Basic Medical Surveillance Essentials for people with Down syndrome

CERVICAL SPINE DISORDERS: CRANIOVERTEBRAL INSTABILITY

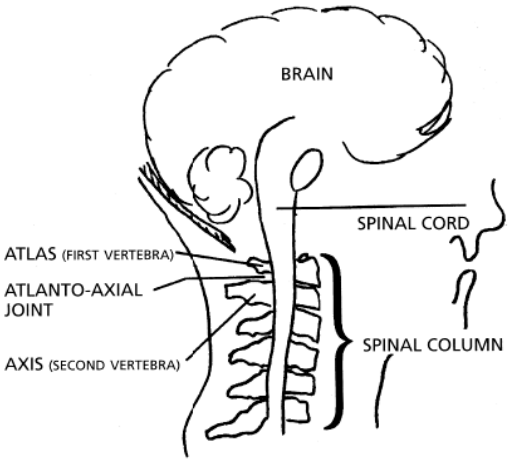
REVISED 2012

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

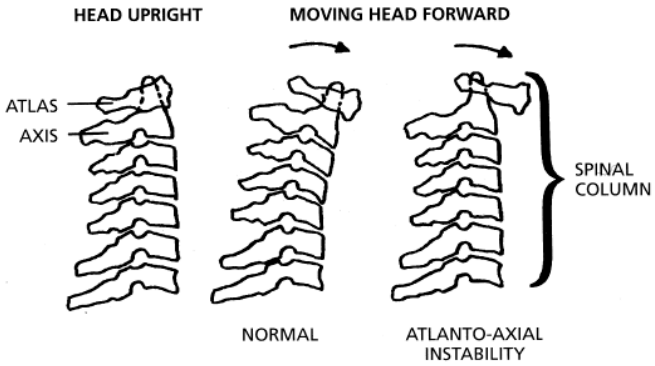
Background

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[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[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[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[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.

Warning Signs

- Neck pain,
- Abnormal head posture,
- Torticollis, (Wry neck)
- Reduced neck movements,
- Deterioration of gait and/or frequent falls
- Increasing fatigability on walking,
- Deterioration of manipulative skills,

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 concerns[17-19].

References

1. Brockmeyer D. Down syndrome and craniovertebral instability. Topic review and treatment recommendations. *Pediatr Neurosurg* 1999;**31**:71-7.

2. Davidson RG. Atlantoaxial instability in individuals with Down syndrome: a fresh look at the evidence. *Pediatrics* 1988;**81**:857-65.

3. Atlantoaxial instability in Down syndrome: subject review. American Academy of Pediatrics Committee on Sports Medicine and Fitness. *Pediatrics* 1995;**96**:151-4.

4. Van Dyke DC.Gahagan CA. Down syndrome. Cervical spine abnormalities and problems. *Clin Pediatr (Phila)* 1988;**27**:415-8.

5. Fidone GS. Degenerative cervical arthritis and Down's syndrome. *N Engl J Med* 1986;**314**:320.

6. Bosma GPM, van Buchem A et al. Cervical spondylarthrotic myelopathy with early onset in Down's syndrome: five cases and a review of the literature." *J Intellect Disabil Res* 1999:**43**(Pt 4): 283-8.

7. Christofaro RL, Donovan RU, Christofaro J. Orthopaedic Abnormalities of Adults with Down's Syndrome. *Dev Med Child Neurol* 1986;103.

8. Morton RE, Khan MA, Murray-Leslie C, Elliott S. Atlantoaxial instability in Down's syndrome: a five year follow up study. *Arch Dis Child* 1995;**72**:115-8.

9. Cremers MJ, Ramos L, Bol E, van Gijn J. Radiological assessment of the atlantoaxial distance in Down's syndrome. *Arch Dis Child* 1993;**69**:347-50.

10. Selby KA, Newton RW, Gupta S, Hunt L. Clinical predictors and radiological reliability in atlantoaxial subluxation in Down's syndrome. *Arch Dis Child* 1991;**66**:876-8.

11. Wellborn CC, Sturm PF et al. Intraobserver reproducibility and interobserver reliability of cervical spine measurements. *J Pediatr Orthop* 2000:**20(1)**: 66-70.

12. Nader-Sepahi A, Casey AT, Hayward R, Crockard HA, Thompson D. Symptomatic atlantoaxial instability in Down syndrome. *J Neurosurg* 2005;**103**:231-7.

13. Taggard DA, Menezes AH et al. "Treatment of Down syndrome-associated craniovertebral junction abnormalities." *J Neurosurg* 2000:**93(2 Suppl)**: 205-13.

14. Callman K. Cervical spine instability in people with Down syndrome. *CMO's Update 7* 1995;4.

15. Cremers MJ, Bol E, de Roos F, van Gijn J. Risk of sports activities in children with Down's syndrome and atlantoaxial instability. *Lancet* 1993;**342**:511-4.

16. Worley G, Shbarou R et al. New onset focal weakness in children with Down Syndrome. *Am J Med Genet.* 2004: July 1. 128A(1): 15 - 18

17. Litman RS, Zerngast BA, Perkins FM. Preoperative evaluation of the cervical spine in children with trisomy-21: results of a questionnaire study. *Paediatr Anaesth* 1995;**5**:355-61.

18. Lin Yuan-Chi. Cervical spine disease and Down syndrome in pediatric anesthesia. *Anesthesiology clinics of North America* 1998;**16**:911-23.

19. Casey AT, O'Brien M, Kumar V, Hayward RD, Crockard HA. Don't twist my child's head off: iatrogenic cervical dislocation. *BMJ* 1995;**311**:1212-3.

Steering group for CSI guideline revision

Pat Charleton. Community paediatrician. Aberdeen

Jennifer Dennis. Paediatrician. Oxford

Jill Ellis. Consultant paediatrician. Hackney

Marian McGowan. Consultant community paediatrician. St George's Hospital

Dominic Thomson . Consultant paediatric spinal neurosurgeon. Hospital for Sick Children. Great Ormond Street

Amy Walmsley. British Gymnastics