Evaluation and treatment of congenital and developmental anomalies of the cervical spine

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Congenital and developmental osseous abnormalities of the cervical spine can result in neural compression ranging from the medulla oblongata to the cervicothoracic spinal cord junction. These may present in infancy as scoliosis and even limb weakness. A high index of suspicion is essential. Neurodiagnostic imaging relies on high-resolution computerized tomography (CT) scanning and three-dimensional CT reconstructions as well as magnetic resonance imaging and angiography. The anatomical/physiological CT factors considered when developing a surgical approach are: 1) the stability and reducibility of the lesions; 2) direction and manner of encroachment of the lesion on the neural structures; 3) neural and vascular abnormalities; and 4) growth potential of the affected area. Primary stabilization is required for reducible lesions, whereas irreducible lesions are decompressed in the manner in which encroachment has occurred. Instability, whether present before or after operative intervention, required spinal stabilization. Illustrative examples of this approach are presented.

KEY WORDS • cervical spine • Klippel–Feil syndrome • congenital scoliosis • quadriparesis • congenital malformation

Abbreviations used in this paper: CCJ = craniovertebral junction; CMJ = cervicomedullary junction; CT = computerized tomography; MR = magnetic resonance; VA = vertebral artery; VB = vertebral body; 2D = two-dimensional; 3D = three-dimensional.

Classification of Congenital Anomalies of the Cervical Spine

Table 1 provides a practical classification of the most frequently encountered congenital cervical anomalies,1–9,13,14 which are divided into congenital and developmental abnormalities. The latter has its basis in abnormal embryology leading to symptomatic abnormalities during early childhood and into adulthood.

Basilar invagination by itself is very rarely congenital but does occur developmentally as a result of atlantal assimilation and segmentation failures of the upper cervical spine, leading to atlantoaxial dislocation and subsequent invagination through developmental changes at the skull base.10 This can be further compounded by condylar hypoplasia and proatlantal segmentation failures. Other anomalies in this region consist of failures of segmentation, failures of fusion of different components of each bone or hypoplasia, and ankylosis. In cases involving absent components of the atlas or hemivertebrae in which there are segmentation failures, the patient may present as early as a few months of age with torticollis and at a later stage with congenital scoliosis.

The developmental abnormalities result from an abnor-
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...mal substrate-in-formation and can lead to the subsequent abnormalities. Os odontoidum is now recognized as being traumatic in origin. Trauma occurs when the child is between the ages of 1 and 4 years and may be unrecognized as a fracture through the neck of the odontoid process, leading to subsequent sequestrum formation (supplied by the occipital artery through the apical ligament) and hypertrophy. Progressive dynamic loads on the craniocervical region lead to instability and subsequent neurological injury. The syndromic abnormalities of skeletal dysplasias, Goldenhar syndrome, the spondyloepiphysial dysplasia, and Conradi syndrome fall into the developmental category. The problems associated with osteogenesis imperfecta may be observed in infancy, but more commonly in childhood.

**Congenital Abnormalities of the Cervical Spine: Symptoms and Signs**

Table 2 provides a summary of acute and chronic signs and symptoms manifesting in patients with abnormalities of the cervical spine. Limb weakness, respiratory failure, and quadriplegia are signs indicative of upper cervical spine lesions; however, torticollis, failure to thrive, repeated aspirations, and swallowing abnormalities may be chronic deficits. Limb weakness may also be chronic. Headache and neck pain were demonstrated in approximately 58% of our 4800 patients with craniocervical abnormalities.

There is a wide variety of these congenital anomalies of the cervical spine and craniocervical region; they develop as single lesions or as multiple anomalies in the same individual and involve both osseous and neural structures. An insult to both may occur and result in a combination of abnormalities.

**Imaging Features of Congenital and Developmental Anomalies of the Cervical Spine**

The factors considered in developing a surgical approach to such lesions include the following: 1) the stability and reducibility of the lesion; 2) the direction and manner of encroachment of a lesion on the neural structures; 3) presence of neural and vascular abnormalities such as a Chiari malformation or syringohydromyelia, meningocele, and tethered cord; and 4) growth potential of the affected area. The stability of the region after a surgical approach must be considered as well as the potential need for radical resection and subsequent iatrogenic instability. Thus, reducible lesions require primary stabilization, whereas irreducible ones require decompression in the manner in which an encroachment has occurred, whether ventral, dorsal, or lateral; in any circumstance, if instability is present or will occur, stabilization becomes paramount.

