



THE **MARFAN**  
FOUNDATION

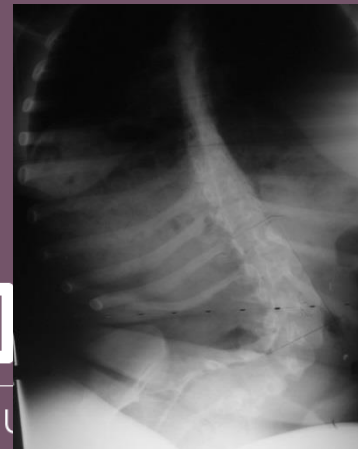
Know the signs. Fight for victory.

# HEALTHY LIMBS, FEET & SPINE

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ANNUAL



# TOPICS

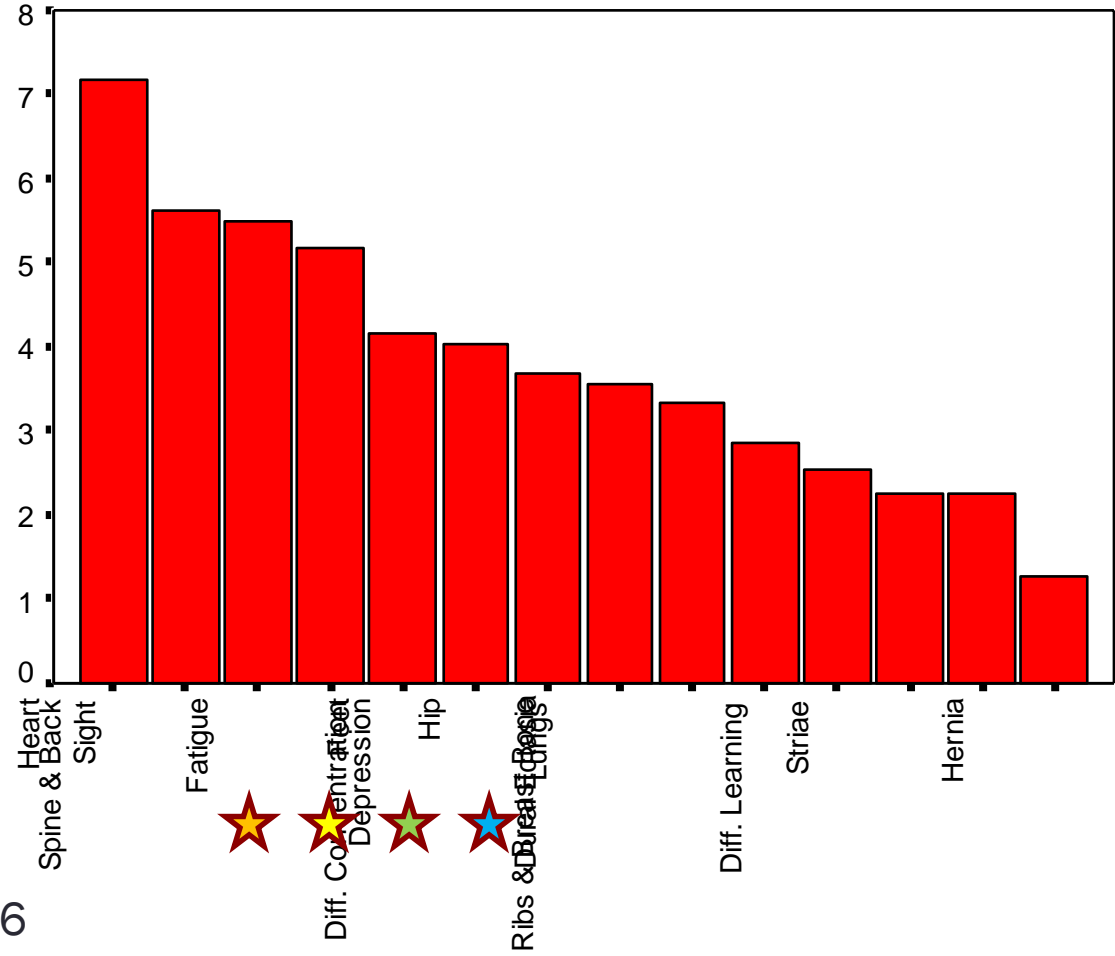
Spine issues

Dural Ectasia

Hips

# I. MAIN HEALTH CONCERNS IN MFS

Heart  
Sight  
Back  
Fatigue

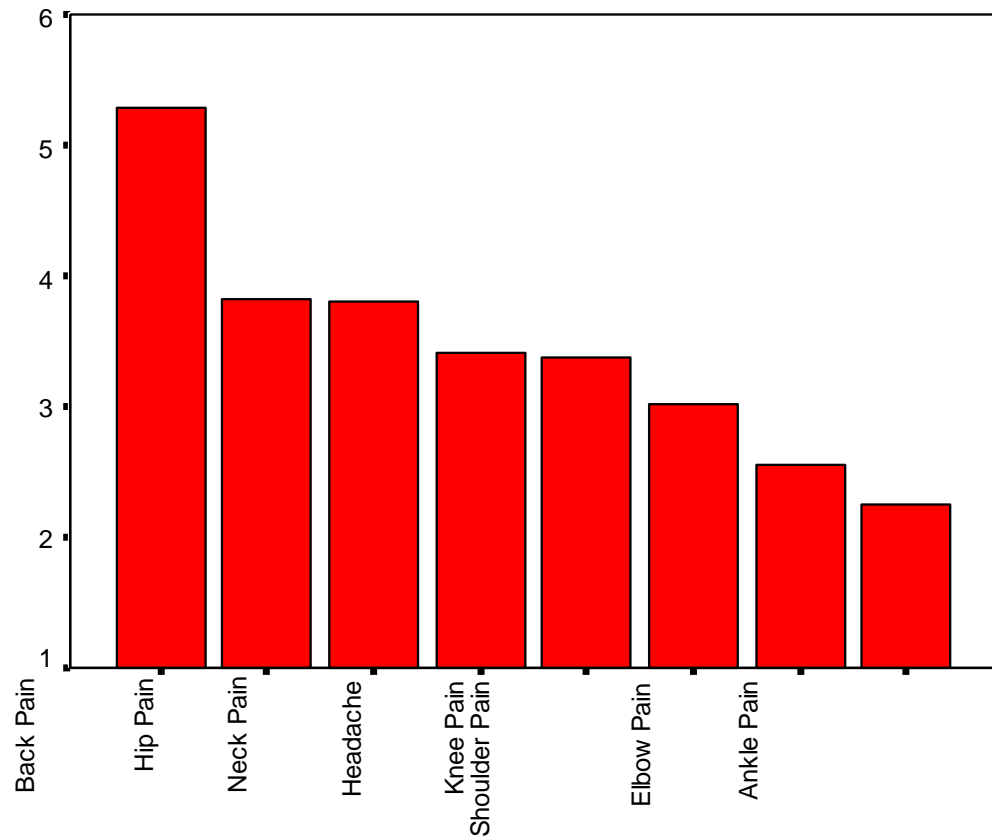


Rao, Sponseller 2016

# MAIN SOURCES OF SKELETAL PAIN

## -FURTHER RESEARCH NEEDED ON THESE

Back  
Hip  
Neck  
Knee  
