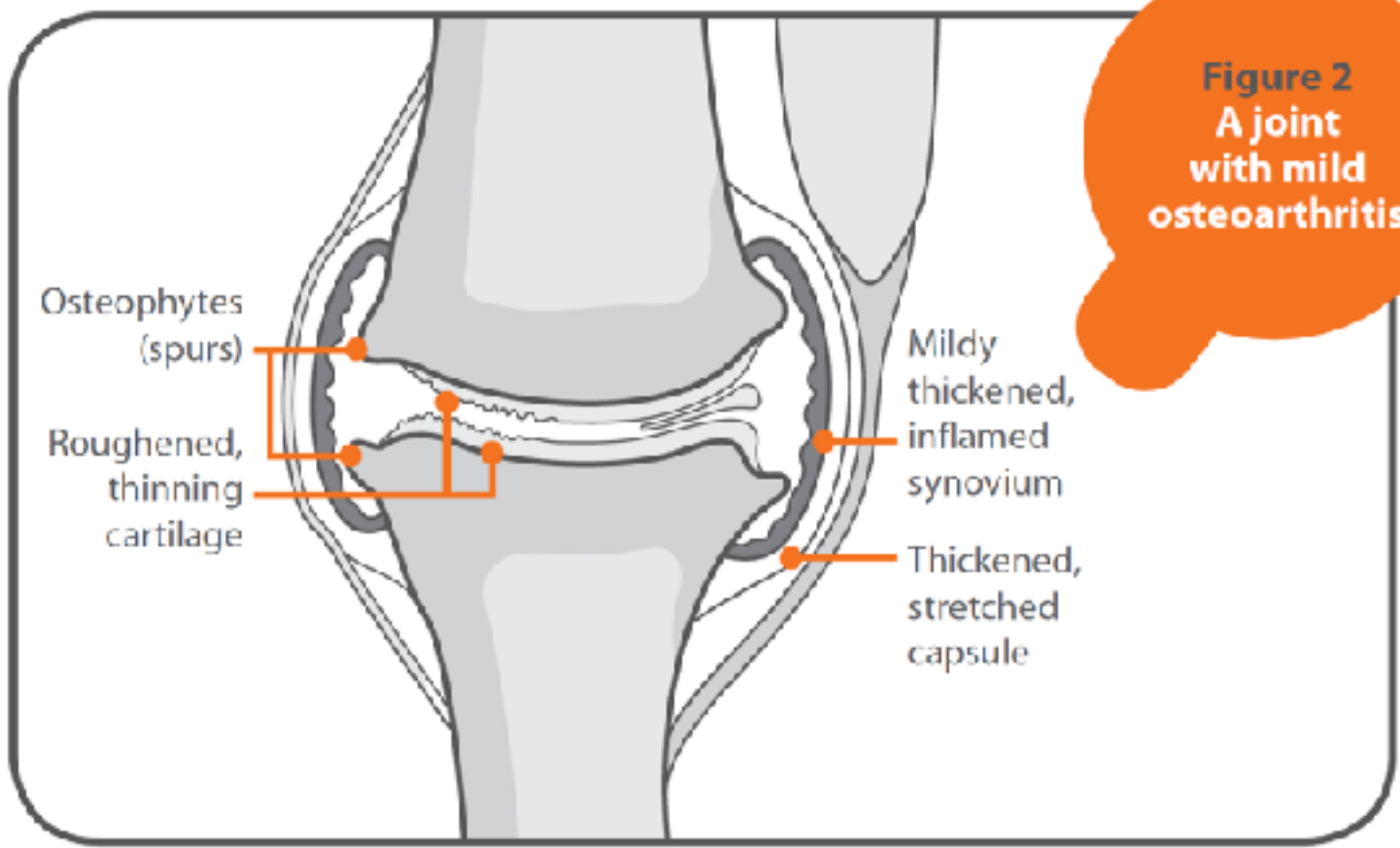


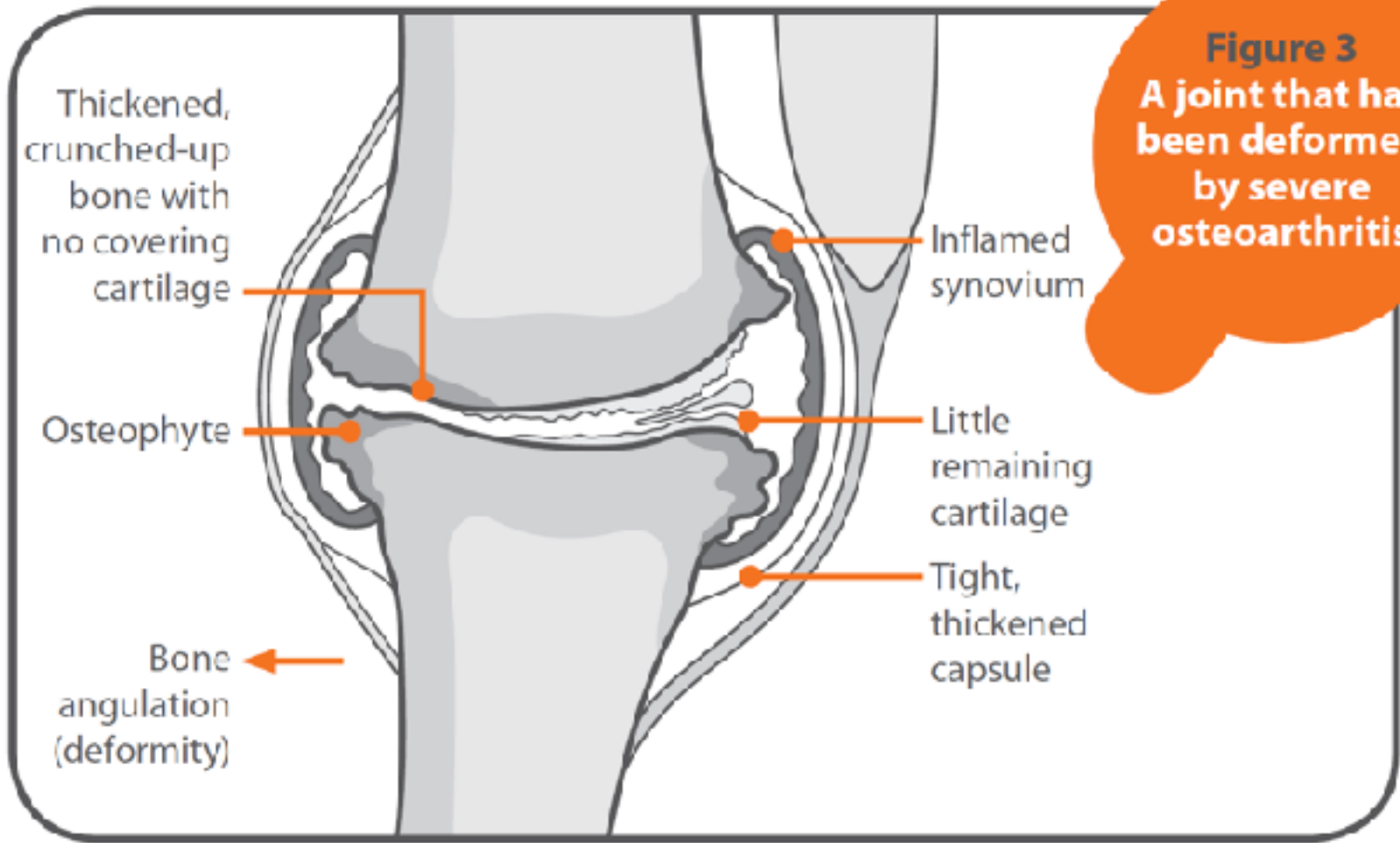
Figure 2
A joint
with mild
osteoarthritis

Osteophytes
(spurs)

Roughened,
thinning
cartilage

Mildly
thickened,
inflamed
synovium

Thickened,
stretched
capsule



What is pain?

International Association for the Study of Pain (IASP)



‘An unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage.’

