Absence of the Posterior Element of C1 and C2 Along With Mild Wedge Shape in Body of C3: Case Report

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Abstract- Due to the wide variety of cervical vertebral congenital anomalies, knowledge of their types and related clinical conditions can prove to be helpful for physicians and surgeons in the diagnosis and treatment of patients. The aim of this study is to report on a rare case of unstable cervical vertebral congenital anomaly in which the patient had a defect in the body of the C3 vertebra in addition to defects in the posterior arch of C1 and posterior element of C2. This type of anomaly has not been reported so far in the literature. To reduce the patient's related neurological disorders, our treatment choice for this patient was occipitocervical fusion (OCF) surgery. At the 3-year postoperative follow-up, all sensory and motor problems related to this anomaly affecting the patient's upper and lower limbs and causing shoulder and neck pain were eliminated. In this rare type of anomaly, OCF surgery seems to be an effective choice in relieving the patient's neurological symptoms.

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Introduction

The development of vertebrae in intrauterine life is a complex biological process that follows the expression of specific genes as well as morphogenetic movements (1). The ossification of the vertebrae occurs at three primary centers: in each half of the vertebral arches and in the centrum (vertebral body). These centers appear in the vertebral arches in the ninth to tenth week of intrauterine life. These centers initially form in the lower cervical and upper thoracic regions and spread rapidly toward the upper cervical region (2,3). Congenital anomalies in the posterior arch of the vertebrae are extremely rare conditions and may be accidentally seen on radiological images of the neck. These anomalies may range from a minor defect to the absence of posterior arch formation. The prevalence of these types of anomalies is reported about 0.15% (4,5). Although it might be no specific symptoms in these patients, and sometimes they are diagnosed accidentally. Some of these patients complain of neck pain as well as neuropathy following a trauma (even minor trauma) (6). Radiographs, computed tomography (CT) scans, and magnetic resonance imaging (MRI) are needed to accurately diagnose all aspects of vertebral anomalies (7).

In this study, we report a rare case of unstable cervical vertebral congenital anomalies in which the patient had a defect in the body of the C3 vertebra in addition to defects in the posterior arch of C1 and posterior element of C2. To reduce the patient's related neurological disorders, our treatment of choice for this patient was occipitocervical fusion (OCF) surgery with a posterior approach. According to our research, this type of anomaly has not been reported so far in the literature.

Case Report

A 13-year-old girl with clinical manifestations, including frequent urination, neck pain, and difficulty in walking, was referred by a urologist to Erfan Hospital,

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Tehran, Iran, in 2018. According to the patient's statements, the strength of the upper limbs, especially the right hand, was reduced. The patient's right leg had difficulty in walking, so that it was dragged on the ground while walking (right hemiparesis). Many visits by various specialists did not lead to a correct diagnosis. According to the urologist, the patient had a urinary tract infection, so antibiotics were started for the patient. Her parents stated that frequent urination and sometimes enuresis had existed since childhood, but a severe decrease in the mobility of her right arm and leg had intensified over the past 3 years. Seven years ago, the patient had a history of seizures following a fall (mild trauma), and for this reason, the patient had a history of using phenobarbital for 1 year. The patient had been suffering from constipation for the past 3 months, and her frequent urination worsened. No pathological findings were observed on gastrointestinal examinations. Hemoglobin A1c was 6.2%. Awake electroencephalogram (EEG) and kidneys sonography were reported normal. In the patient's urodynamic report, decreased bladder capacity, increased sensation, excessive bladder muscle activity, high residual, and diagnosis of the neuropathic bladder were highlighted. According to the muscle strength grading index, the motor strength of the right upper limb was 3 and that of the left upper limb 4 (out of 5 points). The patient had a manifestation of an upper motor neuron. The Plantar reflex of the patient was normal (negative Babinski sign). Lasègue's test was negative. Plain radiographs, CT scan, and MRI of the brain, cervical, thoracic, and lumbar regions were performed. These images of the brain, thoracic, and lumbar regions showed no abnormal results. Plain radiographs (Figure 1), CT scan (Figure 2), and MRI (Figure 3) showed abnormality and instability in the cervical region. The results of these studies in the cervical region indicated the following:

- Posterior arch of C1 and posterior element of C2 were not formed. No fractures or dislocations were observed in the cervical vertebrae. Stenosis of the upper part of the cervical medullary canal was evident. Craniocervical junction stenosis was observed due to forward and upward subluxation of C2. The axis of the C2 was deviated posteriorly. Atlantodental interval (ADI) was increased (9 mm). No masses or lesions were observed in the neck. Pressure on the spinal cord was evident at the level of the foramen magnum to C2.
- Cervical vertebrae were normal in shape, interrelationships, and bone marrow signal intensity, except C3. The body of the C3 had a mild wedge-shaped deformity.
- Decreased cervical lordosis was observed. The space between the intervertebral discs in the neck was normal. The soft tissue around the cervical spine was normal.