Shoulder  
Feet



## II. SPINE ISSUES

Scoliosis

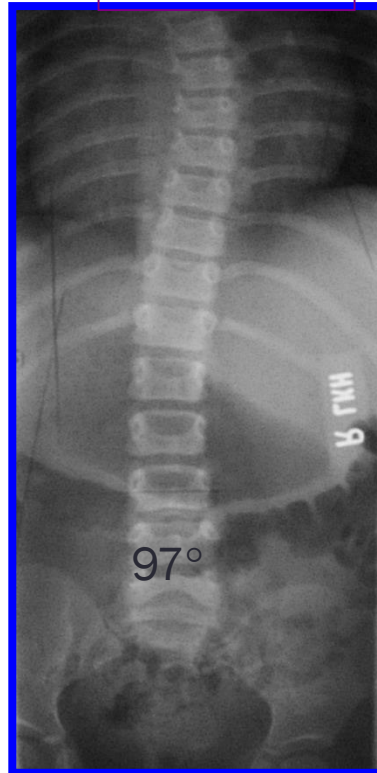
Kyphosis

Slipping (Spondylolisthesis)

# Scoliosis in Marfan Syndrome

Scoliosis affects 2/3 of MF  
Does not always worsen  
10% need brace or surgery  
Curves increase faster  
Increased back pain

Mild  
Scoliosis



3 years  
later



# WILL BRACE HELP SCOLIOSIS?

Straighten curve? – No

“Hold” it? – Maybe

Results of bracing:

- 20% successes
- 80% failures

Same principle for all connective tissue disorders!!



