



Annual Health Check Information for GPs

Cervical Spine Problems

Author: Mr Adrian Casey

Below is Down's syndrome specific information. The information is for GPs and is to be used alongside DSA's Adult Health Book.

About 750 babies with Down's syndrome are born in the UK each year. Most medical practitioners are well aware of thyroid and cardiac issues but perhaps the cervical spine are less well known. Other problems later in life include a higher incidence of acute myeloproliferative disorders and dementia.

Individuals with Down's syndrome can have significant cervical spine problems with neurological problems often ensuing. The most common anomaly is atlanto-axial instability where the first cervical vertebra slips forward over the odontoid peg of the second cervical vertebra, the axis. Figure 1. Instability at the atlanto-occipital level is common and must be considered along with the previously documented atlantoaxial instability.

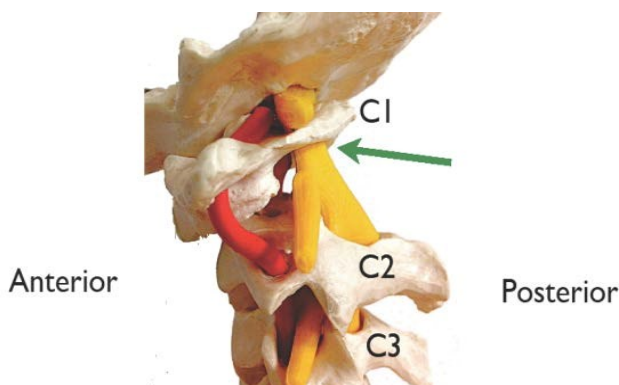


Figure 1

Pueschel and colleagues from Brown University have showed that a significantly greater number of children with Down's syndrome (45 of 78) have cervical spine anomalies than an age and sex-matched group of normal children (3 of 38). (Pueschel, 1990). However, in further longitudinal studies he did not find major progression of instability or neurological problems. (Pueschel, 1992).

Of course people with Down's syndrome can also have similar pathologies to other adults eg slipped disc causing cervical myelopathy or constitutional cervical canal stenosis. These often

present late to the medical team due to communication difficulties as a result cognitive impairment. The latter is quite variable in people with Down's syndrome.

Presentation

Most children will not articulate their neurological difficulties. In most cases there is probably a slow insidious progression of symptoms. The family and carers should be alert to slow clumsy hands, difficulty dressing, drawing or holding a fork or spoon. Many children with Downs are quite keen on dancing, if there are changes here or in other areas of play or mobility, concerns should be raised. Some will complain of neck pain or have a reluctance to turn their head to the left or right. The atlanto-axial complex (c12) is responsible for most of the 50 degrees of head rotation (75%).

Atlanto-axial instability can compress the C2 nerve giving rise to pain in its distribution at the back of the head as well.

It is prudent to suspect atlantoaxial instability in a child with Down's syndrome who presents with decreased motor skills or who develops torticollis, a gait disorder, or any form of progressive paralysis. Although the compression is in the high cervical spine, the first motor symptoms or signs may be discovered in the legs.

In many instances, infections of the pharynx, the middle ear, and/or the upper respiratory tract precede symptoms of atlanto-axial instability.

Clinical deterioration following any of these conditions justifies a complete evaluation. Rarely there will be an acute deterioration following trauma e.g. pushing a child's head underwater playfully, heading a football or even iatrogenic during general anesthesia e.g. for grommets.

On examination there may be no signs if there is just mild instability with no neurological damage. If the spinal cord is damaged one would expect to see upper motor neuron signs of increased tone, brisk reflexes of arms and legs including sustained clonus. In advanced cases there will be a degree of paralysis, and features of a myelopathic hand.

Investigations

Plain x-rays of the cervical spine should ideally include volitional flexion extension x-rays. In the neutral position the c1 c2 bones may well be in normal position.

If radiological abnormalities are picked up or when there are potential neurological symptoms or signs the imaging of choice is MRI of the cranio-cervical junction. Because most children with Down's syndrome will not tolerate an MRI without sedation or GA it is prudent to perform MR angiography and also include the brain, and the whole spine to rule out any other copathologies e.g. slipped disc, vertebral artery occlusion, cva amongst others. The MRA has potential relevance to surgical planning. The spine fixation most commonly performed includes screw fixation of the axis through the pedicle and the atlas. The vertebral artery is at risk with screw misplacement of only a few millimetres. 13% of normal individuals have a high riding vertebral artery near the C2 pedicle (axis) which would be at very high risk. There are also natural variations in caliber of the artery e.g. right or left side dominant or atretic. The radiological abnormality of os odontoideum is believed to be a missed injury with transposition

of the transverse ligament into the fracture site which impairs healing. This is more common in Down's syndrome.

Surgical Treatment

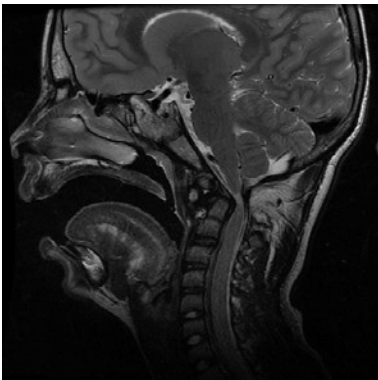
Surgical treatment involves stabilising the atlanto-axial complex. Most authorities would suggest extending the fixation rostrally to include the occiput (skull) in view of the anomalies of their joint (atlanto-occipital joint) and relatively high failure of isolated c12 fusion in Down's syndrome where bony healing and ligamentous laxity compromise the results.

