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Orthopedic care in 22q11.2DS syndrome

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There are no conflicts of interest

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Homans et al 2017

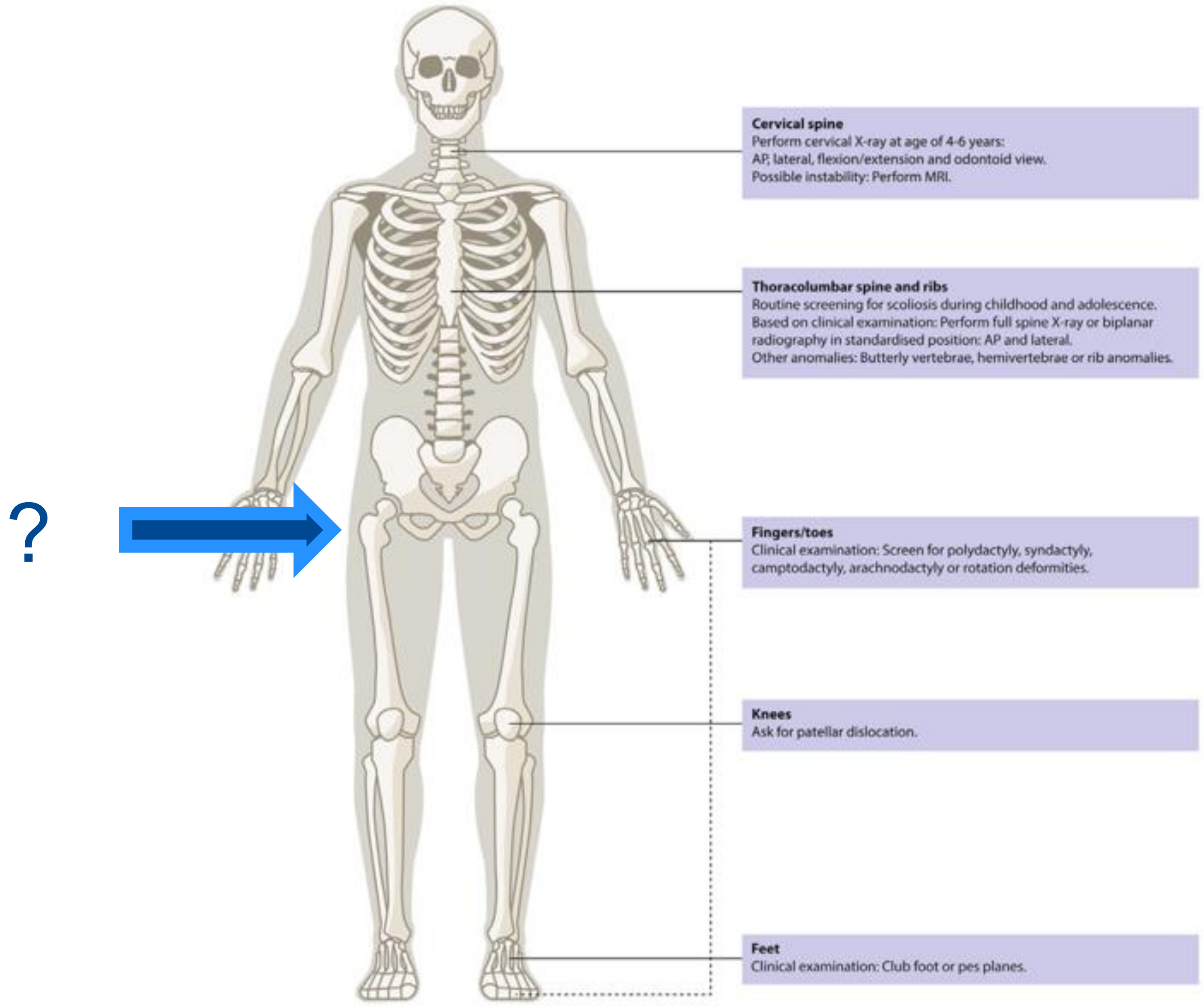
Review of literature

- 69 manuscripts (6055 Patienten), described 58 orthopaedic manifestations
- Prevalence of at least one cervical spine or occipital anomaly was 90 -100%
- Wide scoliosis prevalence in 14 studies (20-60%)
- Anomalies of lower extremities in 15-20%
    - Clubfoot: 1,1-13,3% prevalence
    - Patella dislocation: 10-20% prevalence
- Anomalies of upper extremities in 6%

Additional manifestations

- Developmental delay
  - Walking often starts > 18 months
- Hypermobility of joints
  - Development of foot deformities (flatfeet)
- Hypotonia
- Juvenile idiopathic arthritis
- Growth inhibition

Screening recommendations (Homans et al, 2017)



Scoliosis

- Idiopathic scoliosis
  - Prevalence: 2% in the population
- Neuromuscular scoliosis
- Congenital scoliosis
  - May occur in 22q11.2DS
- **Syndromic scoliosis**
  - Up to 60% in 22q11.2DS syndrome

- Thoracogenic scoliosis
  - After cardiac or thoracic surgery
  - Not unusual for 22q11.2DS syndrome

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22q11.2DS and scoliosis

- Prevalence 45 (20-60)%
- Brace often not effective (literature)
  - **My own experience is different**
- MRI for progressive curves to rule out intraspinal anomalies
- Surgical correction often necessary

22q11.2DS and scoliosis

> J Clin Med. 2021 Oct 20;10(21):4823. doi: 10.3390/jcm10214823.

22q11.2 Deletion Syndrome as a Human Model for Idiopathic Scoliosis

Steven de Reuver [1], Jelle F Homans [1], Tom P C Schlösser [1], Michiel L Houben [2], Vincent F X Deeney [3 4], Terrence B Crowley [5], Ralf Stücker [6 7], Saba Pasha [3 4], Moyo C Kruyt [1], Donna M McDonald-McGinn [4 5], René M Castelein [1]

Affiliations + expand

PMID: 34768342 PMCID: PMC8584329 DOI: 10.3390/jcm10214823

- Multicenter study
  - Curve progression in 54%
  - Curve progression rate 2,5° /year
  - Additional anomalies on MRI 10,5%

- Similar to idiopathic scoliosis

Most effective scoliosis treatment for children < 5 years

Casting for very young children (< 5 years)

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Casting for 3 months, than brace

