

# Atlanto-Axial Instability in Patients with Down Syndrome

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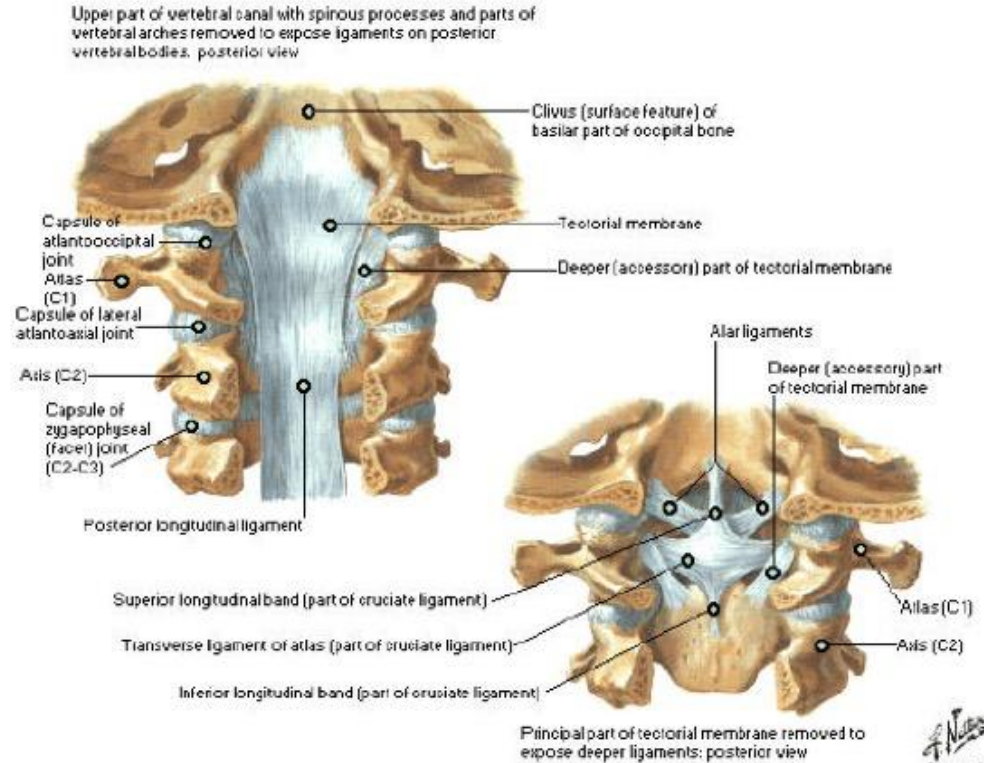
# Atlanto-Axial Instability: Anatomy

Excessive motion at C1/C2

Transverse ligament

Instability can lead to subluxation/dislocation

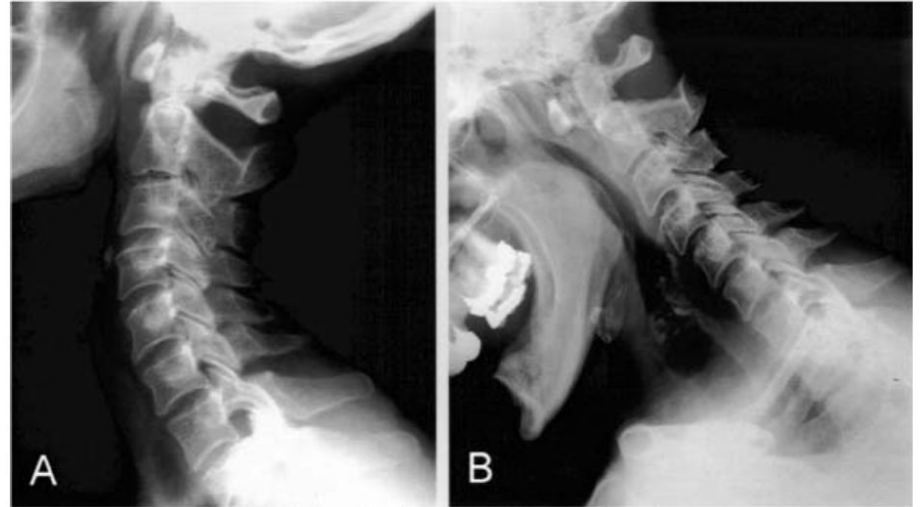
Cervical cord compression



# Atlanto-Axial Instability: Clinical Screen

Imaging (x-ray, CT, MRI)