Table 3 provides a summary of radiographic and neuroradiographic modalities for the detection of abnormalities in the cervical spine. These include plain lateral and anteroposterior radiography as well as dynamic flexion, extension, and lateral radiography. Segmentation failures, abnormal alignment, and fusion and instability can be documented. A predental space of less than 3 mm is considered normal between flexion and extension excursion in a patient older than 8 years of age. When younger, the limit is 5 mm. The interspinous distance between C-1 and C-2 is likewise important in recognizing instability at the craniocervical region. Abnormal dynamics in the relationship of the anterior arch of C-1, the odontoid process, and the clivus are considered pathological.

Computerized tomography, especially with high-resolution 3D reconstruction and its ability to inspect the craniocervical region, cervical spinal canal, and the cervicothoracic region from within, is necessary to identify the true pathological state. Static and dynamic (flexion–extension) MR imaging is necessary and may be combined with MR angiography and cerebrospinal fluid motion dynamics.

To identify the location of the VA and its components, angiography is initially performed, with CT angiography or MR angiography; however, the best definition is achieved using formal VA angiography for direct visualization.

In 1939, Karl List pointed out that numerous dislocations and abnormalities in cases involving abnormal craniocervical, cervical, and cervicothoracic alignment respond to cervical traction. The effects of cervical traction should be documented on plain radiography and MR imaging. This is considered to be a major factor in modulating treatment for cervical and craniocervical abnormalities. Case illustrations are presented to emphasize the sequence of imaging and the surgical–physiological approach to treatment.

**TABLE 1**

<table>
<thead>
<tr>
<th>Congenital Anomaly</th>
<th>Developmental Anomaly</th>
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<tr>
<td>proatlantal segmentation failure</td>
<td>basilar invagination</td>
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<td>basilar invagination</td>
<td>rotary dislocation</td>
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<tr>
<td>atlantal assimilation</td>
<td>os odontodeum</td>
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<tr>
<td>condylar hypoplasia</td>
<td>syndromic abnormalities</td>
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<tr>
<td>absent atlantal components</td>
<td>skeletal dysplasias</td>
</tr>
<tr>
<td>C-2-T spondylysis</td>
<td>Goldenhar</td>
</tr>
<tr>
<td>hemivertebra, segmentation failure</td>
<td>spondyloepiphysial</td>
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<td></td>
<td>Conradi</td>
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<td></td>
<td>osteogenesis imperfecta</td>
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**TABLE 2**

<table>
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<tr>
<th>Signs &amp; Symptoms</th>
<th>Acute</th>
<th>Chronic</th>
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<tbody>
<tr>
<td>quadriplegia</td>
<td>neck pain</td>
<td></td>
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<tr>
<td>respiratory failure</td>
<td>torticollis</td>
<td></td>
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<tr>
<td>limb weakness</td>
<td>failure to thrive</td>
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<td></td>
<td>repeated aspirations</td>
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<tr>
<td>swallowing abnormalities</td>
<td>vertigo</td>
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<tr>
<td>scoliosis</td>
<td>basilar migraine</td>
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<tr>
<td>tinnitus, vertigo</td>
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<tr>
<td>nystagmus</td>
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<tr>
<td>limb weakness &amp; paresthesias</td>
<td>bladder/bowel dysfunction</td>
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Illustrative Cases

Case 1

This 3-year-old child presented with neck pain and intermittent bilateral hand weakness. There was a history of renal and splenic agenesis as well as Sprengel deformity of the shoulder. On referral, a diagnosis of Klippel–Feil syndrome was established. Lateral flexion–extension radiography revealed instability between C-2 and C-3 VBs and blocked vertebra at C3–4 and C1–2 (Fig. 1 upper left and right). On 3D CT scanning block fusion was demonstrated between foramen magnum, the atlas, and the axis in an abnormal position (Fig. 1 center left and right). When obtained in the neutral position, MR images revealed no cervical cord compression (Fig. 1 lower). A C2–3 fusion was performed, and the child’s neurological deficits resolved. Instability in cases involving segmentation abnormalities is not uncommon and must be treated.

Case 2

This 15-year-old boy suffered progressive worsening of neck pain, was unable to turn his head to the right or left, and experienced arm weakness with numbness occurring when moving his head. Lateral radiography (Fig. 2A) demonstrated fusion from the C-1 anterior arch to the C-2 VB. Anterior CT scanning revealed abnormal segmentation of C-2 and C-1 (Fig. 2B). Segmentation failure was observed from C-1 to the lateral mass of C-2 on the right, and the C-1 anterior arch was incorporated into the C-2 VB (Fig. 2C). Flexion–extension MR imaging revealed abnormal motion where the ventral CMJ was indented by the osseous abnormality in the flexed position (Fig. 2D). The patient underwent a posterior occipitocervical fusion. It may be inferred from this case that dynamic/motion studies are essential for assessment.

