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

- 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 1
A normal joint

- Bone
- Muscle
- Synovium
- Capsule
- Cartilage
- Meniscus (knee only)
- Ligament
- Tendon
- Bone
Figure 2
A joint with mild osteoarthritis

- Osteophytes (spurs)
- Roughened, thinning cartilage
- Mildly thickened, inflamed synovium
- Thickened, stretched capsule
Figure 3
A joint that has been deformed by severe osteoarthritis.
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

Peripheral sensitisation

Brain neuromatrix

Medulla

Spinal cord: dorsal horn

Nociceptive neurons

Nociception

Thermal

Mechanical

Chemical
Biopsychosocial model

- Biological
- Psychological
- Social
Biopsychosocial model

Biological

Psychological

Social
HONEY, DID YOU WANT THE ONE THAT CURES HEADACHE AND CAUSES STROKE OR EASES BACK PAIN BUT CAUSES HEART ATTACK?
World Health Organization Pain Ladder

1

Paracetamol
NSAID
+ /- Adjuvant

Non-opioid

2

Codeine
Paracetamol
NSAID

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

Pain persisting or increasing

3

Morphine
Paracetamol
NSAID

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

Pain persisting or increasing

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

- 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
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 100000

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

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 population¹,²

Results and Discussion - Spine

- 3% (2 patients) – scoliosis

Scoliosis

- **Prevalence 0.47 – 5.2%** \(^1,2,3,4,5,6,7\)


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