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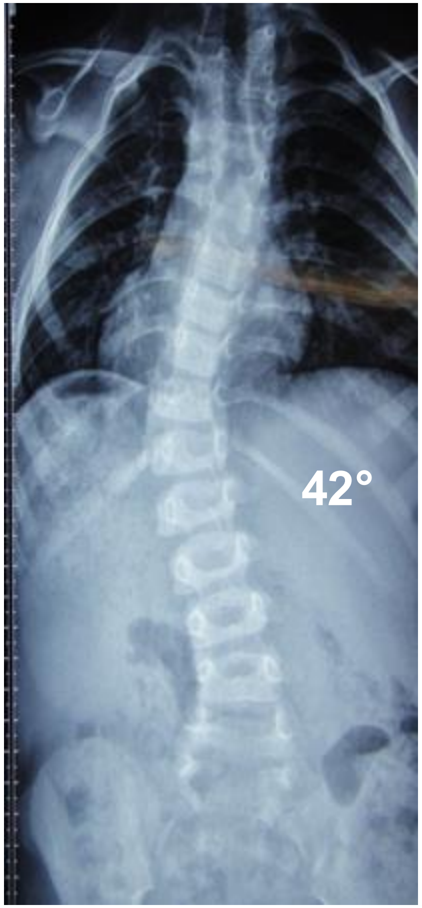
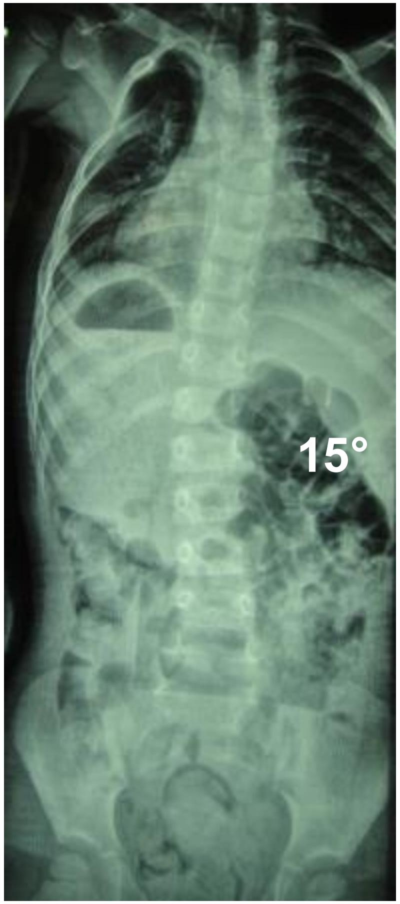
2,5 years old boy, result after 3 casts

At least a very efficient time buying strategy





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6 idiopathic, 10 syndromic patients

3x serial casting in 3 months

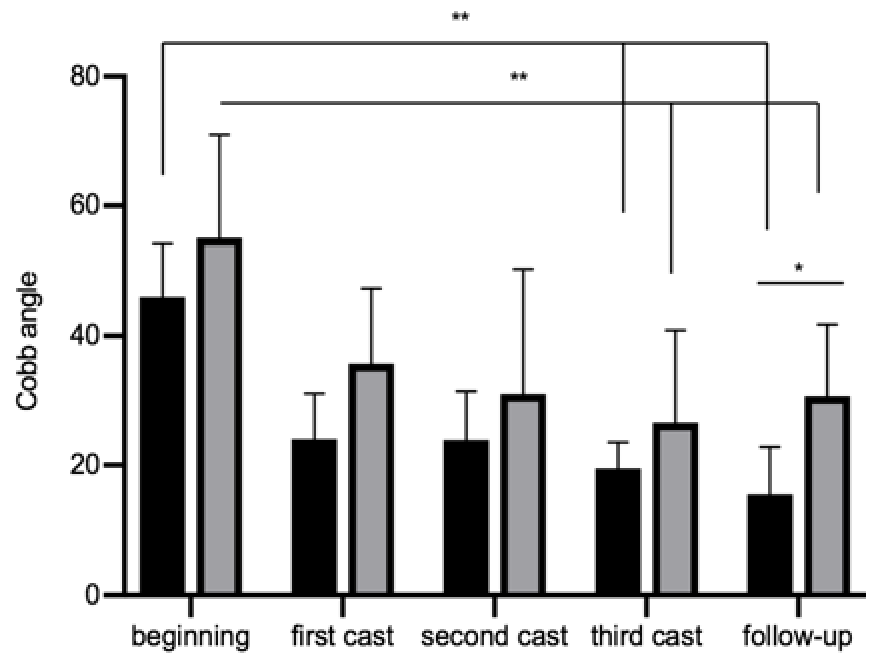
F/U: 21 months

Average age: 35 +/- 15 months

Average Cobb

IS: 46°

NIS: 55°



- idiopathic

- syndromic

RESEARCH ARTICLE | Open Access

Serial casting in early onset scoliosis: syndromic scoliosis is no contraindication

Check for updates

Tobias M. Ballhause[1,2*], Menard Moritz[1,3], Annika Hättich[2], Ralf Stücker[1,3] and Kiril Mladenov[1,3]

Casting as a time buying procedure very effective for syndromic and curative for idiopathic cases

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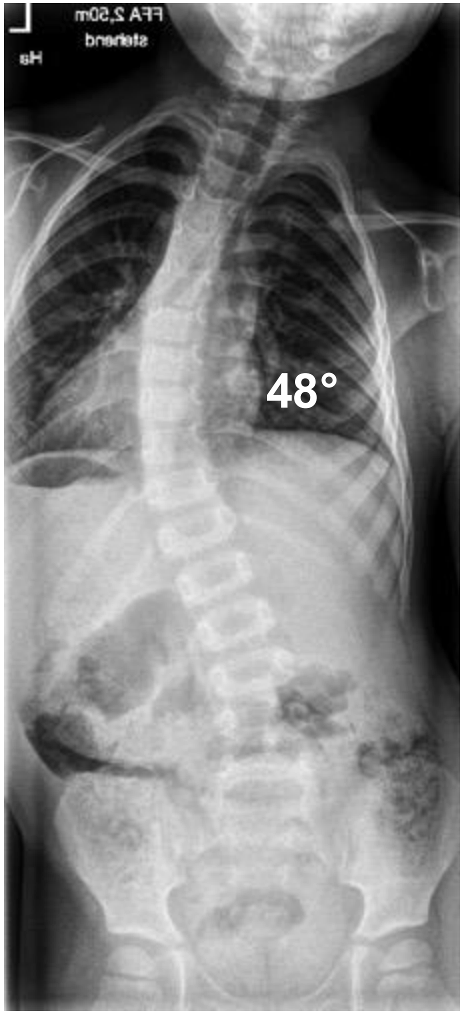
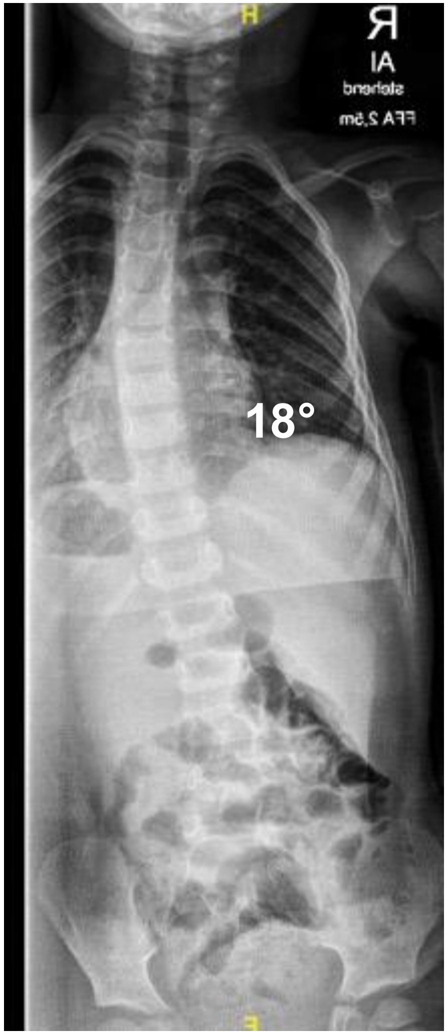
The importance to obtain MRI before treatment

