Basic Medical Surveillance Essentials for people with Down Syndrome

Cervical Spine Disorders: Craniovertebral Instability
Revised 2024

One of a set of clinical guidance drawn up by the Down Syndrome Medical Interest Group (DSMIG UK)

Background

People with Down syndrome are at risk of acute or chronic neurological problems caused by cervical spine disorders. These may present at any age. In childhood, excessive movement between the bones of the craniovertebral junction (CVJ), also known as craniocervical junction (CCJ), is the predominant issue[1-4] and the prime remit of this guideline.
The risk of other problems increases with age as chronic spinal cord compression due to premature degenerative changes becomes an additional underlying mechanism.[5-8]

The CVJ comprises the complex set of joints, muscles and ligaments that allow for articulation between the skull and the upper cervical spine. In Down syndrome, excessive movement at the CVJ can occur due to a combination of ligamentous laxity, hypotonia and/or bone anomalies.[8]

In Down syndrome it is movement between the atlas and axis that is predominantly affected, referred to as atlantoaxial instability (AAI) or atlantoaxial subluxation (Fig 1).[1] The joint between the occiput and the atlas can also be involved (occipito-atlantal instability) but this rarely occurs in isolation and can be more difficult to diagnose. It is considered more correct to use the term craniovertebral instability to encompass movement at both joints rather than atlantoaxial instability alone.

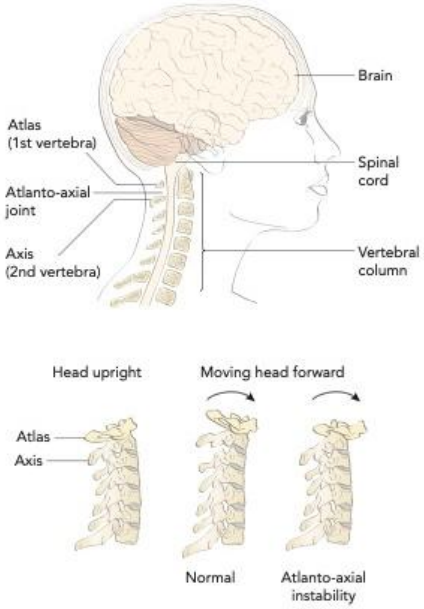

Figure 1: Atlantoaxial Instability

Prevalence and natural history

As many as one third of children with Down syndrome have 'hypermobility' exhibiting a range of movement that is greater than the normal range, however the majority of such patients are asymptomatic.[4,9,10] The term 'instability' is generally reserved for instances when the range of movement is sufficient to pose a potential, or actual, risk to the spinal cord. The incidence of clinically significant/symptomatic atlantoaxial instability is approximately 2%.[4,9,11,12] The natural history of atlantoaxial motion in Down syndrome is unclear, however older studies suggest that the natural history is favourable; radiological parameters of movement more commonly improve than deteriorate with age. A recent study suggested that the risk of progressive instability is less than 2% over 4 years.[13]

Radiological assessment of craniovertebral instability

(i) Radiographs (X rays)
Lateral neutral cervical spine X rays can be used to demonstrate the alignment of the bones of the CVJ (Fig 2).