Pain pathways

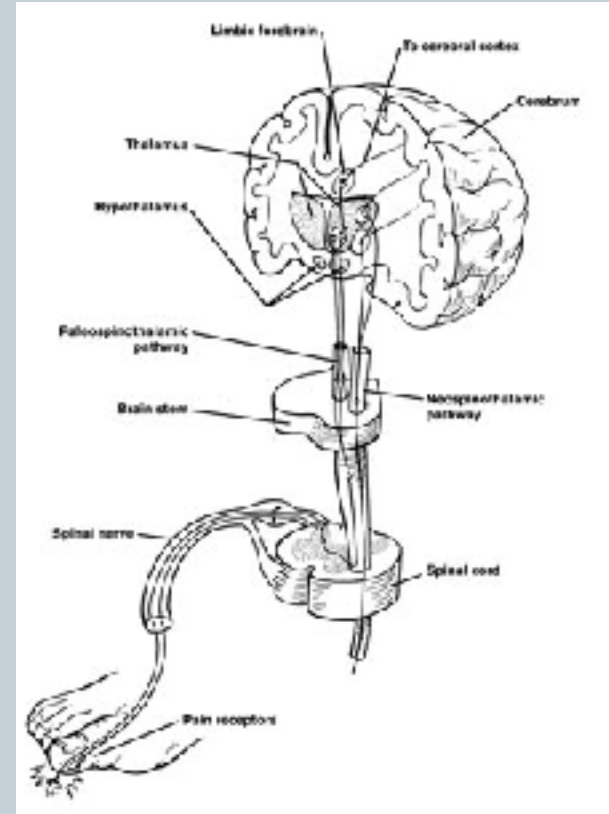
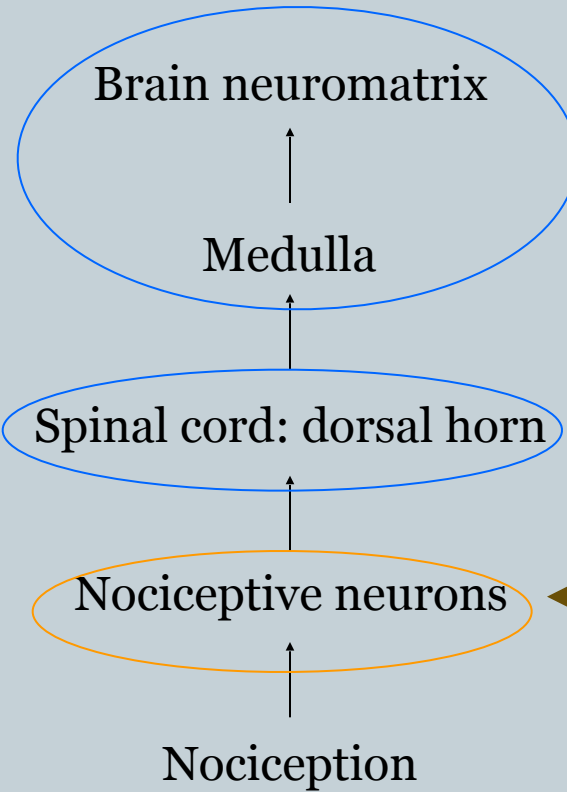


Learning
Neuroplasticity

Central
sensitisation

Central
sensitisation

Peripheral
sensitisation



Thermal

Mechanical

Chemical

Biopsychosocial model



Biological

Psychological

Social

Biopsychosocial model



Biological

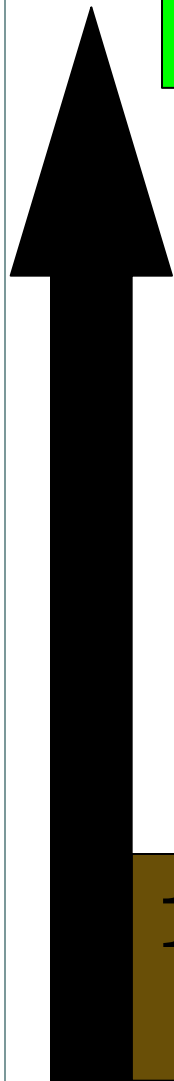
Psychological

Social



World Health Organization Pain Ladder

**Freedom from *cancer* pain, NOT musculoskeletal pain.
Avoid high doses of strong opioids**



3

*** Morphine**
Paracetamol
NSAID

Opioid for moderate to severe pain
+ /- Non-opioid
+ /- Adjuvant

Pain persisting or increasing

2

Codeine
Paracetamol
NSAID

Opioid for mild to moderate pain
+ /- Non-opioid
+ /- Adjuvant

Pain persisting or increasing

1

Paracetamol
NSAID

Non-opioid
+ /- Adjuvant

Antidepressants



- **Tricyclics (nortriptyline, amitriptyline)**
 - Different dosages for pain
 - May help with poor sleep pattern
 - ✦ E.g. Give amitriptyline 10mg two hours prior to sleep
- **SSRI (fluoxetine);**
- **SNRI (venlafaxine, duloxetine);**
- **NARI (reboxetine)**
 - Useful for BOTH pain and anxiety / depression related problems