5 year old boy with progressive scoliosis

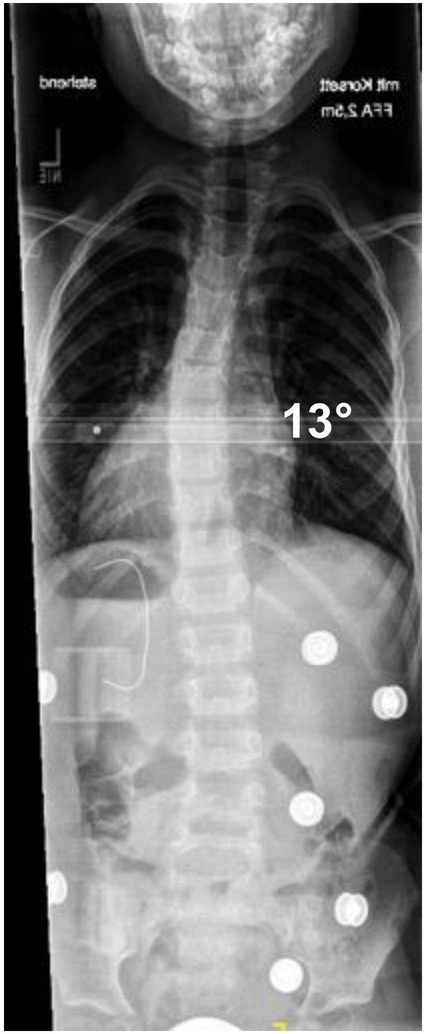


Cervicothoracic syringomyelia and Chiari 1-malformation

Decompression and casting performed



After 3. cast



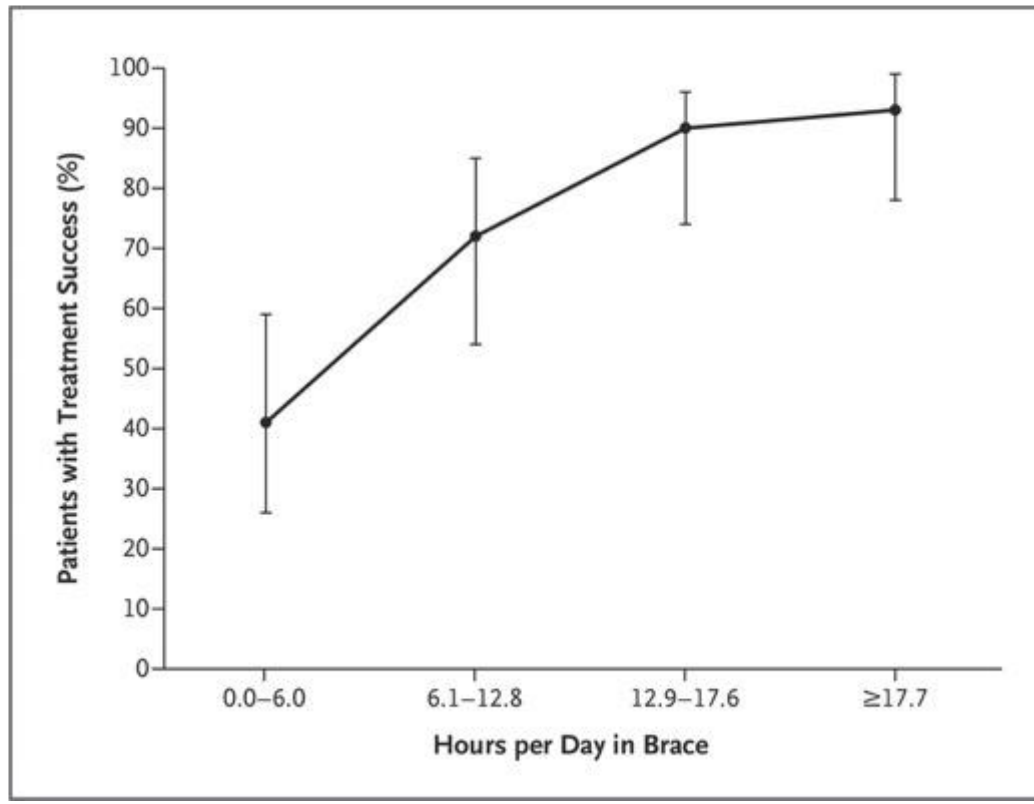
1 year f/u in brace

Scoliosis 5-11 years

- May try casting
- Brace treatment

  Standard 20-50°
- Growing rods
  - > 50-60°

Brace treatment not as effective as in idiopathic scoliosis
But should be tried for curves > 20°