Case 3

This 8-year-old boy experienced significant neck pain and exercise-induced upper-extremity weakness and presented with his neck jutting forward. Cervical radiography (Fig. 3 upper left) demonstrated lordosis of the upper cervical spine and a kyphotic change in the lower cervical region. Three-dimensional CT reconstructions (Fig. 3 upper left) demonstrated C3–4 spondylolysis with a defect at the pars (Fig. 3 upper right [arrow]). This was better defined on the internal view provided by the 3D CT reconstruction (Fig. 3 lower left), which demonstrated the bone separation as well as the VB abnormality. After C3–6 an-
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![Fig. 2. Case 2. A: Lateral cervical radiograph demonstrating lack of segmentation between the C-1 anterior arch and the C-2 VB. B: Composite CT reconstructions obtained through the CCJ and upper cervical spine at and behind the level of the odontoid process. The configuration of the fused atlantal lateral mass in relation to the axis on the right is abnormal. An abnormal articulation also exists between the occipital condyle and the lateral portions of C-2. C: Composite CT reconstructions obtained through the CCJ. The configuration of the posterior aspect of the axial VB is abnormal. This clivus articulates directly with the axis. D: Midsagittal T1-weighted MR image of the CCJ in the extended (left) and the flexed position (right). Note the ventral indentation at the craniovertebral border into the CMJ. Symptoms occurred when the patient’s neck was flexed.]

Case 4

This 11-year-old boy suffered an episode of quadriplegia after falling down while playing soccer. After a week of recovery, he remained paretic and had difficulty swallowing.

Magnetic resonance imaging demonstrated atlantal assimilation with severe basilar invagination and ventral compression of the CMJ. Additionally, a dorsal compression at the foramen magnum was caused by the suspected atlantal assimilation, with the posterior atlantal arch invaginating in a dorsal manner at the foramen magnum. This was confirmed on 2D CT reconstructions of the CCJ (Fig. 4 lower). After anterior resection of the clivus–odontoid complex and the anterior atlantal arch, the foramen magnum was decompressed and occipitocervical region fusion was performed via a posterior approach. Neurological deficits improved gradually. In this case anterior and posterior decompression of the CMJ was required to treat the anomaly.

Case 5

This 12-year-old boy presented with progressive spastic quadriplegia and difficulty swallowing. Midsagittal MR imaging (Fig. 5 left) revealed an anterior myelomeningocele and anterior disruption of the VBs in the midline at C-2 and C-3 (Fig. 5 center), which was better visualized on 3D CT reconstructions. A bifid atlantal anterior arch
was likewise visible, and the cervical kyphoscoliosis was quite evident with a dislocation between C-4 and C-5. Gentle cervical traction was recommended and followed by an anterior approach for release of the cervical cord. The anterior myelomeningocele was repaired, and C2–5 graft- and plate-assisted fusion was performed. Posterior fixation was also necessary.

**Case 6**

This 5-month-old child first presented with torticollis. Lateral cervical radiography demonstrated the absence of the C-1 anterior arch (Fig. 6 upper left). A 3D CT reconstruction (Fig. 6 upper right), obtained when the child was 9 months of age, revealed no anterior C-1 arch and a bifid posterior C-1 arch as well as outward displacement of the lateral atlantal masses. This child was treated with a custom-built cervical collar. Two years later at age 3 years, the patient’s anterior C-1 arch was developing and the posterior arch matured (Fig. 6 lower left). Cervical immobilization therapy was continued. Six months later the child’s head was held straight in a normal neutral position. A 3D CT reconstruction of the CCJ (Fig. 6 lower right) demonstrated that the anterior arch of C-1 had normal ring epiphysis and the occipitocervical alignment was normal. The lateral splaying of the lateral atlantal masses was not...
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Fig. 4. Case 4. Upper: Midsagittal T1-weighted MR image of the posterior fossa and upper cervical spine. The odontoid invagination into the foramen magnum occupies the anterior half of foramen magnum and indents the inferior medulla. The compression is compounded by an osseous abnormality compressing the dorsal CMJ. Lower: Midsagittal CT reconstruction obtained through the CCJ. There is assimilation of the C-1 anterior arch to the inferior aspect of the clivus. There is segmentation failure of C-2 and C-3 as well as severe invagination of the upwardly displaced odontoid process. The assimilated posterior arch of the atlas impinges on the spinal canal.