- Lateral views of c-spine: flex, ext
- Abnormal atlantoaxial gap
  - >5mm children 8yo or younger
  - >3mm in adults
- Relatively poor reproducibility



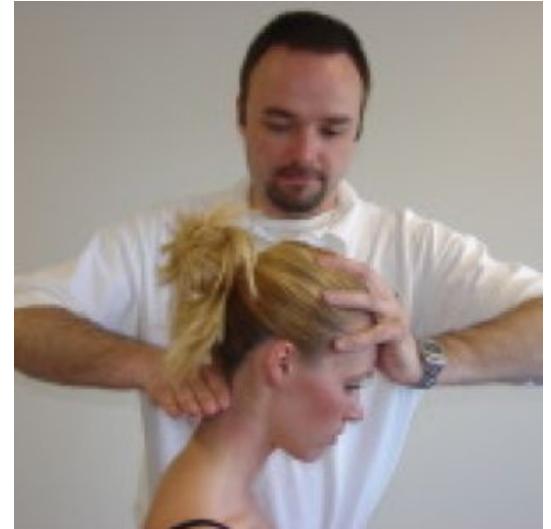
**Figure 3** - Dynamic lateral radiographs of the cervical spine (A- hiperextension; B- hiperflexion), showing atlantoaxial instability

# PT Screening Tools

## Upper Cervical Stability Test



Sharp Purser Test  
(NOT RECOMMENDED TO PERFORM!)



# Atlanto-Axial Instability: Signs and Symptoms

**Most AAI is asymptomatic!**

Be aware of populations at *increased risk* and *cervical cord compression* warning signs

Abnormal head posture

Restricted neck movement and/or pain

Sensation of head falling forward during neck flexion “Clunking” sensation during neck extension

# Atlanto-Axial Instability: Neurological Manifestations

## **Look for changes, esp after trauma**

Fatigue with walking

Abnormal gait

Regression from skills (esp  
ambulation/incontinence)

Neck pain and/or limited ROM

Torticollis or head tilt

Poor coordination, clumsiness

Sensory deficits

Spasticity

Hyperreflexia and clonus

Extensor plantar reflex

Deterioration in bladder/bowel

Upper motor neuron and posterior column s/sx

# Symptomatic AAI

Requires surgical stabilization of vertebrae

Stabilization until surgery

If allowed to progress can lead to paraplegia, hemiplegia, quadriplegia with respiratory failure and possibly death

Difficult to recognize due to low tone

# No AAI

Screen:

Starting age 3-5, every 10 years

Symptoms occur

Before surgery involving breathing tubes





# Asymptomatic AAI

Avoid high risk activities/extra stress/extreme ROM on cervical spine:

Gymnastics (tumbling)

Trampoline - especially under age 6

Diving

Swimming butterfly

High jump

Soccer

Collision Sports (football, hockey, etc)

More frequent medical and x-ray follow-up (debated)



# Atlanto-Axial Instability and Down Syndrome

## Individuals with Down Syndrome

- 13-14% have asymptomatic AAI
- 1-2% have symptomatic AAI

Screening (x-rays) may happen after age 2 (3-5, 12, and 18 years of age)

Before participation in sports or surgeries

Special Olympics

Uncertain whether asymptomatic AAI evolves into symptomatic AAI

- NDSS: more common to have abnormal → normal follow up x-rays than normal  
→ abnormal follow up

# To Screen or Not to Screen?

- Expensive
- Radiation
  
- 2 prospective studies of 95 and 141 individuals w Down Syndrome
  - 1 asymptomatic pt became symptomatic during at least 3y follow-up
- 41 reported cases of symptomatic AAI
  - 1 pt had radiographs before symptoms began (inconclusive)
  - 6y.o. X-ray was normal; symptomatic AAI after tumbling accident at age 18

As of 2011, the American Academy  
of Pediatrics no longer  
recommends routine x-rays for  
patients with Down Syndrome

# Summary

Symptomatic AAI is rare, but out there!

Asymptomatic AAI not necessarily higher risk for symptomatic AAI

Kids with DS are at higher risk for AAI - know s/sx

Advocate for our patients!

# Discussion

Screening tools aren't necessarily effective

Children may not be screened

Keep an eye out for s/sx

Most dislocations: everyday activity, med treatment, sport

Extra precautions for children with Down Syndrome?

Falls, bouncing, extreme neck ROM, spinal manipulations, trampoline?

Posture

3 y.o. with Down syndrome s/p femur fx

Thank you!

*Questions?*

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