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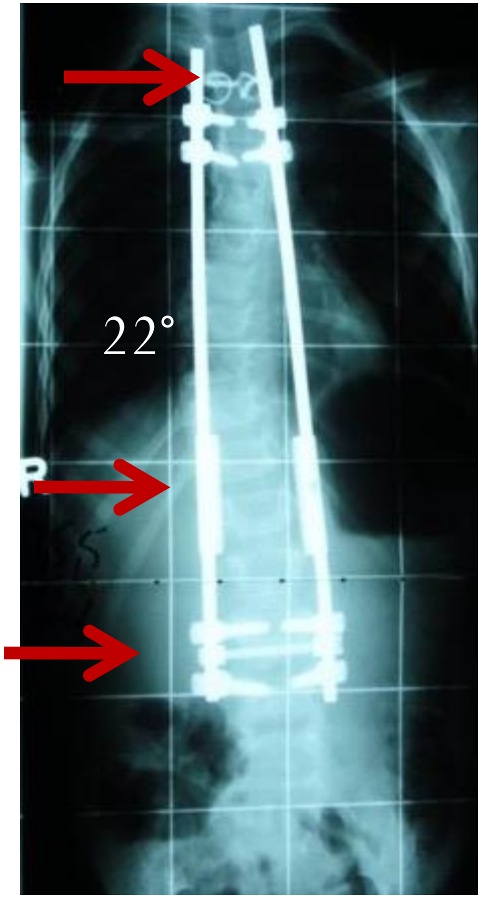
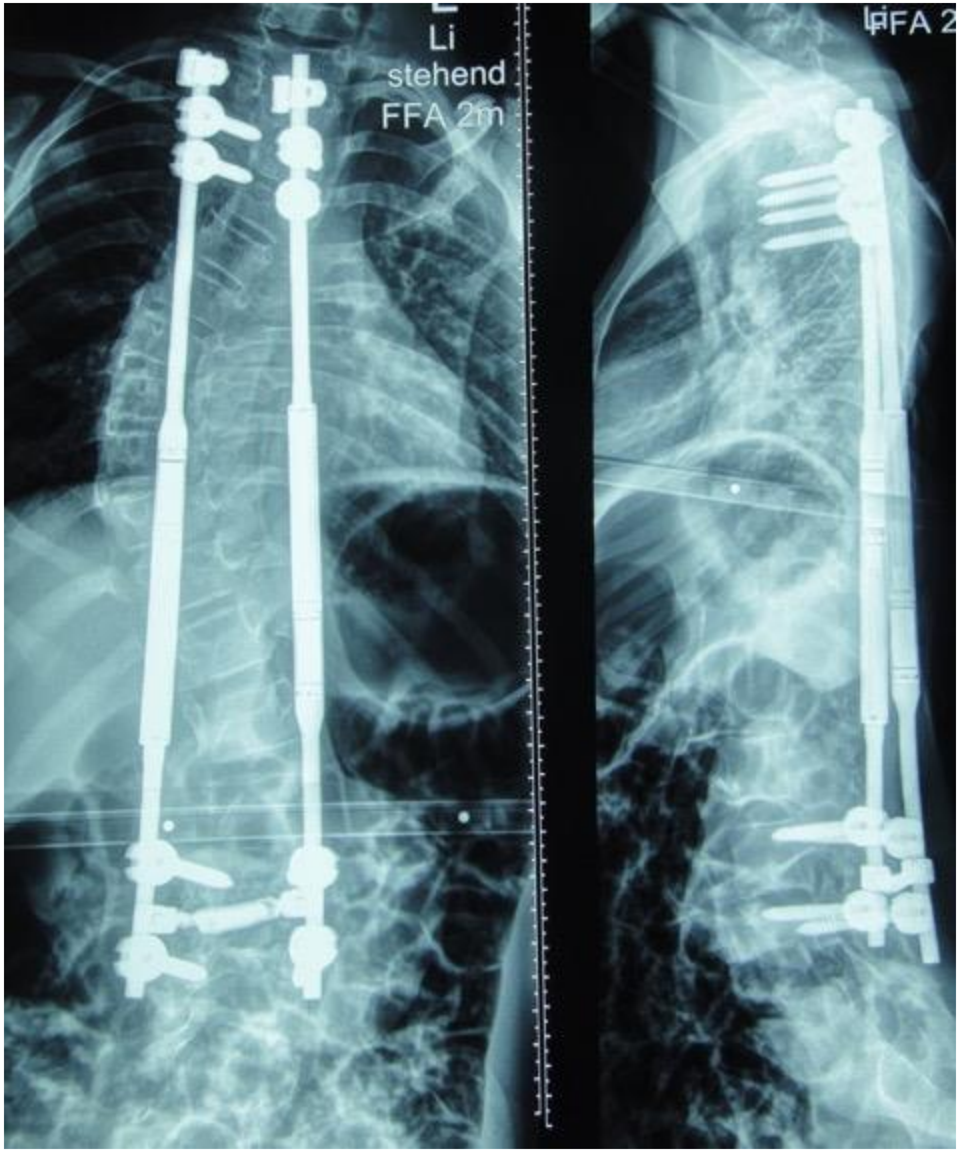
9 years old boy with scoliosis, treated by growing rods



Indication: Progressive curves > 50° and brace not effective

Magnetically controlled Growing Rods



Scoliosis > 11 years

- Brace (20-50°)
- Correction without fusion
  (VBT= vertebral body tethering)
- Final fusion (for curves > 50°)

12 – year old girl with 22q11.2DS
Brace treatment

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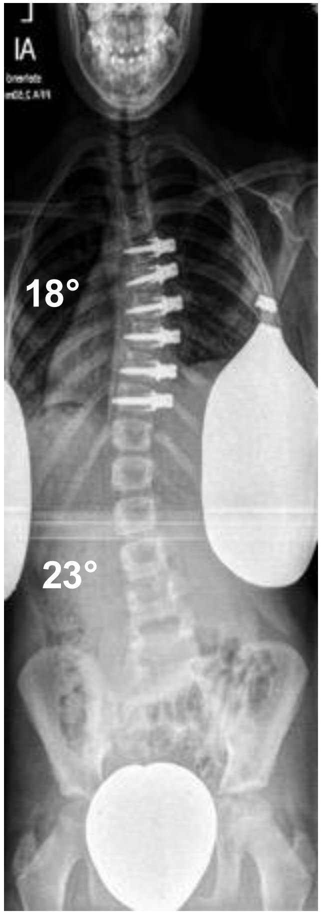
11+ 5 old girl

Progression of scoliosis in spite of brace

Sanders II



1 year F/U

11+6 year old girl, brace not tolerated

Preop – 35°

Bending 17°

Sanders 2

6 months 13°

12 months 7°

14 year old boy with 22q11.2DS and lumbar scoliosis

Currently VBT is more applied in the lumbar spine,
especially to preserve mobiity

Vertebral body tethering (VBT)

- Less enthusiasm in recent years
- Tether breaks frequently
- Own series
  - 60% needed fusion
  - Risk factors: large curves and older age
- Current recommendation
  - Not after the onset of pubertal growth spurt
  - Not for large curves

J Clin Med. 2023 Jun 8;12(12):3933. doi: 10.3390/jcm12123933.

Anterior Vertebral Body Tethering for Skeletally Immature Patients with AIS: Indication for Spinal Fusion at Skeletal Maturity Is Not Obviated in 60% of Cases

Kiril V Mladenov [1, 2], Hans O Pinnschmidt [3], Ralf Stücker [1, 2]

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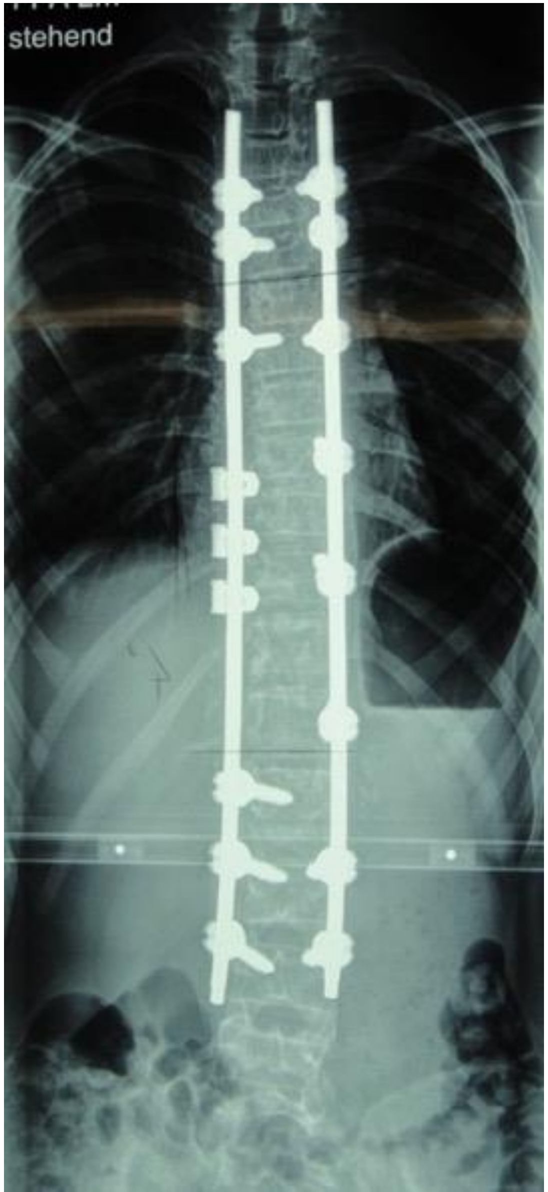
15 year old boy with 22q11.2DS scoliosis = 55°

