MINI-SYMPOSIUM: CHILDREN’S ORTHOPAEDIC SURGERY

(iv) Cervical spine problems in children

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Summary
The paediatric spine is the site of a number of conditions, both congenital and acquired, which are unusual in adult practice if not unique to paediatrics. Some insight into the embryological development of the cervical spine, the normal paediatric anatomy and its development into the standard adult anatomy is crucial to understanding paediatric spinal pathology. Conditions which frequently come under the remit of orthopaedic surgeons such as trauma, bony tumours and congenital bony and ligamentous disorders are discussed. © 2006 Published by Elsevier Ltd.

Introduction
Disorders of the bony paediatric spine are the responsibility of many clinicians including orthopaedic surgeons, neurosurgeons, paediatric surgeons, paediatricians and geneticists. Some understanding of the embryological development of the cervical spine, its growth patterns and normal anatomy, in both the child and adult, is necessary to appreciate the spectrum of disorders which may be encountered in paediatric practice. Disorders of the cervical spine in children are uncommon and therefore any one clinician, unless super-specialised, is unlikely to build up much personal experience and therefore the purpose of this review is to synthesise current opinions on a variety of the more common disorders encountered.

Embryological development and normal anatomy
Once the embryo undergoes the process of gastrulation (i.e., the formation of a trilaminar disc), the formation of the notochord induces the overlying ectoderm to form neuroectoderm, from which the brain and spinal cord are derived, and the para-axial mesoderm is induced to form somites. The craniocervical junction and the cervical spine develop from the four occipital and seven cervical somites. The ventromedial portions of the somites become the sclerotomes and one sclerotome contributes to two adjacent vertebral bodies while the neural arch arises solely from the caudal portion of the sclerotome. This process is driven by, at least in part, two families of regulatory genes: the Box and Pax families and is reviewed in detail by David and Crockard.¹

Chondrification of these mesenchymal elements begins at 6 weeks of development in utero and ossification proceeds by endochondral ossification from 9 weeks. The subaxial vertebrae ossify from three primary centres, one in the vertebral body and one in each of the neural arches. The

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Synchondrosis between the two neural arches fuses by age three and the synchondroses between the arches and the body between the ages of 3 and 6 years. In addition, secondary ossification centres exist at the tips of both the transverse and spinous processes, and ring apophyses at the superior and inferior endplates which may remain unfused until early adulthood.

The first two vertebrae are unique in their ossification. The atlas (C1) is composed of three centres of ossification with individual centres in the anterior arch and in each neural arch. The anterior arch is not ossified in most children at birth, generally becoming visible as an ossification centre by 1 year and fusing with the neural arches around 7 years. The two neural arches generally fuse posteriorly by 3 years. Therefore, developmentally normal non-fusion can appear pathological. The axis (C2) is more complex still, with four ossific centres at birth (body, both neural arches and one in the odontoid process which itself forms from two centres fusing in utero). Fusion of these synchondroses proceeds first with the neural arches posteriorly by the age of 3 years, while the odontoid process fuses with the body of C2 by 3–6 years; this subdental synchondrosis is frequently confused with a fracture (Fig. 1). In addition, a secondary centre of ossification appears in the tip of the dens at 3–6 years fusing with the rest of the dens by age 12.² Radiographically synchondroses appear sclerotic while fractures do not.

Normal radiographic variants

There are a number of anatomical differences between the adult and the paediatric spine which can cause confusion when assessing for pathology. In flexion, the normal adult anterior atlanto-dental interval (ADI) is ≤ 3 mm while in children it may be normal up to 5 mm. In children it is normal to have some overlap of the lateral masses of C1 relative to the dens and up to 6 mm can be seen in children as old as 8 years. In adults, ≥ 8 mm usually indicates disruption of the transverse ligaments indicating an unstable injury. The variable fusion of the C1 synchondroses can further confuse the picture for the unwary. In addition, flexion can result in an increase in the distance between the tips of the spinous processes of C1 and C2 which is a normal finding in children. Pseudosubluxations of C2 on C3 and, to a lesser extent, C3 on C4 are probably the most commonly over-interpreted findings on paediatric cervical spine films (Fig. 1). Up to 40% of children less than 8 years old may demonstrate this phenomenon, with C2 moving 2–3 mm on C3. The spinolaminar line can be used to differentiate normal from abnormal movements (Fig. 2).