# BRACE RECOMMENDATIONS

## Consider brace at earlier point

- 15-20° curves in young children

## Avoid in “low-yield” situations

- Bigger curves, reluctant kids

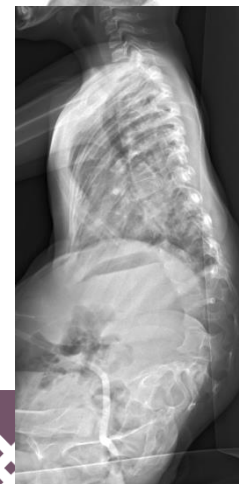


# INFANTILE MARFAN SYNDROME

Requiring surgery before age 9

Fusion arrests trunk growth

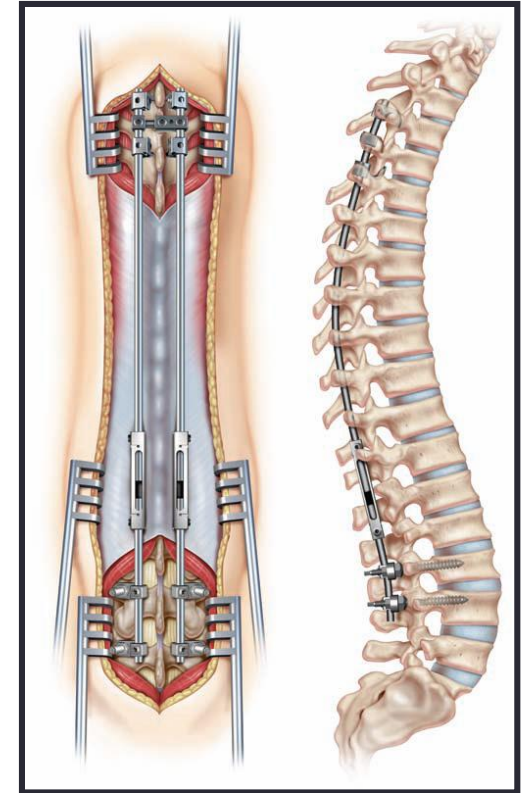
Solution: "Growing" rods (internal brace)



# GROWING ROD RESULTS IN MARFAN

## 10 Marfan children

- 80° curves
- 10 cm length in 5 years
- No infections



# MAGNETICALLY CONTROLLED GROWING RODS ("MAGEC")

If No need for MRI



# ADOLESCENT SCOLIOSIS SURGERY: MFS VERSUS IDIOPATHIC (TYPICAL)

Multicenter, Case-Control

34 Marfan patients

68 AIS patients

- Matched 1:2 for age, gender and degree of major deformity



# RESULTS

## Marfan vs. AIS:

- More Rod Complications (**3 vs 1**,  $p = 0.007$ )
- More Reoperations (**8 vs 0**,  $p = 0.01$ )
- More Intraoperative CSF Leaks (**3 vs 0**,  $p = 0.01$ )
- Progression of Unfused Thoracic Curves (**3 vs 1**,  $p = 0.107$ )