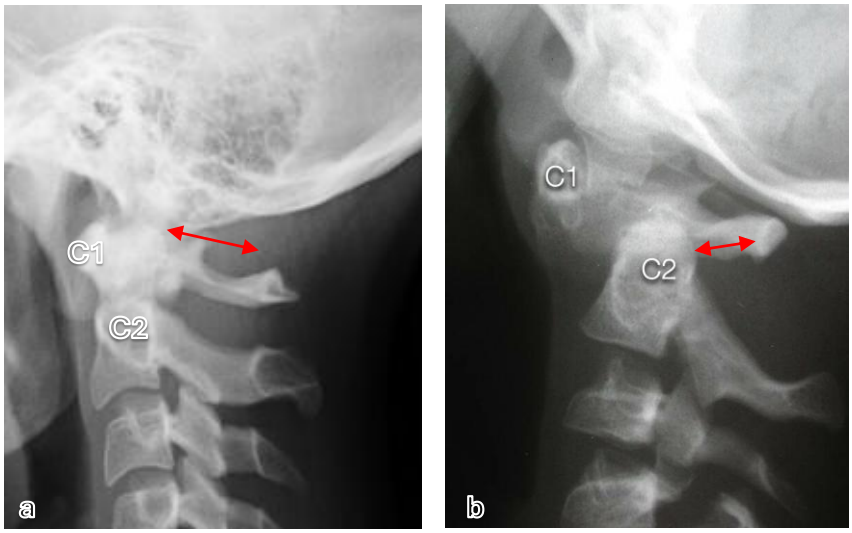

Figure 2: Cervical Spine Plain X rays (a) Normal alignment (b) Atlantoaxial subluxation. Arrow indicates space available for the spinal cord (SAC), which is reduced in the presence of atlantoaxial subluxation.

A single, lateral cervical spine X ray is an appropriate first investigation in the evaluation of a person with Down syndrome who presents with warning signs (see Box 1).[9] Dynamic X rays performed in the flexed and extended positions are used to better define the range of cervical spine motion. Atlantoaxial subluxation, if present, will be accentuated in the flexed position (Fig 3). Such dynamic imaging should not be performed in a sedated or anaesthetised patient.

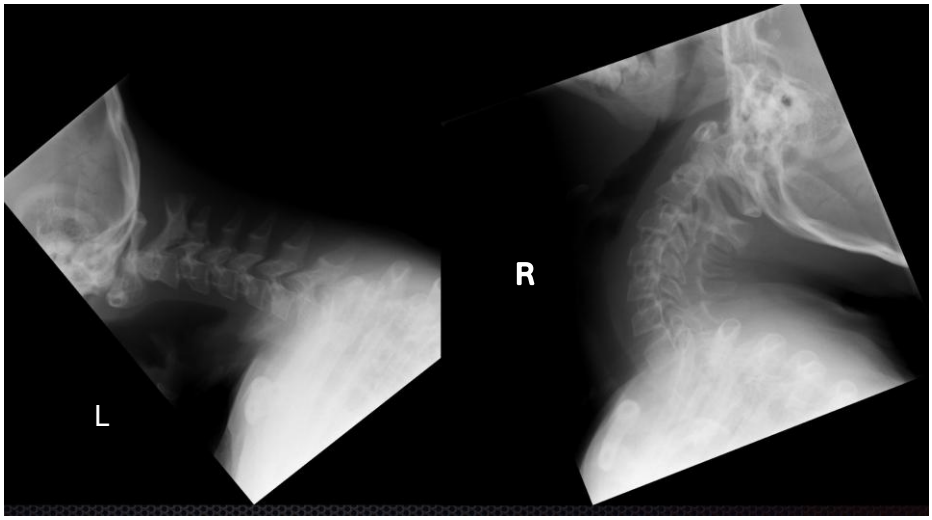

Fig 3: Lateral C spine in flexion (L) and extension (R). Atlantoaxial subluxation is present in the flexed position, but alignment is restored in extension

The correlation between X ray findings and clinical symptoms/signs is poor and so plain radiographs do not have adequate sensitivity and specificity to be used as the sole diagnostic tool for atlantoaxial instability. Furthermore, a normal cervical X ray does not rule out current or potential subluxation.[2,14] If clinical signs of myelopathy are present MRI imaging of the spine should be performed regardless of plain x-ray findings.

(ii) MRI
MRI of the cervical spine is used to assess the cervicomedullary junction and spinal cord. MRI is reserved for symptomatic cases, particularly where there is evidence of myelopathy.

Screening for CVJ instability
(i) Radiological Screening:
Since the natural history is incompletely understood and the correlation between radiological and clinical findings is poor, routine radiological screening for asymptomatic people with Down syndrome is not recommended by DSMIG, the American Academy of Paediatrics and others. [4,11,12,14-17]

(ii) Clinical Screening:
Published guidelines emphasise the importance of patient/carer awareness and the need for clinical vigilance. A careful history and examination should be performed at all annual health reviews/checks looking for 'warning signs', symptoms, and examination findings for CVJ instability and cervical myelopathy (see Box 1and 2) in patients with Down syndrome.[16,18] Parents should be advised to seek medical advice if their child develops symptoms in between health reviews.