Final fusion



The cervical spine in 22q11.2DS

- Prevalence of anomalies: 90-100%
  - Fusion C2/C3 - 34%
  - Instability - 56%
- However, few patients need surgery in childhood
- In case of instability school sport activities should be restricted
- First x-rays of cervical spine including flexion/extension views at 4-6 years
- MRI and CT may be necessary

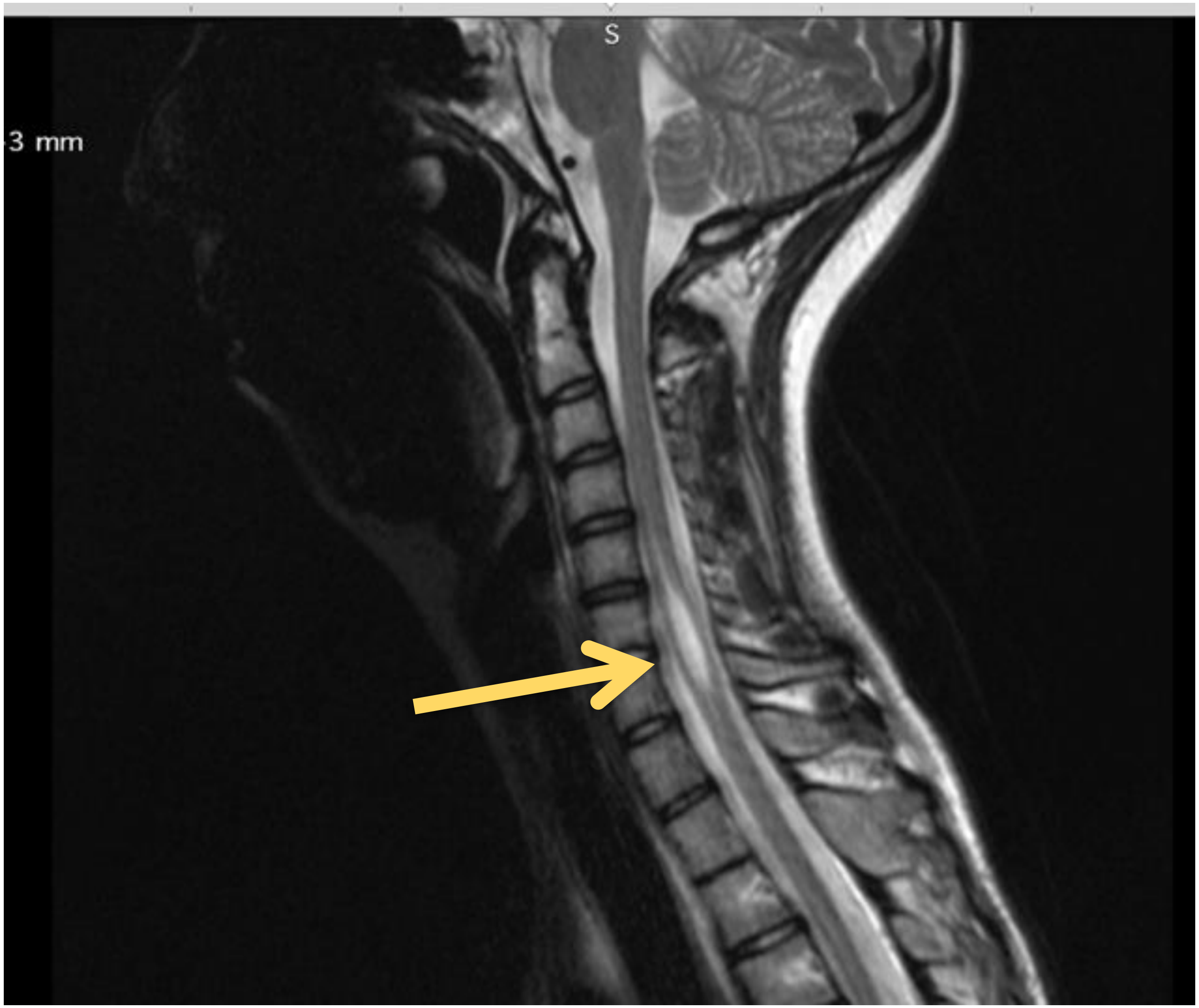
Instability of cervical spine in 22q11.2DS
Ricchetti 2004, n=79 patients

- Segmental instability 56%
- C0/C1 - instability 44%
- C1/C2 instabiliy 10%
- C3/C4 instability 15%
  - In patients with C2/C3 fusion
- Polysegmental instability 33%

Flexion/extension views show fusion C2/C3

Severe C1/C2 instability

Syrinx of spinal cord in a patient with 22q11.2DS



Before any type of surgery with anaesthesia x-rays of the cervical spine should obtained

Important for the anaesthesiologist

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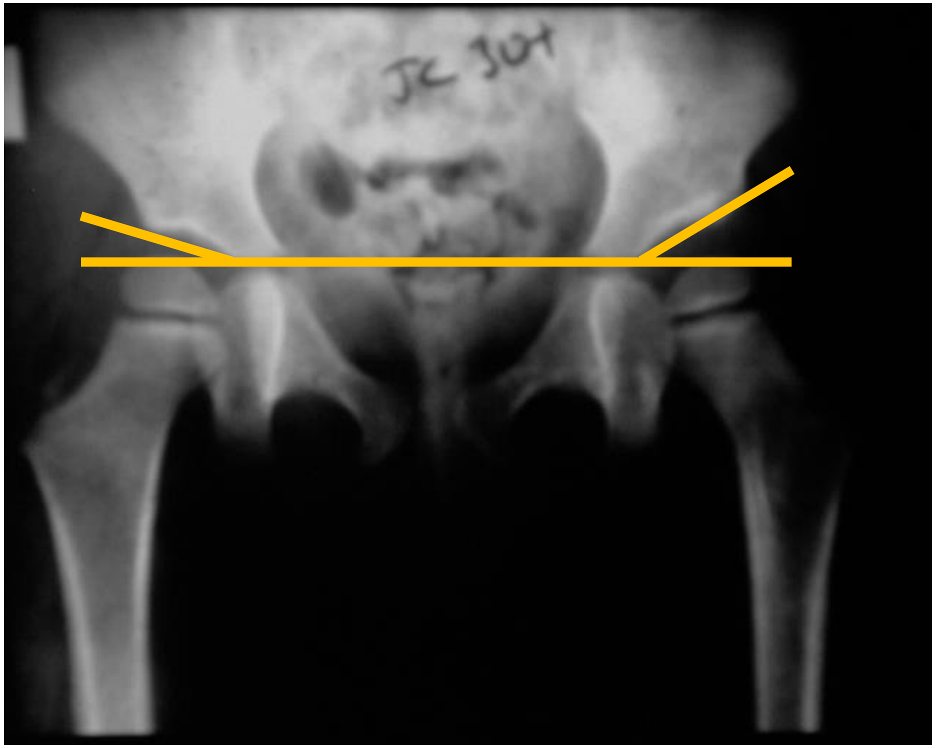
Hip problems in 22q11.2DS