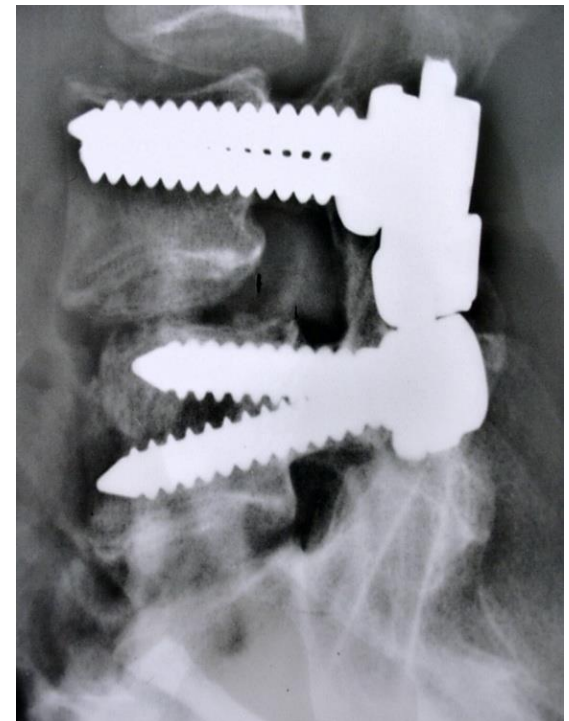
# SPONDYLOLISTHESIS

Not more common, but more likely to slip

L4-5, L5-S1

Grades 2-5

Posterior fusion only



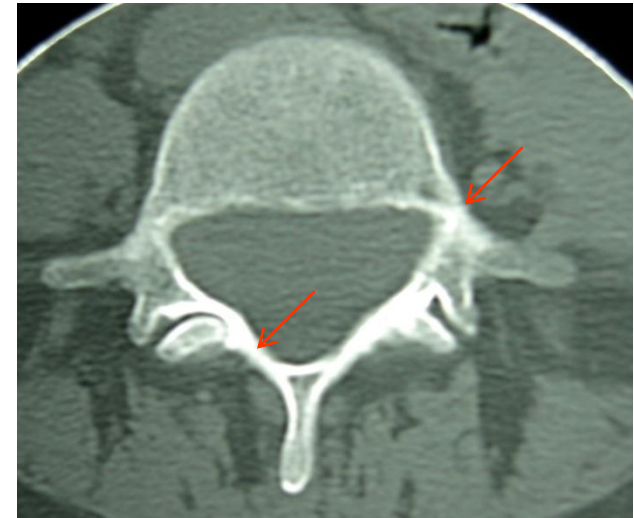


# BONY CHANGES

Pedicles thin

Lamina thin

Vertebrae scalloped



# IIC. DURAL ECTASIA

Present in over 50% of patients

- LDS similar

Visual criteria:

- dural diameter S1 > L4
- scalloping > 4 mm
- anterior meningocele

What is Dura?

- Tough tube protecting spinal cord
- Contains clear fluid - CSF

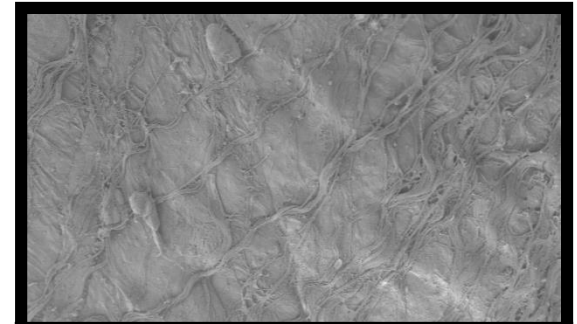




# HOW DOES DURAL ECTASIA DEVELOP?

Spinal Fluid pulsations

NI

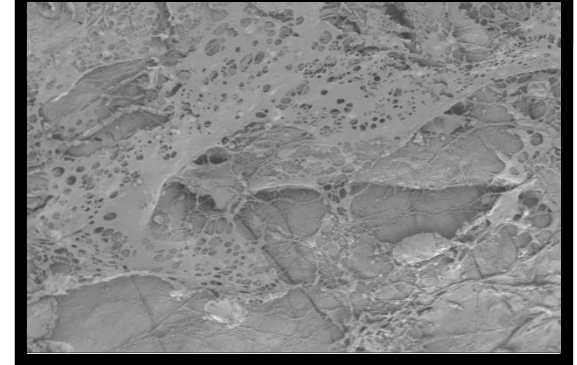


Dilatation of the weak dura



Scalloping/erosion of the vertebral body

DE



Signaling effects?



# DOES DURAL ECTASIA CAUSE PAIN? YES, BUT...

## Case-control study 30-50 y/o

- DE greater in pain group
- But not all with DE had pain!