The cervical lordosis seen in adults, the absence of which may be pathological, is commonly not seen in children. Prevertebral soft tissue swelling, a useful sign in adult cervical spine injury, is unreliable in children, being dramatically increased when crying Table 1.

Figure 1 Lateral radiograph in a child showing the subdental synchondrosis and pseudosubluxation of C2 upon C3 and C3 upon C4.

Figure 2 Lines used in assessing the alignment on a lateral cervical spine radiograph: (1) anterior vertebral line; (2) posterior vertebral line; (3) spinolaminar line and (4) spinous process line.
Fractures are easy to misdiagnose in children especially those in the region of the occiput to C2 where the complex normal developmental anatomy can be confusing. Avellino et al.\(^4\) found that 19% of paediatric cervical spine injuries were misdiagnosed on initial assessment, especially in children under 8 years old, and the majority of these were due to misinterpretation of normal findings.

### Traumatic injury to the cervical spine

Spinal injury in children is rare but can have catastrophic consequences. In the UK, 3.4% of paediatric trauma admissions have spinal column or cord injury and of these approximately 45% are in the cervical spine. Spinal cord injury presents in about 16.5% of all spinal injury admissions and 55% of all cord injuries were in the cervical spinal cord.\(^5\) Spinal column injuries are more frequent after road traffic accidents and falls (especially from heights in excess of 2 m) and should be particularly suspected in those with a depressed level of consciousness (from head injury) as well as in patients with chest/multiple injuries.

### Biomechanical factors

The anatomical differences between the adult and the paediatric spine result in a different injury profile between the two groups which lessens with development. Overall the elasticity of the spinal column results in a lower incidence of spinal column injuries when compared to adults; this elasticity does not, however, extend to the neural structures. Young children have proportionally larger heads and therefore a higher centre of gravity, resulting in relatively greater torque and acceleration forces applied to relatively underdeveloped neck musculature and relatively more lax and elastic ligaments and joint capsules. They are therefore more prone to flexion/extension injuries. In addition, the fulcrum of this force differs from the normal adult fulcrum of C5/6 and instead lies at C2/3. The stability of the adult cervical spine rests, in part, on the coronal orientation of the facet joints which in the child are more horizontally oriented predisposing to subluxation and translational movement of the vertebral bodies. This translational movement is exacerbated further by the normal anterior wedging of the superior aspects of the vertebral bodies in children (Fig. 3).

These anatomical differences result in a pattern of injury which differs from that seen in adults. The differential elasticity of the spinal column in comparison with the neural elements results in the syndrome of spinal cord injury without radiological abnormality (SCIWORA). In addition, children are twice as likely to injure the bony upper cervical segments (C1-4) rather than the lower segments although the frequency of lower cervical injury increases with age. While fractures are the most common bony injury regardless of age, dislocations occur more frequently in the upper segments and in the younger age group (<8 years).\(^6\) Injuries in the younger child are also more likely to result in an associated cord injury.

The change from a paediatric pattern of injury to that of adults has traditionally been held to occur around the age of 8–9 years when injuries in the mid-cervical spine become more frequent. By this age all the synchondroses between the centres of ossification have fused. However, some authors maintain that the trend to an adult pattern of

![Figure 3](image-url)
injury does not start until 10–11 years and does not equal that of the adult until late adolescence.\textsuperscript{7}

**Initial management**

All traumatised children should be suspected of having sustained a significant spinal column or cord injury and managed by standard principles namely appropriate airway management with control of the cervical spine, and assessment of the adequacy of ventilation and circulatory status.

**Immobilisation**

Immobilisation is more complex in children than in adults where ‘sand-bags, cervical collar, tape and spinal board’ are the standard of care. To achieve a neutral alignment in the paediatric cervical spine, account must be taken of the relatively large head compared to the torso which tends to force the neck into a degree of flexion when lying on a flat surface,\textsuperscript{8} a position not prevented by application of a cervical collar. This applies to children under 8 years but particularly to those under 4 years. To counteract this angulation, the torso may need to be elevated 25 mm or the head placed in an occipital recess.

**Imaging**

Imaging is, even to a greater degree than in the adult, a contentious issue. Particular causes of dissent are the imaging of asymptomatic children, of comatose children and those under 5 year of age.