Symptomatic Individuals
New or progressive symptoms that might be indicative of CVJ instability require prompt investigation and specialist neurosurgical/orthopaedic referral.
It is imperative that any person with Down syndrome presenting with new symptoms or signs that may be indicative of craniovertebral instability or myelopathy be examined and investigated expediently. [2,19]

Symptoms can be grouped into those relating to appearance, movement and functional impact (see Box 1).[4,16,17] There should be a low threshold for suspicion as there is good evidence that these early warning signs are often missed and the diagnosis of spinal cord injury made late with otherwise preventable catastrophic consequences.

It is essential that parents, relatives, carers and all healthcare professionals are made aware of these clinical signs and symptoms and that patients with Down syndrome are examined (see Box 2) and consulted for their presence at all annual health reviews.

Box 1: Warning Signs/Symptoms

1) Appearance:
   - Abnormal head posture
   - Torticollis (Wry neck)
2) Movement:
   - Neck pain
   - Reduced neck movements
3) Functional
   - Deterioration of gait and/or frequent falls
   - New onset or worsening fatigability on walking,
   - Regression in manipulative skills
   - Limb weakness
   - New onset Bulbar dysfunction (respiratory insufficiency, sleep apnoea, swallowing difficulties)
   - Change in bowel or bladder function

It is important to recognise that some of the symptoms and signs of CVJ instability may overlap with other pathologies seen in Down syndrome, such as feeding problems, obstructive sleep apnoea or Alzheimer disease in adulthood. Similarly, history taking may be more challenging due to the high incidence of intellectual disability seen in patients with Down syndrome.

When any of these clinical findings are present, plain cervical radiographs in the neutral position should be obtained. If significant abnormalities are identified the patient should be referred promptly to a specialist centre. If no significant radiographic abnormalities are present, additional flexion-extension radiographs may be obtained but only in collaboration with the specialist centre.[16]

Box 2: Clinical Examination

1) Neck position
   - Torticollis
   - Reduced range of movement
2) Neurological examination
   - Increased tone
   - Clonus
   - Pyramidal pattern of limb weakness
   - Brisk reflexes
   - Extensor plantars
   - Gait unsteadiness
   - Loss of joint position sense

Treatment of CVJ instability

Treatment of symptomatic cases will typically entail instrumented fusion between the atlas and the axis, or between the occiput and the atlas. Using current surgical techniques increasingly good outcomes are reported where timely intervention is performed in experienced centres. [20,21]

Sport

Asymptomatic individuals with Down syndrome should not be barred from normal sporting activities because there is no evidence that participation in sports increases the risk of cervical spine injury any more than for the general population. [17,22,23] Clinical guidelines from the Journal of the American Medical Association in 2020 advised: *"while avoiding potential SCI is important, restricting asymptomatic individuals with AAI from participating in physical activities is also undesirable for reasons related to physical and psychological health. Additional indirect evidence has suggested that SCI from AAI is uncommon. A 1995 review*

from the American Academy of Pediatrics Committee on Sports Medicine noted only 41 well-documented, published cases of symptomatic AAI in adults with Down syndrome. In addition, Special Olympics organizers report no spinal cord injuries from more than 50 000 individuals with Down syndrome who participated in Special Olympics activities over 20 years."[17]

For specialised sport, such as gymnastics, children with Down syndrome should not be automatically excluded. Currently, national governing bodies for certain sports have a clinical screening protocol which should be observed (www.british-gymnastics.org-Atlanto-Axial Information Pack).

The Special Olympics no longer requires radiographic screening of athletes with Down syndrome.[24]

Counselling of individuals with Down syndrome and a shared decision-making approach that considers the benefits and potential harms of participation in high risk activities including, but not limited to, contact sports, gymnastics, horse riding, trampolining, skiing and diving is encouraged. [16,17]

Anaesthesia
Very few cases of perioperative cervical spine injury in patients with Down syndrome have been reported in the literature.[25] This complication is therefore considered very rare.

- Prior to general anaesthesia, a careful history and examination should be undertaken with reference to the above warning signs.
- Evaluation of the patient's comfortable range of movement can be carried out at the pre-operative assessment.
- Routine pre-operative radiography is not recommended in the absence of clinical concerns. [26-28]
- Universal cervical precautions are recommended during anaesthesia, including the use of video laryngoscopy, if available. [25]
- Flexion of the cervical spine should be avoided at induction or whilst under anaesthesia.

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