Illustrative Cases

CASE 1

This is a 14 years old boy who through screening was recognised to have atlanto-axial subluxation for 7 years prior to developing neurological problems. He complained to his mother that his legs felt tired and he became progressively less mobile. On examination he had a spastic gait and was unsteady on heel toe gait. Clonus was present bilaterally.

X-rays revealed atlanto-axial instability. MRI showed severe compression at the cervicomedullary junction.



CASE 2

This is a 19 years old male who presented to his medical team when he became less involved at school in sports and walking ability. His family also noticed that he played his computer games with his non dominant hand due to loss of fine dexterity (myelopathic hand) This was secondary to atlanto-axial subluxation with cord compression. He underwent decompression (c1 laminectomy) and occipitocervical fixation. He recovered well and returned to full activity, including his computer games and iPad.



Figure 3 plain x-rays showing atlanto-axial subluxation, with increased atlanto dens interval.

CASE 3

This is a 44 years old man in residential care who became progressively less mobile and had speech problems. There had been visits to his local doctors complaining about a painful neck several months before. After admission to hospital with severe neurological disability he was initially thought to have had a stroke (vertebro-basilar territory).

He had severe tetraparesis with global reduction in muscle power with loss of antigravity function MRC 1-2.

A high definition CT (figure 3) scan showed irreducible atlanto-axial subluxation, with severe cord compression.

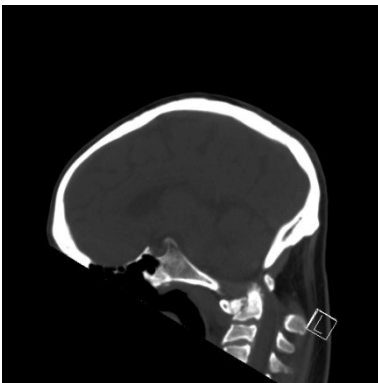


Figure 3 - severe atlantoaxial subluxation. The ring of the atlas has slipped forward and down to the base of c2 odontoid peg of the axis.

The role of surgery for this man is controversial. Surgery if deemed appropriate will involve decompression by removing the posterior arch of the atlas and stabilisation using screws in the occiput and axis C2, & C3. The prospects for neurological recover when anti-gravity power has been lost will be slim. Arguably if his problems had been diagnosed earlier and surgery performed to stabilise his spine, well established neurological problems may have been averted. His original presentation to his GP, and separately to his local casualty department, with neck pain was several months before he became paralysed. These represent missed opportunities.

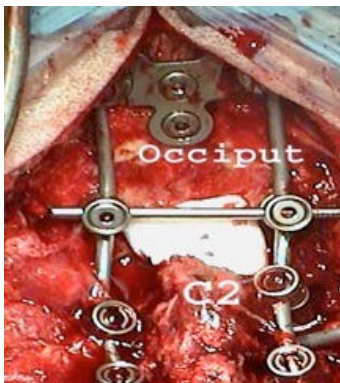
Screening

There is an argument for screening by means of plain x-ray but there is no consensus. The arguments are well rehearsed by both sides. Screening may cause unnecessary stress to the family. There are various degrees of atlanto-axial subluxation and mild ones 5-7mm are probably not at any great risk of neurological problems. Neurological sequelae overall are quite rare. The other side argues that there are many other conditions that are screened for with low pick up rate and less severe consequences if missed. The Olympic committee has changed its mind about the need for screening but certain activities such as swimming, gymnastics and contact sports are risky.

Surgery

Surgery if required, consists of stabilising the unstable atlantoaxial joint. This is typically effected by a screw rod construct joining C1 & C2. Often this is extended to include the occiput due to instability of occipito-atlantal joints and high rate of non-union for Down's syndrome patients, perhaps due to collagen abnormalities and relative hypermobility. Successful fixation of C12 prevents the majority of head rotation. Specific risks of surgery include the potential to damage local structures e.g. spinal cord, dura, C2 nerve or vertebral artery. Instrumentation failure with screw misplacement or pseudo-arthrosis non-union also may occur due to challenging patho-anatomy.

Neurological recovery is related to preoperative status, with those patients with mild neurological dysfunction improving close to normal. Those individuals who are unable to walk preoperatively will have some improvement over several months, but are likely to remain with significant disability.



References

- Pueschel, S.M., Scola, F.H., Tupper, T.B. & Pezzullo, J.C. (1990). Skeletal anomalies of the upper cervical spine in children with Down syndrome. *J. Pediatr. Orthop.* Sep-Oct;10(5):607-11.
- Pueschel, S.M., Scola, F.H. & Pezzullo, J.C. (1992). A longitudinal study of atlanto-dens relationships in asymptomatic individuals with Down syndrome. *Pediatrics.* Jun;89(6 Pt 2):1194-8.
- Nader-Sepahi, A., Casey, A.T., Hayward, R., Crockard, H.A. & Thompson, D. Symptomatic atlantoaxial instability in Down syndrome. *J. Neurosurg* 2005;103:231-7.

The Down's Syndrome Association (DSA) is the only organisation in England, Wales and Northern Ireland which supports people with Down's syndrome at every stage of life.

Down's Syndrome Association, Langdon Down Centre, 2A Langdon Park, Teddington, Middlesex, TW11 9PS. Tel: 0333 1212 300 | Email: info@downs-syndrome.org.uk | Web: www.downs-syndrome.org.uk

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