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Sport Preparticipation Screening for Asymptomatic Atlantoaxial Instability in Patients With Down Syndrome

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Sport pre-participation screening for AAI in Down Syndrome patients

Position Statement

Introduction

Down Syndrome (DS) is a clinical syndrome comprising of typical facial features and various physical and intellectual disabilities due to extra genetic material on chromosome 21, with one in every 1,000 babies born in the UK affected (1). DS patients are at risk of atlanto-axial instability (AAI) although AAI can occur in other conditions, such as rheumatoid arthritis, but this position statement deals specifically with DS patients and asymptomatic AAI.

AAI, also referred to as atlanto-axial subluxation, is defined as increased movement between the 1st(atlas) and 2nd (axial) cervical vertebra joint articulation, the atlantoaxial joint (2). AAI in DS patients is due to a combination of ligament laxity and bony abnormalities of the atlantoaxial joint. AAI is reported to occur in 6.8 – 27% of the DS population (3) (4) although this varies depending on the age of the patients which you are screening. Less than 1-2% (3) (5) of these patients are then thought to later develop symptomatic AAI although the natural history and progression of AAI is not well understood (2).

The risks associated with AAI are neurological injury from excessive movement of the cervical vertebra impinging on and then damaging the spinal cord although the risk of this during sporting activities is extremely rare (2) (6). Clearly physical activity and sports participation for DS patients has many biological, psychological and social benefits (7) and the Faculty of Sport and Exercise Medicine (FSEM), United Kingdom (UK) wish to promote safe physical activity and sport for all. The FSEM, UK has therefore produced a statement regarding sport pre-participation screening for asymptomatic AAI in DS patients.

Plain lateral cervical spine radiography as a screening test for asymptomatic AAI

1. Cremers et al (6) followed up 91 asymptomatic AAI patients for 1 year after screening them with lateral cervical spine radiography, letting them either participate in unrestricted sport or restricting their sporting activities. They found no difference between the groups in terms of motor or neurological function. They concluded that there is no evidence to support plain radiography screening for asymptomatic AAI.
2. There are diagnostic inconsistencies surrounding asymptomatic AAI on plain radiography, with, for example, Morton et al (8) reporting that it occurs at 4mm whilst other authors 3mm (9) (10) and some 4.5mm (5).

3. AAI diagnosis on plain xray can also change as the patient gets older (4): in 1986 Morton et al (8) followed up 90 children aged between 4-19 yo for 5 years after screening them with plain cervical x-ray and overall the AAI prevalence decreased at follow-up.
4. Obtaining plain lateral cervical spine radiography and then the necessary measurements for AAI is technically difficult (6).
5. Symptomatic AAI is rare in DS patients (2).
6. Plain lateral cervical spine radiography (including neutral, flexion and/or extension views) cannot therefore be recommended as a screening test for asymptomatic AAI in sport (10) (6) (8) (2) (4).

Alternative sport pre-participation screening techniques for asymptomatic AAI

- For sport pre-participation assessment, a focused history and neurological examination of DS patients should be undertaken by an appropriately qualified medical professional or chartered physiotherapist (6) (4), with the preference for the professional to be someone who cares for the patient regularly, on an ongoing basis and is therefore aware of their baseline function (6), e.g. General Practitioner (GP)/Family Physician.
- Selby et al (10) conclude that both plain x-ray and physical examination are insensitive for screening for AAI in DS patients.
- For the neurological examination, Morton et al advise (8) checking for gait disturbance, neck movements, tendon reflexes, and plantar responses. This is similar to British Gymnastics, who have developed their own information sheet and undertake their own screening questions, with no radiological screening (11). This is also supported by Down Syndrome Association (12).
- The 3 screening questions consist of (11):
 - Does the person show evidence of progressive Myopathy? Yes/No
 - Does the person have poor head/neck muscular control? Yes/No
 - Does the person's neck flexion allow the chin to rest on their chest? Yes/No

To help with question B, the person's neck control can be assessed by: laying the person on their back with legs straight and they are then pulled to a sitting position by their hands, with the examiner pulling them from the front (11).

Depending on the results of the neurological examination, assessment of neck control and the 3 screening questions, there will essentially be 2 options for the patient:

- Unrestricted sports participation;
- Restricted sports participation.

High-risk sports for DS patients and symptoms of AAI to be aware of

- Sports considered to put DS patients at higher risk of developing symptomatic AAI include (2):
 - Gymnastics including Trampolining;
 - Diving,
 - Butterfly stroke and diving starts at swimming;
 - Pentathlon;
 - Contact sports such as martial arts, rugby and soccer;
 - High jump.

AAI symptoms for patients, family members, health and sport professionals to be aware of include (13) (11) (4):

- change in gait or use of arms or hands,
- change in bowel or bladder function,
- neck pain,
- stiff neck,
- head tilt,
- how the child positions his or her head,
- change in general function, or weakness.

These 'warning' symptoms need to be promoted to the general population, patients, family members and health professionals, e.g. through a public health campaign (4), to allow them to act if they identify an issue.

Specific signs and symptoms for health professionals to be aware of when they are examining a DS patient at risk of AAI include (2):

- Easy fatigability;
- Difficulties in walking;
- Abnormal gait;
- Neck pain;
- Torticollis or head tilt;
- Incoordination and clumsiness;
- Sensory deficits;
- Spasticity;
- Hyper-reflexia;
- Clonus;
- Extensor-plantar reflex;
- Other upper motor neuron and posterior column signs and symptoms.

Acute management of symptomatic AAI

If you suspect symptomatic AAI, the patient's spine should be immobilized and an urgent assessment, including neurosurgical consult, undertaken within the Emergency department along with MRI spinal imaging (5).

Promoting safe sport for DS patients

To facilitate safe sport for DS patients we need to promote neck conditioning exercises for this cohort of patients, similar to the neck programme promoted in patients with chronic neck discomfort (14), as well as safe sporting practices, e.g. appropriate supervision when undertaking higher risk sports such as trampolining or rugby.

Conclusion

Plain radiography cannot be currently recommended to screen for asymptomatic AAI. DS patients undergoing a sport pre-participation screening should have 3 questions asked as per the British Gymnastics programme and a neurological and neck control assessment undertaken. Neck conditioning exercises should be generally promoted amongst the DS population. Awareness of potential signs and symptoms of symptomatic AAI need to be raised amongst DS patients, family members and professionals caring for this group of patients as well as the need to provide appropriate supervision when DS patients are undertaking certain 'high-risk' sports. These measures will then allow DS patients to maximise the biological, social and psychological benefits of physical activity and sport participation.

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