- Anticonvulsant medications
- Gabapentin, Pregabalin, Carbamazepine, Phenytoin, Topiramate, Valproate etc
- Side effects – drowsiness, weight gain

Findings



- Hypermobility demonstrable or historical in 60%
- Small number of Marfanoid habitus (@5%)
- Hypermobile hindfoot evident
 - Pes planus
 - Ankle medial laxity
- Osteoarthritis
 - Patellofemoral

Other findings



- Scoliosis rare
- Short stature prevalent
- Normal skin, no bruising,
- No cardiac involvement
 - 1 childhood murmur

Brief Pain Inventory



- Average pain score 4.1/10
- Best - worst pain: 2.5 -5.5/10
- 47/91 patients took no medication (av. pain 3.4/10)
- 44/91 patients took medication that provided about 50% (av. pain 4.8/10)
- Most frequent medication was NSAID +/- paracetamol (31/44)
- 65/91 reported that their knee was painful and those that did reported a pain score of 4.4 versus 3.3)

Pain interference scores



- General activity 3.79
 - Mood 3.50
 - Walking 4.16
 - Working 4.15
 - Relationships 2.36
 - Sleep 3.65
 - Quality of life 3.72
-
- *Weak correlations with age.*
 - *No correlation with gender nor Stickler type*

Stickler syndrome



A musculoskeletal overview

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Research and Development Department approval



- R&D ref: A093076
- Mr McArthur, Mr Rehm, Dr Tanner, Dr Bearcroft
- Radiological abnormalities in Stickler syndrome patients
- Identified 240 children with Stickler syndrome on our database. 75 of these had radiographs taken of their knees, pelvis and/or spine.

Patient demographics



- **Stickler subtype**
 - Type 1 – 51
 - Type 2 – 17
 - Not available – 7
- **Gender ratio**
 - M 44 : F 31
- **Average age**
 - Knee radiographs: 9.4 y
 - Hip radiographs: 10.8 y
 - Spine radiographs: 8.8 y

Radiographs



- AP Pelvis – 61
- Knee – 102
- Spine - 61

Results - Knees



- 59 % of knees present no abnormality of note

Results and discussion - Knees



- 24 % - multiple Harris-lines in proximal tibia and distal femur

- Cause of Harris lines still debated



Results and discussion knees

- 5 % - hypoplastic lateral femoral condyles
- 4 % - Osteochondral defects

- Lateral femoral Hypoplasia not described in literature
- OCD present in 15 – 30 in 100000¹



- 3 % - fibrous cortical defects
- 1% - Varus deformity at tibial metaphysis
- 1% - Valgus deformity at tibial metaphysis

¹ Obedian RS, Grelsamer RP (January 1997). "Osteochondritis dissecans of the distal femur and patella". Clinical Journal of Sports Medicine 16 (1): 157–74. doi: 10.1016/S0278-5919(05)70012-0. PMID 9012566

Results - Hips



- 82 % (50 patients) of AP Pelvis x-rays reveal no abnormality

Results and Discussion - Hips



- 7 % (4 patients) - lower centre of femoral head rotation bilaterally
- 1.6 % (1 patient) - left sided Perthes disease
- 1.6 % (1 patient) - epiphyseal fragmentation