## Possible reasons for pain:

- pressure on bone, soft tissues
- folding of dura or nerve roots

## Treatment?



# SIGNS/SYMPTOMS ASSOCIATED WITH DURAL ECTASIA

Very Low Back pain (91%)

Headaches (77%)

Leg pain (64%)

Pelvic & Perineal pain (30%)

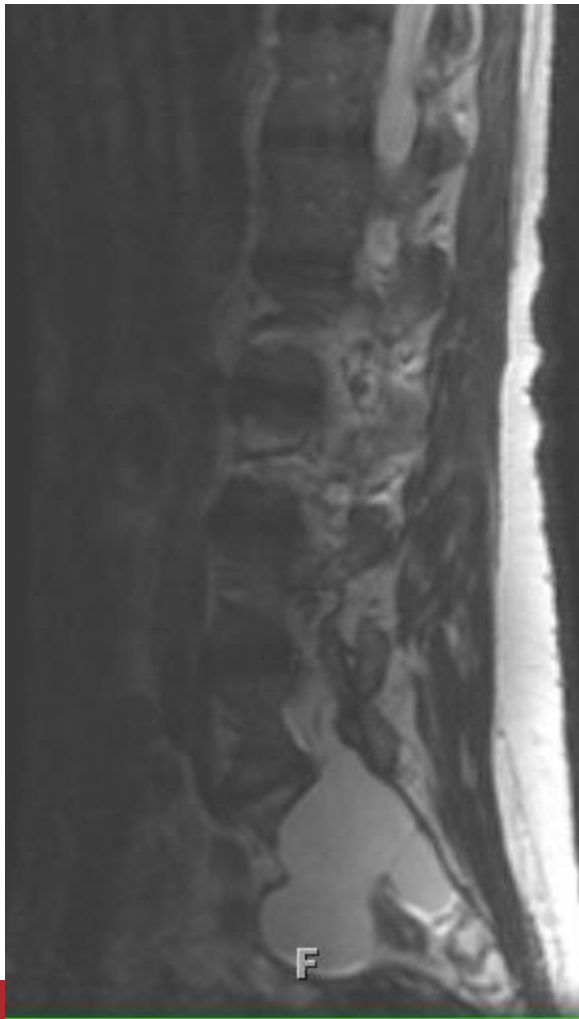
# DOES DURAL ECTASIA GET WORSE?

11 yr follow up

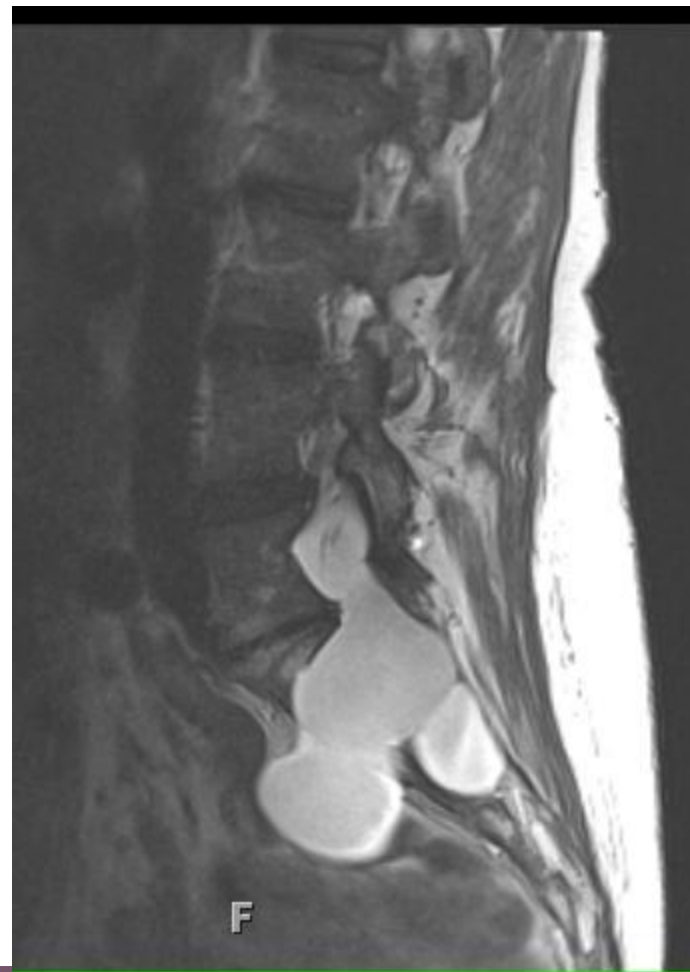
12 patients

- repeat MRI/CT/pain score
- age 52y/o (42-60)