Laham et al.\textsuperscript{9} evaluated 268 children who they retrospectively divided into high risk (those complaining of neck pain and those unable to communicate because of age (<2y) or injury) and low risk for spinal injury after head injury. They used the standard trauma cervical spine series (AP, lateral and open mouth radiographs) and none of the low-risk group versus 7.5% of the high-risk group had injuries. A prospective study applying the NEXUS decision instrument to children indicated that the presence of specific risk factors (midline cervical tenderness, altered level of consciousness, intoxication, neurological abnormality or other injury such as a long bone fracture which might distract from cervical pain) rendered children at high risk of cervical spine injury, correctly identifying all children with cervical spine injury. MRI is useful in non-assessable children and may supplant plain radiographs in children at high risk of cervical spine injury. MRI is useful in non-assessable children and in children with fixed or transitory deficits.

**Paediatric patterns of injury**

Some patterns of injury are unique in children and are particular to certain stages of development i.e., neonatal spinal cord injuries and odontoid epiphysiolysis. Other injuries that are most common in, but not unique to, children are: os odontoideum, SCIWORA, atlanto-occipital dislocation (AOD) and atlanto-axial rotatory fixation. Older children in addition present with injuries seen in the adult population and management of these pathologies is not discussed here.

**Neonatal spinal cord injuries**

Spinal cord injury at the time of delivery is rare, occurring in around 1/60000 live births. It occurs most frequently in the cervical or cervico-thoracic region, the former being most common and associated with rotational forceps deliveries of cephalic presentations, the latter in breech presentations. Clinically, they tend to present with apnoea and flaccid quadriplegia. Of the 22 neonates reported by MacKinnon et al.\textsuperscript{15} five of the 14 with cervical cord injury died before 3 months of age and six of the seven still alive at last follow-up were ventilator dependent. A smaller series from Oakes indicated that bedside diagnosis was often incorrect and in their series of five infants only one had an abnormal lateral radiograph.\textsuperscript{16}
Odontoid epiphysiodesis

The subdental synchondrosis (neurocentral synchondrosis) is relatively vulnerable to injury in children until fused. The lateral radiograph may show abnormal angulation (usually anteriorly) or indeed frank dislocation of the dens. As it is a fracture through a physis it is very likely to heal and it can be successfully managed in a halo orthosis. If it is significantly dislocated, then reduction in the halo ring under fluoroscopic guidance is appropriate. Successful fusion rates of 80% have been described following 10–18 weeks of immobilization. Others have opted for operative management (anterior odontoid screw fixation or posterior C1–C2 fixation) although the published numbers are much smaller.17

Spinal cord injury without radiographic abnormality (SCIWORA)

The SCIWORA lesion results in a sensory and/or motor deficit with no injury apparent on imaging.19 The name is somewhat misleading as in reality it refers to no evidence of bony injury on plain radiology or CT; with the advent of MRI it has become apparent that these children often have neural (haemorrhage/oedema in spinal cord) or extraneural pathological processes (traumatic disc protrusion, epidural haematoma, ligamentous injuries) which correlate with the clinical findings. SCIWORA is rare, accounting for around 4.5% of children with spinal injuries, and following serious trauma (motor vehicle accident, fall from height). It usually occurs in the cervical spine and less frequently in the thoracic spine. It presents with a complete or partial deficit although initial assessment is often clouded by haemodynamic instability. About a quarter of patients present in a delayed fashion.19 It was the incidence of delayed deterioration and of recurrent SCIWORA that has led to the suggestion that there exists in such children a state of ‘occult instability’, which renders them particularly at risk, of further deterioration from repeated stress.20

Children with abnormal or transiently abnormal symptoms should be immobilised as for an unstable injury. If there are any doubts about the normality of whole spine radiographs, then the patient should undergo high-resolution CT and 3D reconstruction, particularly of poorly visualised areas. If there is any evidence of a fracture or fracture/dislocation, then these children are managed as appropriate for that fracture. If there are no radiographic abnormalities, then MRI should be obtained with some urgency, looking for a compressive lesion (e.g., traumatic soft disc herniation, haematoma). In patients with neurological abnormalities, the MRI is almost always abnormal with respect to the neural and/or the extraneural tissues although these abnormalities may be subtle. Children who have had transient symptoms or ongoing neck pain/paravertebral spasm should be immobilised and flexion/extension views performed looking for overt instability when neck pain has settled.21

The majority of cases of SCIWORA with no compressive lesion on MRI are managed non-operatively with a short course of corticosteroids and rigid external immobilisation for 12 weeks, checking flexion/extension views after this period to rule out late instability. Outcome is ultimately related to the admission neurological status. Those admitted with complete lesions rarely improve; hence, younger children tend to do worse as they often have more severe and segmentally higher injuries. Patients with mild-to-moderate injuries or no cord changes on MRI will tend make a better recovery.