Fig. 5. Case 5. Left and Center: Midsagittal (left) and parasagittal (center) T1-weighted MR images of the posterior fossa and cervical spine. There is an anterior myelomeningocele that extrudes through the C2–4 VBs into the retropharyngeal space. Right: A 3D CT reconstruction demonstrating bifid anterior arch of C-1 and gross opening of the ventral bifid C2–4 VBs. A C4–5 dislocation can be visualized.

These findings demonstrate the growth potential if the CCJ is stabilized; 3D CT reconstructions were essential in the care of this child.

Case 7

This 12-month-old boy presented with torticollis and an absent right atlantal arch with a hypoplastic dens. He underwent cervical brace therapy until the age of 4 years. At such time 3D CT reconstructions (Fig. 7) demonstrated complete absence of the entire right lateral portion of C-1 (from the anterior midline to the posterior aspect). Marked neck deformity was exhibited. Posterior occipitocervical rib graft–augmented with fusion and halo vest immobilization were performed. Failure of formation of atlantal or axial vertebral component when a child is older than 3 years requires permanent stabilization.

Case 8

This 13-year-old girl with osteogenesis imperfecta presented with quadriplegia and a 3-year history of spasticity. Because she had previously undergone numerous joint replacements, MR imaging could not be conducted. Lateral cervical radiography demonstrated marked compression of the C-4 VB with a kyphoscoliosis and a rotary change between C-3 and C-6 (Fig. 8 upper left). This was further confirmed on 3D CT scanning (Fig. 8 upper right), which demonstrated the marked canal compromise between C-3 and C-5. Axial CT scanning revealed the thin ribbonlike space available for the cord between C-3 and C-5 (Fig. 8 lower left). Anterior C2–6 corpectomies were performed and followed by bone fusion and plate fixation. The procedure was undertaken with the patient in halo traction (Fig. 8 lower right). Arm function improved, but leg function improved less so. The axial CT scans appropriately revealed the need for anterior decompression of the spinal canal and stabilization.

Case 9

This 15-year-old boy presented with neck pain and occup-
ital headaches, and he suffered from repeated falls. A history of skeletal dysplasia was noted in siblings. A reducible atlantoaxial dislocation was demonstrated on lateral cervical spine radiography and 3D CT scanning demonstrated atlantal assimilation (Fig. 9 left); the CMJ compression was evident with early basilar invagination on MR imaging (Fig. 9 center). Crown halo cervical traction was undertaken to reduce the atlantoaxial dislocation. Atlantoaxial transarticular screw fixation was performed and supplemented with rib graft fixation (Fig. 9 right). Cervical traction–induced reduction of the dislocation was followed by placement of internal instrumentation and a bone graft construct.

Case 10

This 1-year-old boy presented with a small right arm and right hand, a Horner pupil on the right, and cervical myelopathy. At 2 years of age, cervical spine radiography demonstrated fusion of the C-2 and C-3 VBs. A gross abnormality of the C-4 and C-5 VBs was evident (Fig. 10A). The C-5 VB resided within the spinal canal and was not connected to the C-6 VB. The patient was placed in a cervical collar for immobilization. The 2D CT reconstructions revealed the abnormality of the C-4 and C-5 VBs within the canal itself (Fig. 10B). The family was reluctant for him to undergo surgery at this time. He subsequently underwent MR imaging (Fig. 10C). The C-5 VB on axial views appeared to fill the ventral aspect of the spinal cord, which conformed to it in a caplike fashion (Fig. 10D). At age 6 years, he underwent angiography.
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Fig. 9. Case 9. **Left:** Midsagittal 3D CT reconstruction of the CCJ and upper cervical spine viewed from within. There is assimilation of the anterior C-1 arch and the lateral mass. An atlantoaxial dislocation is evident; the diameter of the canal is grossly reduced. **Center:** Midsagittal T₁-weighted MR image revealing odontoid invagination into the foramen magnum and atlantoaxial dislocation compressing the ventral CMJ. **Right:** Postoperative lateral cervical radiograph demonstrating normal alignment at the cranio cervical border. Transarticular screws can be seen between C-2 and the C-1 anterior arch. An atlantoaxial interlaminar rib graft fusion is evident.