# 60Y/O WOMAN WITH SEVERE DE (ANTERIOR MENINGOCELE)



1998



2009

# 48Y/O MALE WITH MODERATE DE



1998

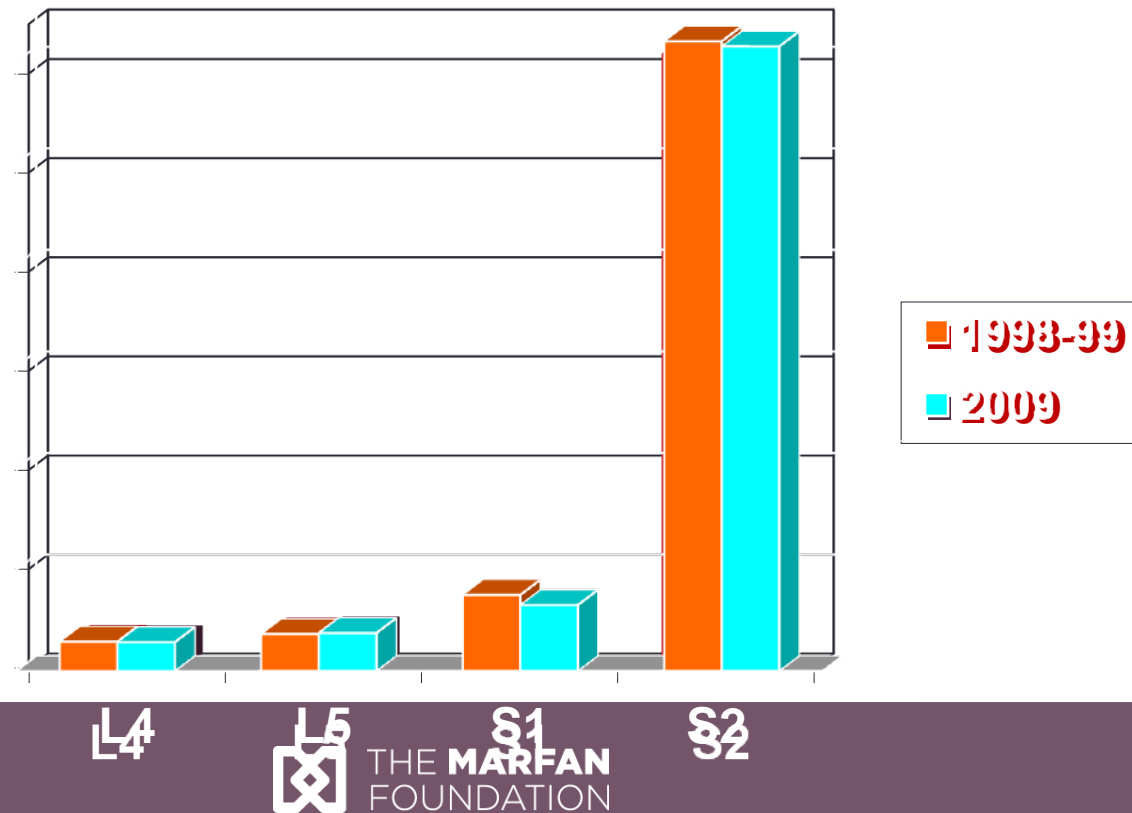


2009

# RESULTS

No statistically significant difference in:

- dural ectasia size noted after 11 yrs
- Symptoms (pain scores)



## IV. HIPS

### NEONATAL HIP DISLOCATION IN MARFAN

Incidence about 2 %

Pavlik Harness not successful

Surprisingly good response (3/4) to closed reduction and hip spica cast





# HIPS

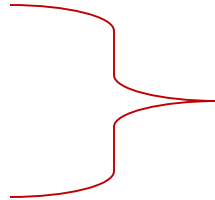
## “PROTRUSIO”