Atlanto-occipital dislocation (AOD)

Traumatic AOD is a common cause of death.22 It is an injury that children are anatomically predisposed to because of the higher fulcrum and increased ligamentous laxity already described. Over the last decade, there have been around 100 reports of survivors of AOD of whom 41 have been in the paediatric age group.23,24 The stability of the craniovertebral junction in children is very reliant on the ligamentous structures as the reciprocal concavities of the atlas and convexities of the occipital condyles, a feature of the spine at maturity (>8–10 years), are absent. The particular ligamentous structures of relevance are the tectorial membrane (a continuation of the posterior longitudinal ligament), and the cruciate, alar and apical ligaments.

Traumatic AOD results from high-energy impacts causing rupture of the tectorial membrane and the alar ligaments, most frequently by hyperextension/rotation injuries. Patients often die at the scene or present after major trauma, often with an associated head injury, a quadriplegia and cardiopulmonary instability. There may in addition be lower cranial nerve palsies. Many will have been endotracheally intubated and ventilated making assessment of neurology very difficult. A small number of patients present without initial deficit. AOD may therefore be missed on the initial examination.

These injuries have been classified into three specific types, namely anterior occipital displacement (Type I), longitudinal distraction (Type II) and posterior displacement (Type III).25 A number of methods have been devised to diagnose AOD from plain lateral radiographs; some rely upon absolute distance measured on lateral radiographs (Dens-basion interval, Dublin measure), others on ratios of measurements (Powers ratio, Lee’s X-line). None of these techniques is ideal in children because visualisation of the opisthion is difficult and the odontoid process is incompletely ossified in young children. More recently, Sun et al.26 introduced the C1-2:C2-3 posterior interspinous ratio as a sensitive method of diagnosing tectorial membrane abnormalities on MRI.

Cursory examination of the above assessments reveals that all of the measurements are taken from bony landmarks which do not themselves participate in the articulation of the occiput and C1. Therefore, one of the best methods of assessing the articulation is with fine cut (1–2 mm) CT through C0–C3 and coronal/sagittal reconstructions. Generally no condyle to C1 measurement is greater than 3 mm.21 MRI gives a good assessment of the joints and the ligamentous structures, as well as identifying spinal cord injury. MRI when combined with CT may allow diagnosis of partial AOD when there is bony dislocation but the tectorial membrane is intact. The tectorial membrane is probably the critical structure for ensuring stability.26
Initial management comprises in line immobilisation followed by early application of a halo vest. Definitive management thereafter requires fusion across the whole occipito-atlantoaxial unit either with a contoured loop construct or a combined transarticular C1-2 screw fixation and suboccipital screw fixation. Both methods require bone grafting in addition.\textsuperscript{27}

**Atlanto-axial rotatory fixation (AARF)**

Fixed subluxation of the atlanto-axial complex, responsible for 40% of cervical rotation, is caused by a variety of conditions including otolaryngological infection/surgery (synonymous with Grisel’s syndrome) and is predisposed to by a variety of others (Down syndrome, juvenile chronic arthritis, the mucopolysaccharidoses (MPS)). Trauma, often insignificant, accounts for a substantial minority of cases. Patients present with neck discomfort, limited rotation and torticollis. They may assume what has been described as the ‘cock robin’ deformity with the head rotated to one side and laterally flexed to the other. Patients will be unable to rotate the head past the midline to the opposite side. Neurological deficit is rare (Fig. 4).