- Little is known about the prevalence of hip problems
- Our own experience is, that hip problems are more common than previously reported, especially
  - Hip dysplasia
    - Developmetal delay, late beginn of walking
  - Acetabular protrusion (deep socket)
    - Restricted flexion and rotation of hip joint

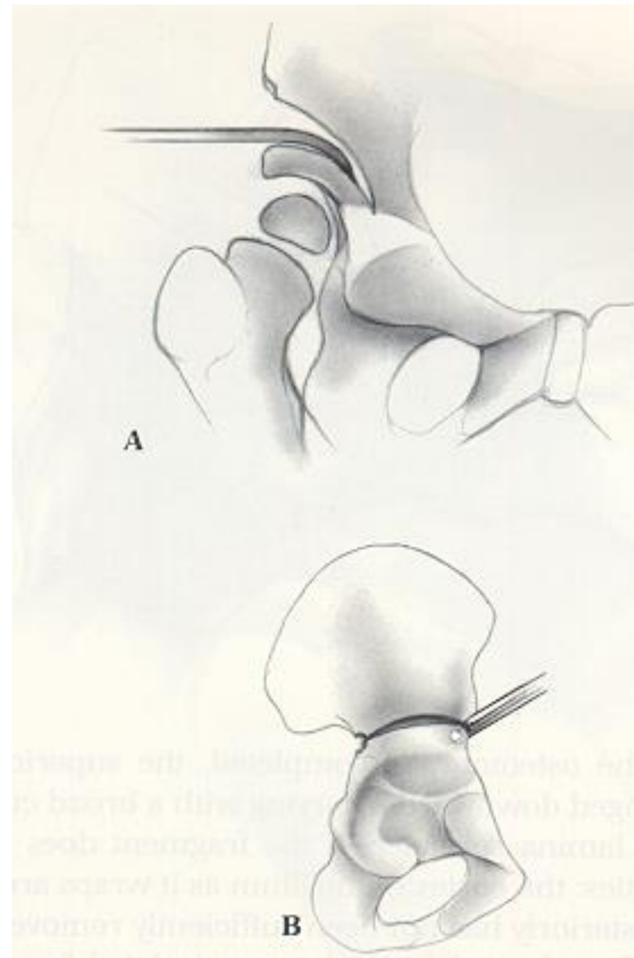
Hip dysplasia can lead to hip dislocation



Hip dyplasia left side

Hip dislocation left side

Treatment is always surgical



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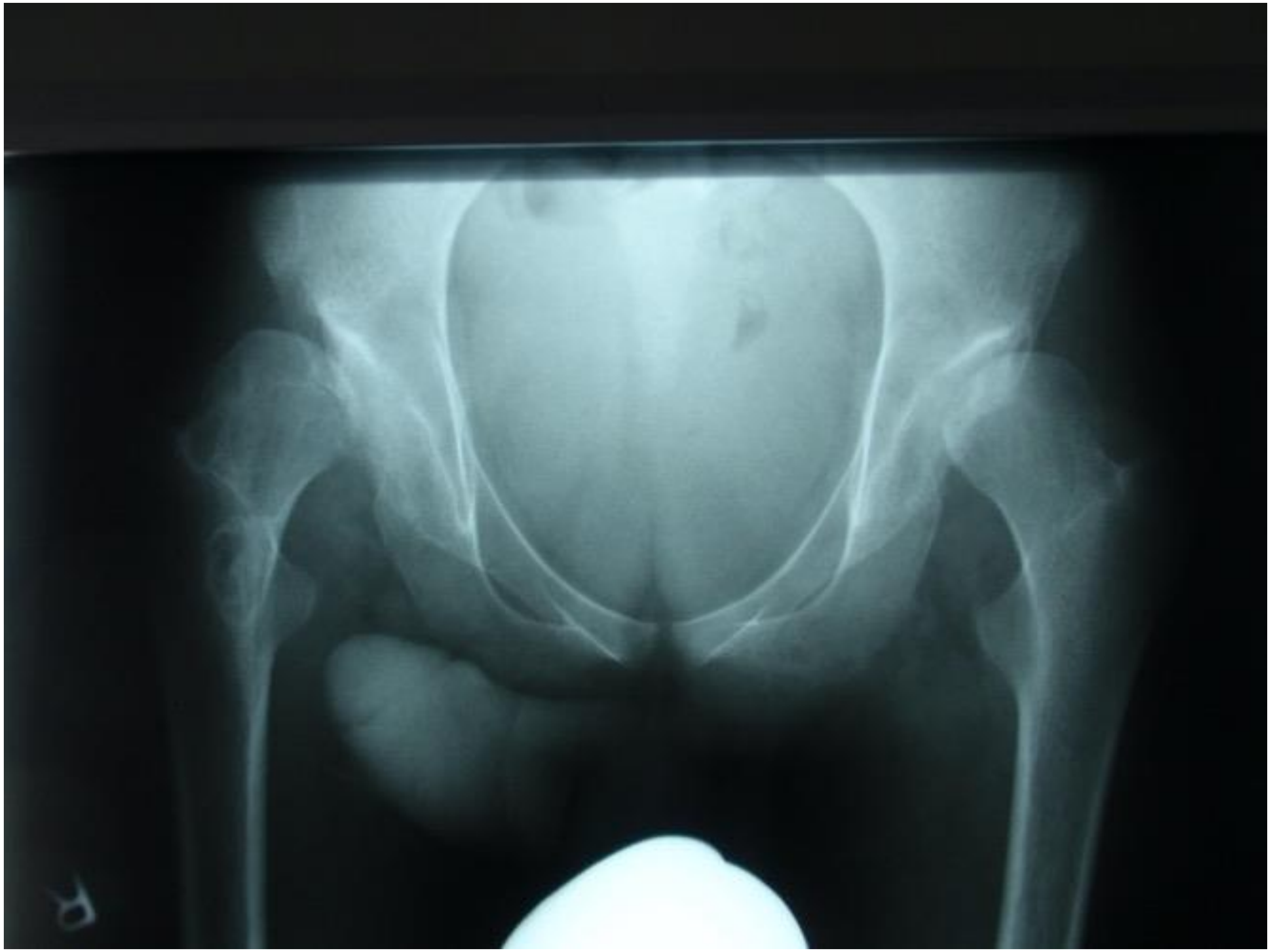
Severe hip dysplasia in a 17 year old girl with 22q11.2DS