Fig. 10. A: Lateral flexion (left) and extension (right) radiographs demonstrating segmentation failure of the C-2 and C-3 VBs. The C-4 and C-5 VBs appear to occupy a position within the spinal canal and do not articulate with the superior aspect of C-6. B: Composite 2D CT reconstructions in the frontal (left) and the sagittal (right) planes. A fusion of C-2 and C-3 VBs is evident; the C-4 and C-5 VBs are dorsal to the C-6 VB and inside the spinal canal. C: Sagittal (left) and parasagittal (right) T₁-weighted MR images obtained in the neutral position. The apparent dislocation between C4–5 and C-6 VBs is evident. This is better seen in the parasagittal view (right). D: Axial Gd-enhanced MR image obtained through the C-5 VB, which appears to be hyperintense and lodged inside the spinal cord (which is wrapped around it). E: Selective lateral left VA angiogram revealing the serpiginous course of the vessel between the C-6 and C4–5 VBs. F: Axial (left) and coronal (right) CT angiograms obtained through the C-4, C-5, and C-6 VBs. The VA location is near the midline at C-5. G: Lateral (left) and anteroposterior (right) radiographs acquired 6 months after surgery. The C-5 and C-4 VBs have been resected and C3–6 strut graft fusion performed.
This demonstrated an absent right VA. The left VA had a circuitous course from C-6 to C-3, and there was an anomalous midline location at C-5 and C-4 (Fig. 10E). This was further confirmed on CT angiography. The dominant left VA was anomalous and located near midline at C-5 (Fig. 10F). Via an anterior approach the C-4 and C-5 VBs were resected and the spinal canal thus realigned. The VA position was confirmed to be near the midline. A C3–6 interbody fusion was performed (Fig. 10G). Postoperatively the cervical myelopathy resolved. The findings and results in this case demonstrate the need for complete imaging analysis to define the neurovascular abnormalities prior to surgical treatment. Sound knowledge of the VA anatomy allowed for appropriate surgical planning.

**Case 11**

This 11-year-old girl presented with occipital headaches, sleep apnea, repeated aspiration pneumonia, slurred speech, and quadriplegia. Midsagittal MR imaging (Fig. 11A) demonstrated a proatlantal segmentation abnormality and an osseous spur indenting the inferior medulla. A hindbrain herniation was also observed. Three-dimensional CT scanning demonstrated proatlantal segmentation failure and continuation of the clivus into the spinal canal at foramen magnum (Fig. 11B). An atlantal assimilation was also seen. Magnetic resonance imaging also demonstrated a cervicothoracic syrinx.

This patient underwent transpalatopharyngeal resection of the ventral clivus–odontoid osseous abnormality, thus decompressing the medulla, and a dorsal decompression of the posterior fossa and foramen magnum as well as occipitocervical fusion. Postoperative MR imaging (Fig. 11C) revealed ventral decompression, ascent of the cerebellar tonsils, and absence of the syrinx. She made an uneventful recovery.

![Fig. 11. A: Midsagittal T1-weighted MR image revealing extension of an osseous mass from the dorsal aspect of the clivus indenting the inferior medulla. A hindbrain herniation is evident. B: Midsagittal 3D CT reconstruction of the CCJ. Atlantal assimilation is present, and a proatlantal segmentation abnormality extends from the ventral inferior aspect of the clivus into the foramen magnum. C: Midsagittal T1- (left) and T2-weighted (right) MR images acquired 6 months after anterior resection of the abnormal craniocervical indentation into the medulla. The compression is now absent, and posterior occipitocervical fusion is evident. Note the fat pad present in the bone defect between the resected inferior clivus abnormality and the odontoid process. The previously recognized syrinx is absent.](image)

![Fig. 12. Treatment algorithm for management of cervical congenital anomalies.](image)
Treatment of Cervical Spine Congenital/Developmental Abnormalities

It is evident that precise identification of the pathological lesion(s) is essential. The findings and results in the cases described in this article demonstrate the necessity of neurodiagnostic studies and subsequent treatment. A surgical/physiological treatment algorithm is shown in Fig. 12. Reducible osseous abnormalities require primary stabilization. If this lesion is a ligamentous abnormality such as that seen after trauma or infection, then temporary brace therapy may be useful; however, a fusion procedure is mandated otherwise. In cases of irreducible lesions, the decompression should be performed via the direction in which the encroachment has occurred—that is, dorsal, ventral, or lateral. An appropriate operative procedure is tailored to the pathological entity and stabilization ensured by performing fusion.

References


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