On both plain films and static CT, it can be difficult to differentiate normal rotation from a fixed subluxation, although the value of both of these investigations is in defining associated congenital abnormalities or fracture. This problem has been studied systematically by Pang and Li.\textsuperscript{28,29} They utilised 3-mm axial CT from the clivus to the base of C3 firstly to, define the dynamic relationship between the occiput and C1 (which move as a single unit) as well as C1 and C2, in normal children. This allowed construction of a normal motion curve, which was highly reproducible between individuals. Around 0°, C1 crosses C2 in either direction to start rotation, and for the first 23° of rotational movement from neutral C1 rotates without C2 moving. From 23° to 65°, C2 starts to rotate progressively with C2 and by 65–90° of rotation C1 and C2 move in unison at a fixed angle of separation (mean 43° of separation). Pang and Li then went on to define the motion curves obtained in forty children presenting with painful ‘cock-robin’ deformities by imaging them in the presenting position, in a neutral rotational position and with the head forcefully turned to the opposite side as much as the patient could tolerate. They defined five groups ranging from a fixed dynamically irreducible angle between C1 and C2 (AARF I), to an abnormal but reducible C1-2 separation angle which never reached zero no matter how forceful the rotation (AARF II), to patients in whom C1 would cross C2 but only in extremes of rotation to the contralateral side (AARF III). Some of the patients had normal dynamics of motion despite the deformity and a small number fell into a diagnostic grey zone.\textsuperscript{29}

The success of treatment modalities in AARF is related to the duration of the preceding period of subluxation. The quicker the intervention the more likely it is to succeed. In the paper from Pollack’s group in Pittsburgh a quarter of the patients in whom C1 would cross C2 but only in extremes of rotation to the contralateral side (AARF III) went on to require open reduction and fusion.\textsuperscript{27}

Figure 4 3D reconstruction of cervical spine CT in a child with atlanto-axial rotatory fixation. The left C1 articular facet is dislocated anteriorly over the C2 articular facet.

The third part of the landmark series by Pang and Li assessed 50 children presenting with a painful torticollis. It subjected them to the three position CT described above; 29 were diagnosed with AARF fairly equally distributed between the three classes. All went on to halter or calliper traction on diagnosis and upon reduction were either immobilised in a cervicothoracic brace or halo jacket, those whose deformity was irreducible or recurred in a halo orthosis underwent fusion in the best possible alignment. There was a clear gradation in failure of reduction, degree of difficulty in obtaining and then maintaining reduction, and the need for operative fusion between Type I and III patients with Type II being intermediate. As in other studies, delays in diagnosis and treatment resulted in worse outcomes independent of degree.\textsuperscript{30}

Patients with ‘cock-robin’ deformities should be rapidly assessed with dynamic CT, then typed and put in traction rapidly to reduce the deformity with acute cases being placed in cervicothoracic bracing. Early intervention is likely to result in improved outcome and avoidance of operative interventions. Chronic cases may require halo bracing when the deformity is reduced. Recurrent dislocation and failure of reduction are indications for operative intervention.
Non-traumatic disorders of the paediatric cervical spine

Primary tumours of the cervical spine

Primary tumours of the cervical spine in children are extremely rare. It is therefore unlikely that any one surgeon will build up a large series of cases, and indeed such series are lacking from the literature. Fortunately, the majority of such neoplasms are benign and present in a fairly typical fashion with persistent neck pain, especially at night or when resting; neurological compromise is infrequent. Tumours may be primary tumours of bone, tumours of non-osseous bony elements or be related to diffuse neoplastic processes elsewhere (acute leukaemia and lymphoma). Plain X-rays may demonstrate lytic lesions or vertebral collapse but MRI is the most useful screening tool. Once an abnormality is demonstrated on MRI, then fine-cut computed tomography through the vertebra involved gives better definition of the anatomy.

Osteochondroma

Osteochondromata (synonymous with exostoses) are the most common benign bone tumours although less than 3% occur in the spine. They may cause local pain but are often asymptomatic, presenting with a painless mass related to a spinous process or with symptoms of neural compression. A recent literature review identified 165 reported cases of spinal exostoses of which about half were solitary and one-third part of the multiple hereditary exostosis syndrome (diaphyseal aclasis). About half of the reported lesions were in the cervical spine and the majority arose from posterior elements of the vertebra giving rise to neurological symptoms. A significant proportion of cases were younger than 20 years. Complete excision is curative with a very low recurrence rate, although thought should be given to stabilisation in wide resections.

Osteoid osteoma/osteoblastoma

These tumours are histopathologically similar entities, both bone-producing tumours that arise in the long bones and in the posterior vertebral elements especially the neural arch. They tend to occur in adolescents and young adults. Osteoid osteomas are less than 2 cm in diameter and tend to present with localised pain, which classically, although infrequently, responds to salicylate analgesia. Cervical spine osteomas can present with torticollis or radicular pain. Radiographically both lesions have a lytic nidus surrounded by a sclerotic margin and appear “hot” on bone scans.