The patient became a candidate for OCF. In the normal position of the neck (not extension) and using fiber optics, an endotracheal tube was inserted through the nose into the trachea for the patient. The patient was then placed in the prone position, and the head was fixed by Mayfield. The posterior incision of the neck was made from occiput to C4. Posterior elements of C1 and C2 were not observed during surgery. Then, in order to decompress the spinal cord in the area of the craniocervical junction, a small craniotomy similar to Chiari decompression surgery was performed near the foramen magnum. Then, the translaminar screws were inserted into the C3 laminae (Figure 4). A double-sided occipital plate was placed and fixed on both sides with translaminar screws using a rod; due to an anomaly in the body of C3, it was not possible to place the screw in lateral masses. Bone allografts were also used for better fusion (Figure 5). The final fixation using a fluoroscopic guide under a microscopic view was completed. Finally, incision layers were sutured. The postoperative patient evaluation was based on face-to-face interviews with the patient, her parents, and physical examinations. In the follow-up at 10 days after surgery, mobility of the arms was much more improved than legs. The paresthesia of the lower limbs was eliminated. During this time, the patient had pain in the neck and shoulders. In another follow-up at 30 days after surgery, the patient's pain was significantly reduced. Physiotherapy was performed on the shoulders, neck, and legs. No significant changes were reported in the next months. At a 3-year postoperative follow-up, the patient and her parents expressed great satisfaction with the surgery. All sensory and motor problems of the patient's upper and lower limbs were eliminated. His shoulder and neck pain had been eliminated. At a 3-year follow-up after surgery, according to the urologist's examinations, the patient's frequent urination was diagnosed due to decreased bladder capacity, which was slightly reduced but not completely resolved.

It should be noted that separate consent was obtained from the patient's parents at the preoperative time to use their child's medical information for research studies.

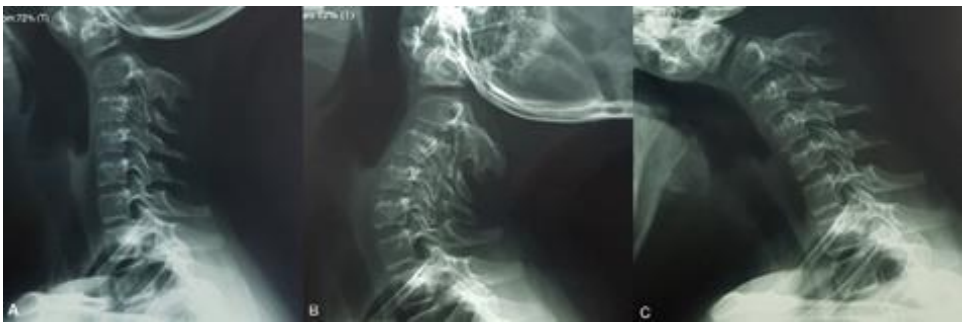

Figure 1. Lateral cervical radiographs showing the absence of the posterior element of atlas and axis vertebrae along with a mild wedge shape in the body of C3. (A) Neutral position. (B) Extension position. (C) Flexion position