Own experience – deep socket

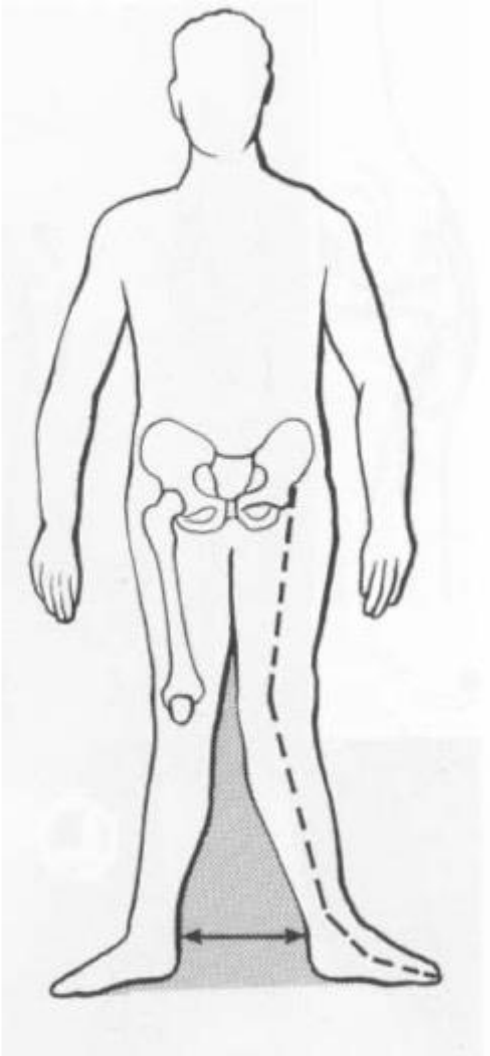
22q11.2DS patients have a significant incidence of acetabular protrusion (deep socket), flexion and rotation is usually reduced

Problems of the knee in 22q11.2DS

- Genu valgum (knock knee deformity)
  - Quite common
  - Prevalence not reported

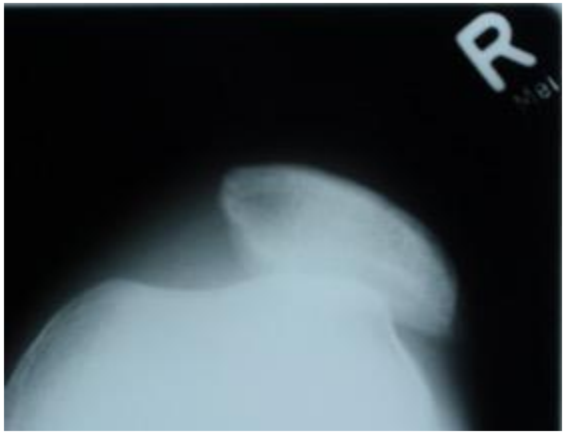
- Patellar dislocation
  - Prevalence – gen. population - 3%
  - Prevalence - 22q11DS – 10-20%

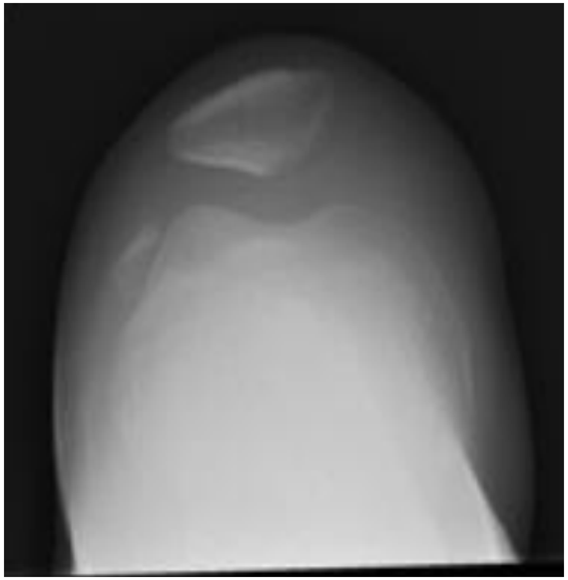
  - Can also occur without knock knees

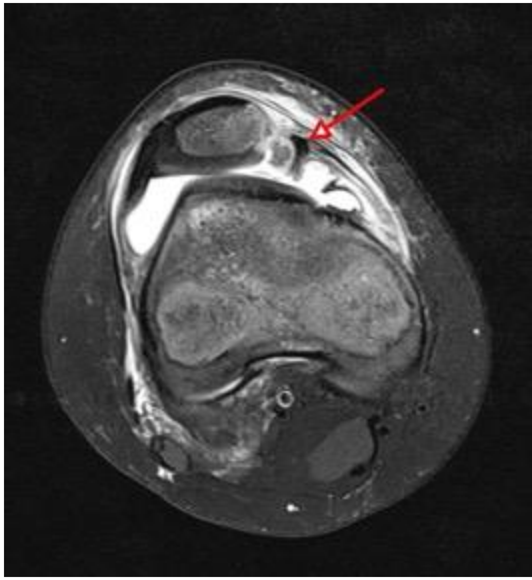


Patellar dislocation

- Associated with trauma
- Without trauma
- Cartilage lesions !!!
- In22q11.2DS often associated with genu valgum without trauma







Patellar dislocation in DS 22q11 patients

- Is common in association with genu valgum
- Other risk factors include
  - Patella alta – Patella is too high
  - Rotational deformities of lower extremities
    - Increased torsion of thigh or lower leg

- Does not respond to conservative treatment
- Early surgical treatment recommended at least after 2nd dislocation +/- correction of genu valgum

Guided growth at the knee

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Foot deformities in 22q11.2DS

- **Clubfoot: 1,1-13%; 0,1% in general population**
  - At birth
  - Ponseti treatment very effective
- **Flat foot**
  - Is seen frequently
  - Often associated with hypermobility of joints
  - Does not respond to conservative treatment
- **Hallux valgus**
  - Often associated with flat foot
  - Does not respond to conservative treatment

Flat foot deformity and treatment with insoles

Flatfoot is associated with hypermobility of joints and hypotonia

Insoles may be able to avoid progression of deformity but are not able to correct the deformity

Guided growth by arthrorisis

Before and after arthrorisis

Hallux valgus and mallet toes

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Guided growth for hallux valgus

Juvenile rheumatoid arthritis and del(22q11) syndrome: a non-random association

Verloes et al, 1998

These observations, and five other recently published cases, indicate that a JRA-like syndrome is a component of the del(22q11) spectrum