Histologically both tumours consist of osteoid trabecula containing a fibro-vascular stroma surrounded by dense cortical bone. Osteoblastomas are differentiated by size (osteoblastomas being greater than 2 cm in diameter) and have an increased predilection for the spine (30–50%) with a tendency to form expansile masses. Osteoid osteomas are curable by resection; similarly a proportion of osteoblastomas are cured by surgery although they have a significant recurrence rate. Frank malignant transformation is rare.

Aneurysmal bone cyst

Aneurysmal bone cysts are benign bone pseudo-tumours of unknown aetiology. They are expansile, containing thin-walled, blood-filled, cystic cavities and are often located in the metaphyses of long bones; they comprise about 15% of primary spine tumours with a predilection for the thoracolumbar area (Fig. 5). They tend to occur in the posterior elements extending into the pedicles and vertebral bodies. Individuals tend to present with nocturnal/recumbency pain or localised swelling and less frequently with neurological compromise. There are reported cases of rapid onset of neurological compromise after vertebral collapse. They are most frequent in the first and second decades of life (mean age at diagnosis of 14 years), with a slight female preponderance.

Optimal therapy is controversial and depends upon the age of the patient, the size and accessibility of the lesion and the need to minimise intraoperative blood loss. Preoperative embolisation followed by wide excision and if necessary spinal instrumentation to maintain stability is probably the intervention of choice. Curettage alone is
likely to result in early progression of residual disease and the role of radiotherapy is controversial.\textsuperscript{35,36}

**Giant cell tumour**

Giant cell tumours are expansile, vascular lesions that account for less than 2\% of all vertebral tumours. They are rapidly progressive, aggressive tumours and often present with pain and neurological compromise. They are most often found in the vertebral body (especially in the sacrum) and appear radiolucent and expansile. Aggressive surgical treatment, often requiring multiple surgical approaches, is the usual requirement with recurrence rates of 8–16\% after complete excision and bone grafting. Radiotherapy may have a role for tumours in which complete resection is not possible (Fig. 6).\textsuperscript{37}

**Langerhans cell histiocytosis (LCH) and eosinophilic granulomata**

Eosinophilic granulomata arise from the reticuloendothelial system and are a localised form of LCH. LCH is a challenging disease which presents in a spectrum of disorders ranging from a spontaneously regressing solitary bone lesion to a multisystem and life-threatening disorder. The incidence of LCH is 2–5/million/annum and is probably underdiagnosed; the peak age is between 1 and 4 years with a slight male preponderance.

Bone lesions are found in both localised and disseminated forms of the disease and are frequently apparent in the vertebra where they are typically osteolytic lesions which may progress to a collapsed vertebral body (\textit{vertebra plana}). These lesions are frequently asymptomatic or can present with localised pain. Treatment depends upon the extent of disease, and for those with limited disease (i.e., bone disease without visceral involvement) the prognosis is excellent. Therefore, appropriate staging of the disease is vital and children should be referred to a paediatric oncologist and screened radiologically. Treatment for localised bone disease is primarily conservative with rest, analgesia and bracing; over time the vertebra often reconstitutes. Some recommend curettage of lesions and corticosteroid instillation.\textsuperscript{38}

**Congenital anomalies of the cervical spine**

There are a large number of congenital malformations of the cervical spine and craniocervical junction, the detail of which are beyond the scope of this review. However, some of the more common and illustrative have been selected for discussion.

**Down syndrome**

Trisomy 21, or Down syndrome, is the commonest inherited chromosomal disorder occurring in 1 in 660 live births. Children and adults with Down syndrome are prone to cervical spine disease, including instability, although the natural history is unclear. The majority of pathology occurs at the craniocervical junction particularly at the atlanto-axial and atlanto-occipital joints.