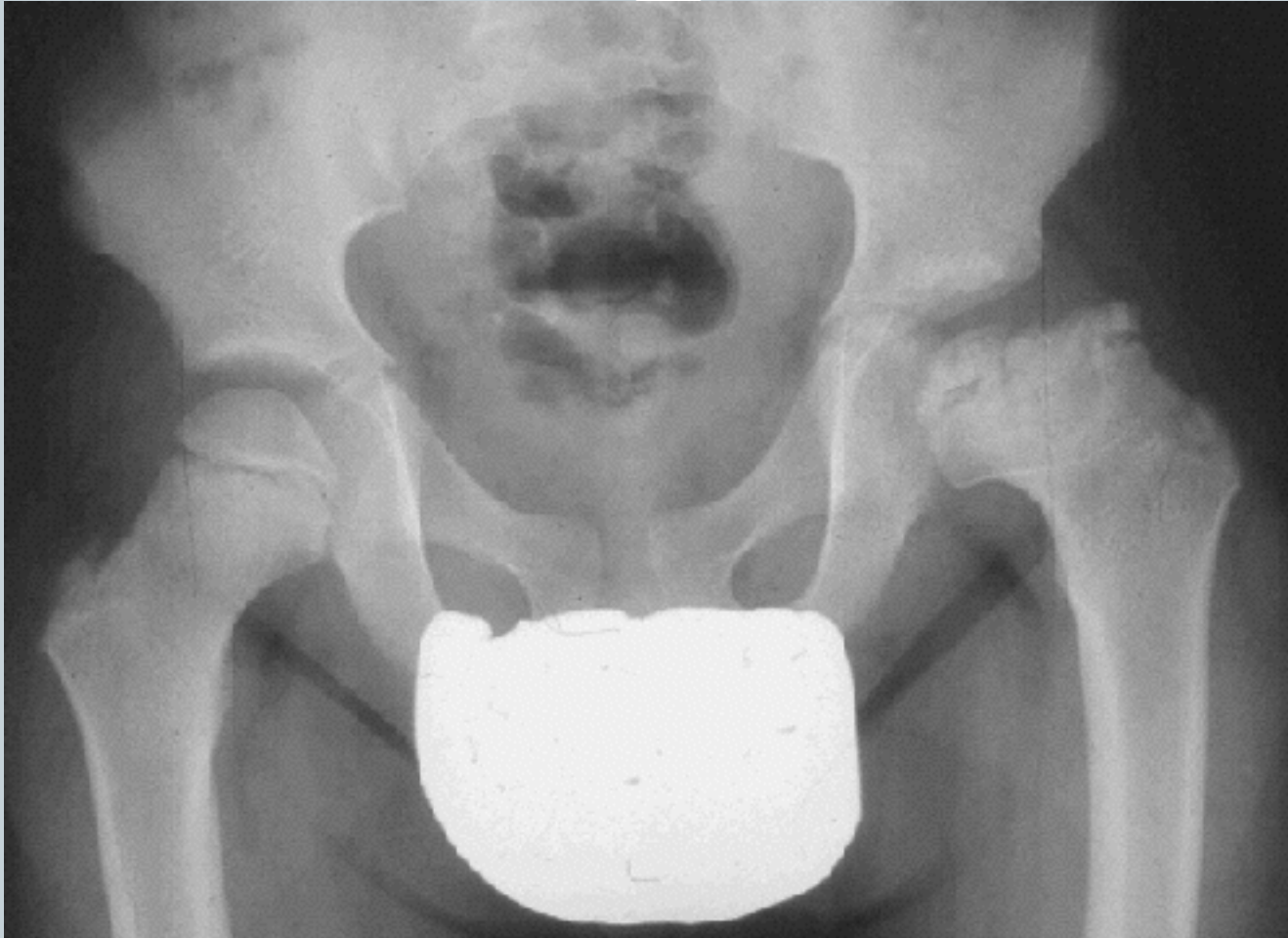
- Low centre of hip rotation can occur post Perthes disease. Femoral heads however appeared normal in our radiographs
- No incidence of epiphyseal fragmentation described. DD include:
 - Hypothyroidism
 - Perthes
 - Multiple epiphyseal dysplasia

Perthes Disease



- Self limiting hip disorder caused by a varying degree of ischemia and subsequent necrosis of the femoral head
- Avascular necrosis of nucleus of proximal femoral epiphysis, abnormal growth of the physis, and eventual remodelling of regenerated bone are the key features of this disorder
- Loss of blood supply to the epiphysis is thought to be the essential lesion
- Normally seen in 4 to 8 yr old boy with delayed skeletal maturity
- male to female ratio: 4-5 to 1
- Increased incidence with a positive family history, low birth weight, and abnormal pregnancy / delivery;
 - up to 12% of cases are bilateral but will be at different stages & are asymmetric
 - age is the key to the prognosis - after 8 yr represents poor prognosis