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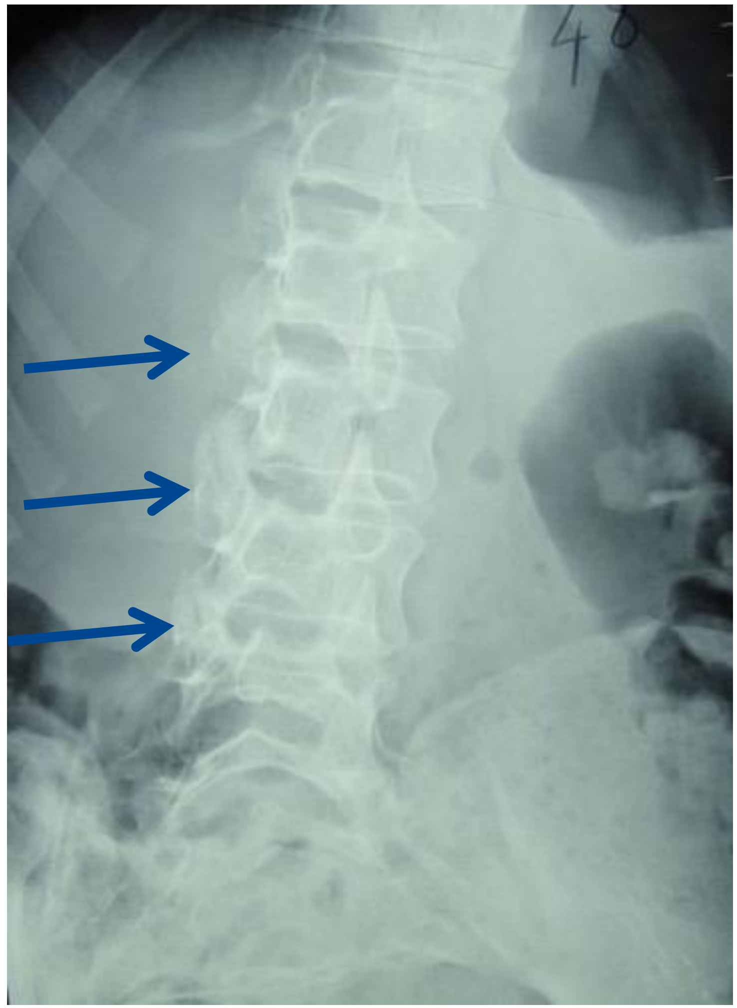
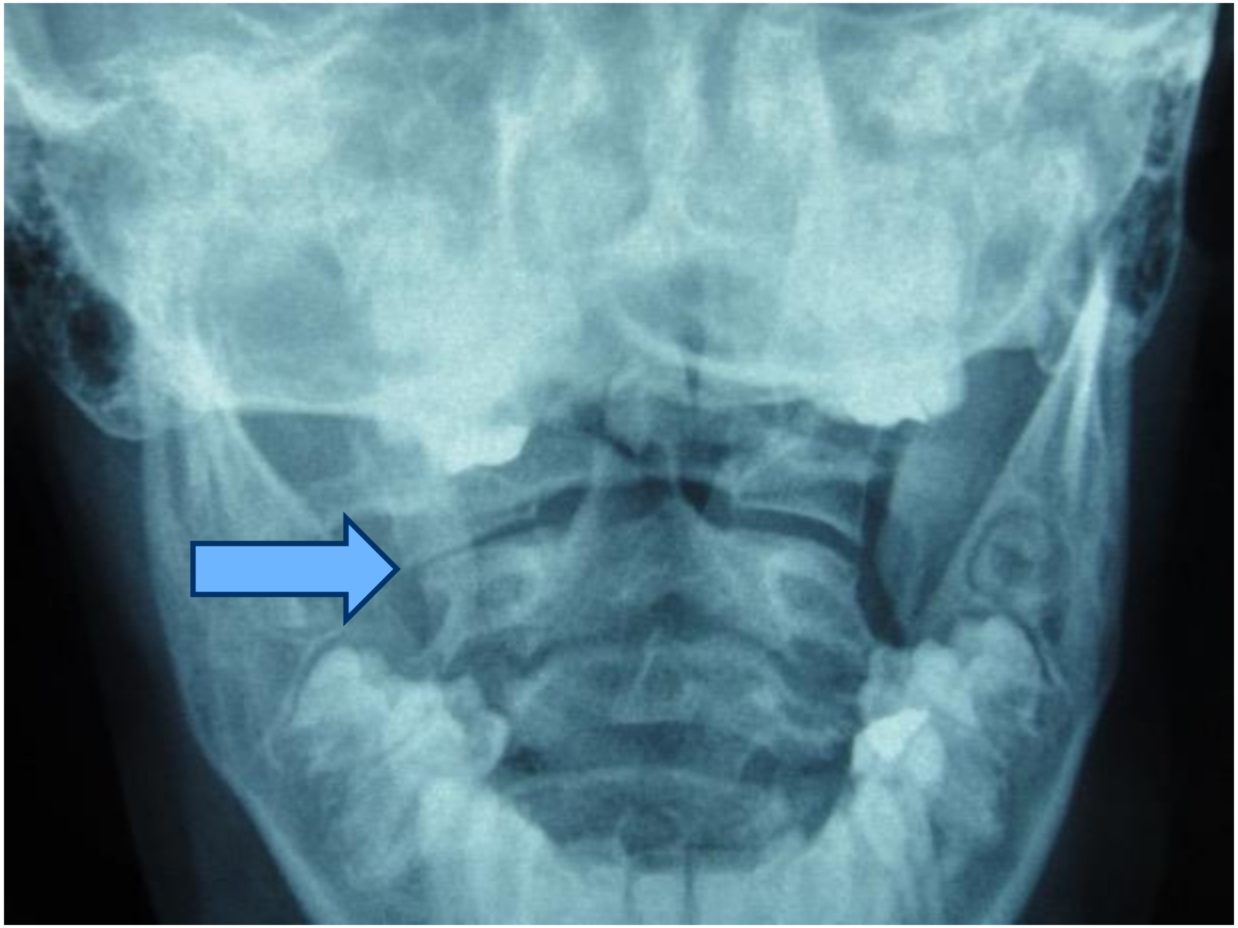
15 year old boy with 22q11.2DS and back pain and infamammatory disease of facet joints of the lumbar spine



11 year old girl with 22q11.2DS and restricted motion of the cervical spine



Own experience

- 10-20% of children with 22q11.2DS have signs of JIA
  - 3,75 % in the literature (Homans, 2018)
- All had not been diagnosed at presentation

Recommendations for orthopedic care for children with 22q11.2DS

AWMF German guidelines for 22q11.2DS (2025)

- Orthopedic evaluations should be performed yearly, especially spine and cervical spine and for patellar dislocations
- If scoliosis is suspected x-rays of the spine should be ordered
- First X-rays of cervical spine including fexion/extension views are recommended at age 4-6

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Summary

- Orthopedic problems in 22q11.2DS are common, especially cervical spine anomalies and scoliosis
  - In case of possible instability of the cervical spine, MRI is recommended
- Before any type of surgery and anesthesia the cervical spine has to be cleared by x-rays
- Some anomalies like knock-knees and flat feet can be surgically treated by guided growth
- Patella dislocation often needs surgery