The reason for this increased incidence is two-fold: abnormal anatomy and the effects of ligamentous laxity. At maturity, the atlanto-occipital joint is stabilised by the reciprocal joint surfaces of the occipital condyles on the superior \textit{C1} articular surface, the associated joint capsules, atlanto-occipital membranes and the tectorial membrane. Down syndrome patients have variable joint morphology with flat or ‘rocker-bottom’ joints and lax ligaments. At the atlanto-axial joint where stability relies on the bony integrity of the dens and the transverse ligament, ligamentous laxity again predisposes to instability, further exacerbated by the significant incidence of bony abnormalities of the dens (os odontoideum, hypoplasia and abnormal ossification of the arch of \textit{C1}). One of the major areas of controversy is the demarcation between \textit{instability} i.e., pathological intersegmental motion which threatens neural integrity and \textit{hypermobility} i.e., increased mobility not indicative of loss of structural integrity that is known to occur in trisomy 21. The standard measurements used to indicate instability may not apply in the patients with Down syndrome.

Atlanto-occipital hypermobility is observed in more than 60\% of individuals with Down syndrome although it is not usually associated with neurological risk.\textsuperscript{39} If hypermobility exists in the context of bony anomalies of \textit{C1} or the skull base then there is an increased risk of neurological compromise, which usually co-exists with atlantoaxial instability.\textsuperscript{40}

![Figure 6](image-url) **Figure 6** Midsagittal MRI showing expansile space occupying lesion involving the \textit{C4} spinous process. Histologically this was demonstrated to be a giant cell tumour.
The majority of studies of this population have concentrated on atlanto-axial instability, the incidence of which is variously reported as between 10 and 30% depending on the study methodology. Pueschel and Scola found an incidence of 14.6% based upon measurements of the ADI in flexion and extension in 404 children with Down syndrome; of these children only six were symptomatic (1.5% of the cohort). The ADI (distance from the back of the arch of C1 to the front of the dens) is normally less than 4 mm in children and gives indirect information about the neural canal width. When the ADI is 10 mm or more it is likely that the cord is significantly compromised (Fig. 7). This method of screening has been criticised and is probably useful only as a guide as to which patients should go on to MR imaging looking for evidence of cord pathology.

Children tend to present with gait abnormalities, diminished exercise tolerance and occasionally neck pain or with a spinal cord injury. Posterior cervical fusion either instrumented or with autologous bone graft is warranted after reducibility has been established. The occiput should be incorporated into the construct when there is, in addition, significant atlanto-occipital subluxation, a congenital osseous abnormality of the C1 ring following transoral odontoidectomy for ventral compressive pathology.

Anomalies of the odontoid process/os odontoideum
Congenital anomalies of the odontoid process include aplasia (its complete absence), hypoplasia (its partial absence) and os odontoideum, defined as an osicle with smooth circumferential cortical margins lying cephalad to, but without osseous continuity with, the body of C2. It can be described as orthotopic when the osicle moves with the ring of C1, and dystopic when functionally fused to the basion.

It is debatable whether os odontoideum should be included in a section on congenital disorders of the spine. Although it is found with increased frequency in children with congenital disorders of the spine characterised by instability at the craniovertebral junction e.g., Down syndrome and Morquio syndrome, there is evidence of a traumatic origin for the condition; there are, for example, well-documented cases of normal odontoid processes being replaced by an os on later examination. Crockard and Stevens have suggested that the instability secondary to the underlying congenital condition results in fracturing of the dens and the subsequent interposition of the lax transverse ligament results in an established non-union and an unstable atlanto-axial joint.

Children tend to present asymptptomatically after screening radiographs or symptomatic with neck pain, loss of neck movement or severe neurological injury after relatively minor trauma. Asymptomatic patients without symptoms or signs of neurological compromise may be managed conservatively and followed closely. Symptomatic individuals require posterior C1–C2 fixation and fusion with postoperative Halo fixation or alternatively transarticular screw fixation.

Klippel–Feil syndrome (KFS) and disorders of segmentation
KFS describes a heterogeneous cohort of patients unified only by the presence of a failure of normal segmentation of the cervical spine (cervical synostosis). The classic triad of short neck, low posterior hairline and limited neck movement is seen in fewer than 50% of people with a KF type anomaly. A wide variety of congenital anomalies are associated with the finding of failure of cervical segmentation including congenital scoliosis, rib abnormalities, deafness, cardiac and genitourinary abnormalities and Sprengel’s deformity (an abnormally elevated and hypoplastic scapula). In some children, the KF anomaly may be associated with another eponymous syndrome e.g., Goldenhar syndrome (synonymous with hemifacial microsmia and oculoauricular dysplasia) or Wildervanck syndrome (synonymous with cervico-oculoacoustic syndrome). A minority of cases of KFS have a genetic inheritance basis with both autosomal dominant and recessive variants.