Perthes – coxa magna



Results and Discussion - Hips



- 1.6 % (1 patient) - bilateral valgus hips (NSA 160°)

- No incidence of coxa valga described. DD include:
 - Trauma
 - Cerebral palsy

Results - Spine



- 10 % (6 patients) – platyspondyly



- Congenital Platyspondyly – present in
 - Thanatophoric dwarfism
 - Metatropic dwarfism
 - Osteogenesis imperfecta type IIA
 - Homozygous achondroplasia

- Platyspondyly in later childhood
 - Morquio`s disease
 - Spondyloepiphyseal dysplasia congenita
 - Spondyloepiphyseal dysplasia tarda
 - Kniest syndrome

Results and Discussion - Spine



- 10 % (6 patients) – loss of lumbar lordosis
- 5 % (3 patients) – flatspine
- 3 % (2 patients) – kyphosis lumbar spine

- **Loss of lordosis and flatspine**
 - De Novo Scoliosis
 - Iatrogenic
- Scheuermann's kyphosis of the lumbar spine.

Results and Discussion - Spine



- 5 % (3 patients) – spina bifida occulta

- Present in 10 - 20 % of the population^{1,2}

1) Lambert, H. Wayne; Wineski, Lawrence E. (2011). *Anatomy & Embryology*. Wolters Kluwer. p. 100.

2) Jump up ^ "Spina Bifida Fact Sheet". National Institute of Neurological Disorders and Stroke. 2013.

Results and Discussion - Spine



- 3 % (2 patients) – scoliosis

• Scoliosis

○ Prevalence 0.47 – 5.2%^{1,2,3,4,5,6,7}

1. Suh SW, Modi HN, Yang JH, Hong JY. Idiopathic scoliosis in Korean schoolchildren: a prospective screening study of over 1 million children. *Eur Spine J.* 2011;20(7):1087–1094. doi: 10.1007/s00586-011-1695-8.
2. Nery LS, Halpern R, Nery PC, Nehme KP, Stein AT. Prevalence of scoliosis among school students in a town in southern Brazil. *Sao Paulo Med J.* 2010;128(2):69–73. doi: 10.1590/S1516-31802010000200005.
3. Daruwalla JS, Balasubramaniam P, Chay SO, Rajan U, Lee HP. Idiopathic scoliosis. Prevalence and ethnic distribution in Singapore schoolchildren. *J Bone Joint Surg Br.* 1985;67(2):182–184.
4. Wong HK, Hui JH, Rajan U, Chia HP. Idiopathic scoliosis in Singapore schoolchildren: a prevalence study 15 years into the screening program. *Spine (Phila Pa 1976)* 2005;30(10):1188–1196. doi: 10.1097/01.brs.0000162280.95076.bb. [PubMed] [Cross Ref]
5. Cilli K, Tezeren G, Taş T, Bulut O, Oztürk H, Oztemur Z, Unsaldi T. School screening for scoliosis in Sivas, Turkey. *Acta Orthop Traumatol Turc.* 2009;43(5):426–430. doi: 10.3944/AOTT.2009.426.
6. Soucacos PN, Soucacos PK, Zacharis KC, Beris AE, Xenakis TA. School-screening for scoliosis. A prospective epidemiological study in northwestern and central Greece. *J Bone Joint Surg Am.* 1997;79(10):1498–1503.
7. Wynne-Davies R. Familial (idiopathic) scoliosis. A family survey. *J Bone Joint Surg Br.* 1968;50:24–30.

Conclusion



- Our study cannot confirm the high prevalence of orthopaedic abnormalities as described in previous literature
- Fibrous cortical defects, scoliosis and spina bifida occulta have a similar incidence in the Stickler population as they do in the general population.

Conclusion



- Relatively mild musculoskeletal disease
- Knee is the most symptomatic
- Small number of patients with structural defects
- Hindfoot may have a role to play in the growing skeleton

- Stickler may be a useful model for other hypermobile conditions