### Deep hip socket

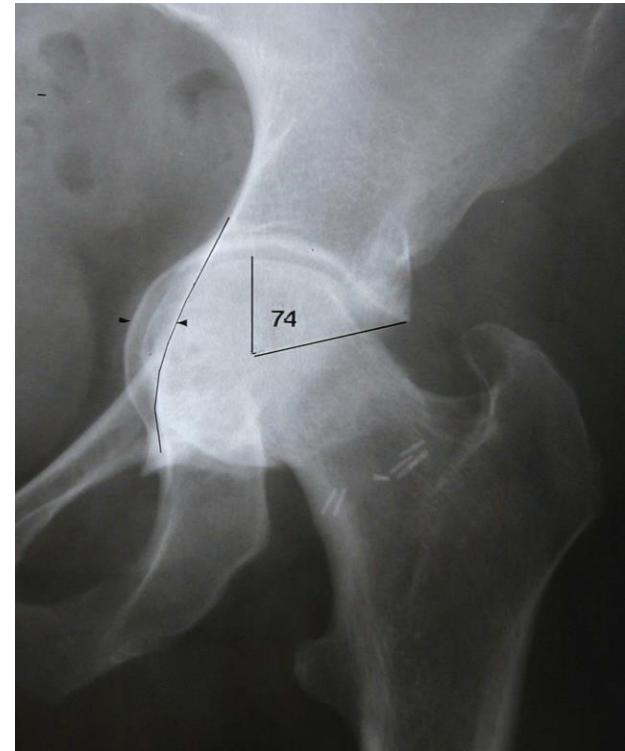
- Center-edge angle
- Medial displacement

### Bone more plastic under load

- With growth
- Mechanism:
  - Activation of osteoclasts?



Are there other reasons for hip degeneration?



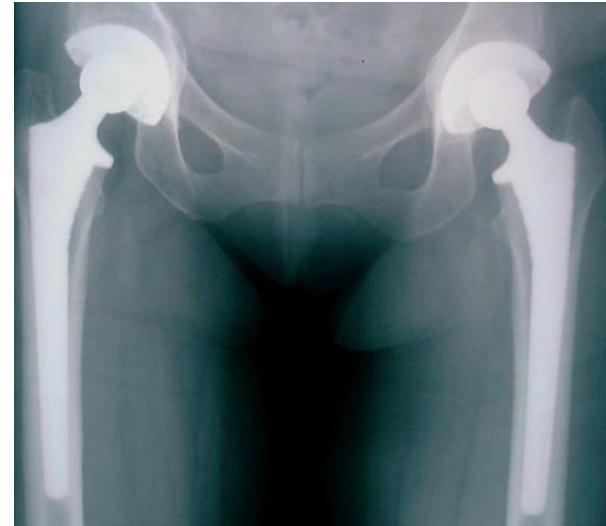
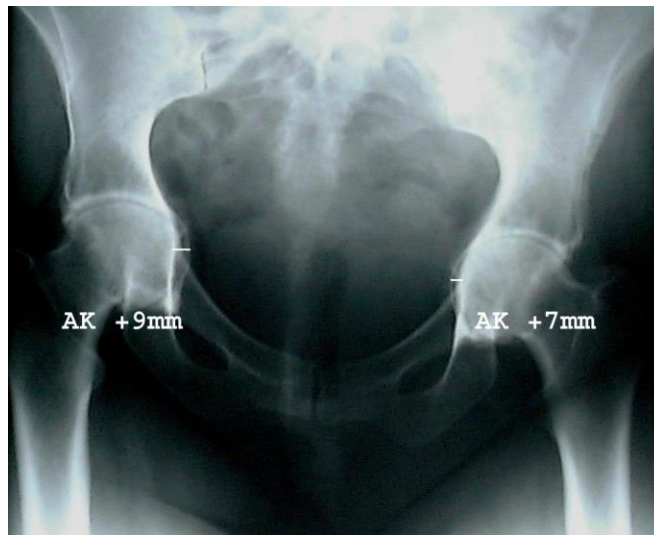
# RESULTS- PROTRUSIO STUDY

Pain only present in ~5% of 300 hips

- Role of prophylactic surgery seems limited

Few people needed hip replacement

Hip replacement has good results



# UNANSWERED QUESTIONS

Mechanism for osteopenia/osteoporosis

Pathogenesis of dysmorphic changes

Mechanism/treatment of

- Skeletal pain,
- fatigue,
- disability