Excitingly KFS has been linked to Sprengel deformity and Chiari malformations as being primarily a defect in post-otic neural crest cell fate.

Figure 7 Axial MRI demonstrating a large atlanto-dental interval in a child with Down syndrome.
choices i.e., ectopic ossification or connective tissue formation.

KFS occurs in 1:40,000 live births with a slight female preponderance. Clinical features are determined by the pathoanatomy of the fusion with more rostral and more extensive fusions tending to present earlier with cosmetic complaints, neck pain or cervical radicular or myelopathic symptoms. Alternatively asymptomatic KFS may be noted during assessment of other congenital anomalies (Fig. 8).

Three specific patterns of fusion are of particular concern as they connote high risk for symptomatic instability: C2–C3 fusion with occipitalisation of the atlas, extensive cervical fusion with an abnormal occipitocervical junction and two fused segments with an interposed open joint space. In each instance, congenital fusion of motion segments predisposes to altered biomechanics at mobile segments and predisposition to pathological instability and neurological compromise. Patients with symptomatic instability or neurological compromise are candidates for an appropriate surgical fusion.

Osteochondral dysplasias
Osteochondral dysplasias are defined as abnormalities of bone or cartilage growth and remodelling in development. They are a heterogeneous group of heritable disorders with more than 120 having been identified. Achondroplasia and spondyloepiphyseal dysplasia (SED) and its subtypes are the most frequently encountered.

Achondroplasia is a common form of disproportionate dwarfism with autosomal dominant (20%) and sporadic (80%) inheritance with an incidence of 1 in 26,000 live births. Generalised spinal stenosis with spinal cord compression can occur which may require operative decompression in childhood; more frequently reported, however, is cervicomedullary compression. Ryken and Menezes described six achondroplastic children presenting with pain, ataxia, incontinence, recurrent apnoeic spells and respiratory arrest. Radiological evaluation revealed marked foramen magnum stenosis, ventrolateral cervicomedullary junction compression secondary to basilar invagination and dorsal cervicomedullary junction compression secondary to ligamentous hypertrophy and invagination of the posterior atlantal arch. All improved with dorsal decompression without fixation. In a prospectively evaluated series of 11 children, two had radiological evidence of cervicomedullary damage on MR1 and underwent immediate decompression; two underwent interval decompression after developing signs of compression. All 11 children were asymptomatic at a mean of 4.6 years. Simple posterior decompression without fusion may predispose to deformity and simultaneous fusion may be most appropriate.

SED is a term for a group of conditions resulting in short trunk disproportionate dwarfism. SED congenita (SEDC) is complicated by ligamentous laxity often combined with odontoid hypoplasia or os odontoideum. This, as in Down syndrome, can result in atlanto-axial subluxation and myelopathy.

The incidence of myelopathy may be as high as 35% in SEDC and symptomatic myelopathy requires posterior decompression and fusion.

Mucopolysaccharidoses (MPS)
The MPS are a heterogeneous group of lysosomal storage disorders each caused by a deficiency in an enzyme involved in degradation of glycosaminoglycans, resulting in cell, tissue and organ dysfunction. Children with both Morquio
syndrome (MPS IV) and Maroteaux–Lamy syndrome (MPS VI) are at risk of cervical myelopathy secondary to craniovertebral junction abnormalities. In Morquio syndrome, children have an os odontoideum secondary to ligamentous laxity with repeated trauma retarding ossification; in addition there is a stenotic foramen magnum secondary to invagination of the posterior rim of C1 and a soft tissue mass of reactive tissue around the unossified dens (Fig. 9a). This results in ventral and posterior compression and a myelopathy. Craniovertebral anomalies in MPS VI are less common than in Morquio’s syndrome and atlanto-axial subluxation is rare. They too suffer from foramen magnum stenosis, in this syndrome secondary to marked thickening of the posterior longitudinal ligament (Fig. 9b).

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References


Figure 9  (a) Lateral radiograph demonstrating cranio cervical instability and os odontoideum in Morquio syndrome. (b) Sagittal MRI demonstrating the thickened posterior longitudinal ligament found in Maroteaux–Lamy syndrome.
Cervical spine problems in children


