Basic Medical Surveillance Essentials for people with Down syndrome

CERVICAL SPINE DISORDERS:
CRANIOVERTEBRAL INSTABILITY

REVISED 2012

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

Background

People with Down syndrome are at risk for acute or chronic neurological problems caused by cervical spine disorders. These may present at any age. In childhood the incidence is low and craniovertebral instability is the predominant issue\(^1\)\(^2\)\(^3\) 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\(^4\)\(^7\).

The craniovertebral junction comprises the complex set of joints, muscles and ligaments that allow for articulation between the skull and the upper cervical spine. Hypotonia and ligamentous laxity in Down Syndrome may result in excessive movement at the craniovertebral junction both between the atlas and the axis (atlantoaxial subluxation, see diagram) and between the occiput and the atlas (occipito-atlantal subluxation)\(^1\). It is thus more correct to use the term craniovertebral instability to cover both joints rather than atlantoaxial instability.

Depending on the study 10 – 27% of all individuals with Down syndrome have radiological evidence of increased movement at the craniovertebral junction however in only a very small proportion of cases is this associated with clinical symptoms\(^1\)\(^8\)\(^-\)\(^11\). Furthermore a normal cervical spine X ray does not preclude the subsequent development of problems related to craniovertebral instability\(^2\)\(^8\).

In a very small proportion of cases
craniovertebral instability is associated with clinical symptoms and surgical fusion of involved levels is indicated. Using current surgical techniques increasingly good outcomes are being reported where timely intervention is performed in experienced centres\textsuperscript{12, 13}.

**Asymptomatic individuals**

*a) X Rays*
Cervical spine X rays are unreliable and in asymptomatic children have no proven predictive validity for subsequent acute dislocation/subluxation at the atlantoaxial or occipitoatlantal joints therefore on the basis of current evidence\textsuperscript{8-11} routine radiological screening for asymptomatic people with Down syndrome is not recommended.

*b) 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\textsuperscript{14, 15}. For specialised sport, such as gymnastics, children with Down syndrome should not be automatically excluded but the requirements of national governing bodies which include a clinical screening protocol should be observed. (www.british-gymnastics.org- Atlanto-Axial Information Pack)

**Symptomatic individuals**

*a) Warning Signs*
It is imperative that any person with Down syndrome presenting with new symptoms or signs (see below) that may be indicative of craniovertebral instability or myelopathy be examined and investigated expediently\textsuperscript{2, 16}. There should be a low threshold for suspicion as there is good evidence that these early warning signs are often missed and diagnosis of CSI 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.

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<th>Warning Signs</th>
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<td>• Neck pain,</td>
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<td>• Abnormal head posture,</td>
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<td>• Torticollis, (Wry neck)</td>
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<td>• Reduced neck movements,</td>
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<td>• Deterioration of gait and/or frequent falls</td>
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<td>• Increasing fatigability on walking,</td>
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<td>• Deterioration of manipulative skills,</td>
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It is important to recognise that the above symptomatology in adult life may be falsely attributed to Alzheimer disease or other progressive cerebral deterioration.
b) Management
In the presence of any of the above warning signs a clinical history, physical and neurological examination must be carried out.

Unless this reveals a good alternative explanation for symptoms, and provided the person has a mobile neck and can extend their neck to look up to the ceiling and flex to look to the floor, good quality flexion and extension cervical spine X-rays should be taken.

Following this, if either clinical or radiological abnormality is found expedient referral to a specialist centre is indicated.

Anaesthesia

Prior to general anaesthesia a careful history and examination should be undertaken with reference to the above warning signs. Routine pre-operative radiography is not recommended in the absence of clinical concerns17-19.

References


Steering group for CSI guideline revision

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