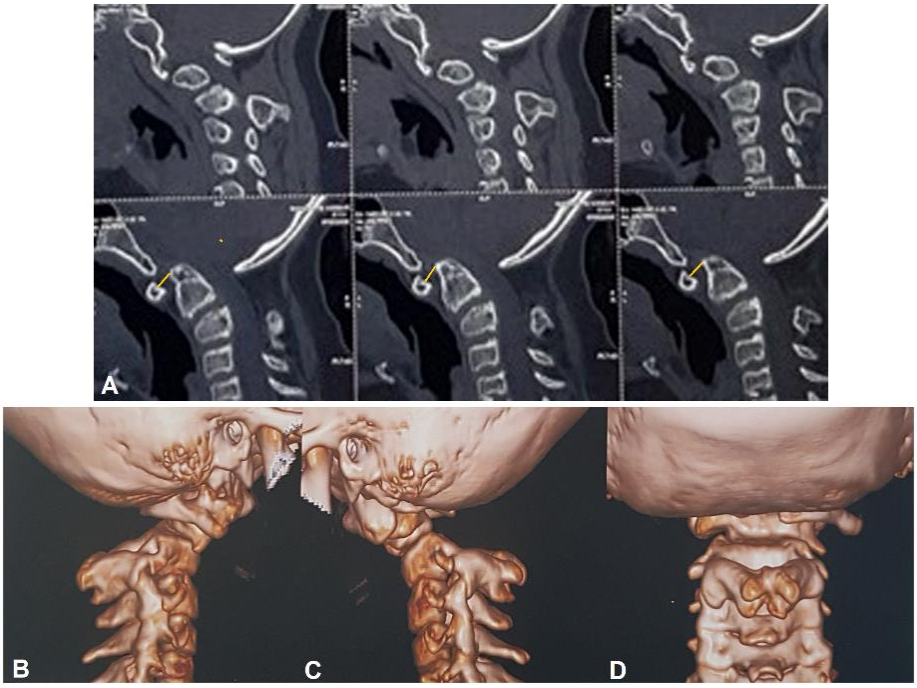

Figure 2. Computed tomography scans of the cervical region, showing the absence of the posterior element of atlas and axis vertebrae along with a mild wedge shape in the body of C3. (A) Sagittal views. The orange line indicates an increased atlantodental interval (ADI). (B) Right lateral view of three-dimensional scan. (C) Left lateral view of three-dimensional scan. (D) Posterior view of three-dimensional scan

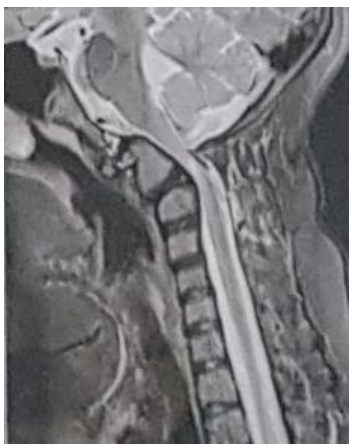

Figure 3. Sagittal view of the T2 MRI of the craniocervical region, which shows pressure on the spinal cord at the levels of foramen magnum to C2

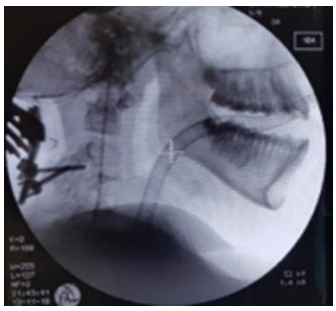

Figure 4. Intraoperative fluoroscopic image during the placement of translaminar screws on the C3 vertebra, with rods on either side of the vertebra

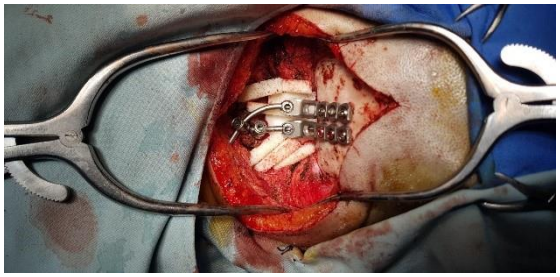

Figure 5. Photographic image of the final result of occipitocervical fusion surgery. Connection of the occipital plate on both sides; placement of two translaminar screws on the C3 vertebra; fixation by the rod on both sides; and using bone allografts to facilitate fusion

Discussion

The aim of this study is to report a rare case of an unstable cervical vertebral congenital anomaly with effective surgical results. At 4-12 weeks of intrauterine life, conditions, including vascular insufficiency, compressive forces, intrauterine trauma, systemic teratogens, and mutations, can cause axial skeletal anomalies (8). Developmental anomalies of the cervical vertebrae are very common (9). Most of these anomalies are asymptomatic and, in most cases, remain undiagnosed. Anomalies of the posterior element of vertebrae are a rare condition that can be the result of an abnormal change or an interruption during the developmental process (10). The craniocervical junction has a mesodermal origin and appears in the third week of intrauterine life. This mesodermal tissue transforms into structures called somites. The craniocervical junction is formed from four occipital somites and the first three cervical somites. Somites' cells are organized in the form of a donut around a small duct after an epithelialization process. By the beginning of the seventh week, the cells in the ventral and medial walls of each somite lose their epithelial structure and become mesenchymal again, migrating around the neural tube and notochord. These cells, which together make up the sclerotome, differentiate into vertebrae (3,10). The lateral zone of the first cervical sclerotome forms the posterior arch of C1, and the lateral zone of the second cervical sclerotome forms the arch of C2. The first ossification center in the C2 vertebra is formed in the body of this vertebra at the fourth month of intrauterine life (11-13). Cervical vertebral congenital anomalies are often associated with systemic anomalies of the head and neck, which should be evaluated by radiological images as soon as possible to reduce the risk of neurological disorders (8). Adisen *et al*., stated that the malocclusion classes and the dimensions of the airway do not have a direct effect on the etiology of craniocervical junction anomalies (14). On the other hand, Faruqui et al. stated that the incidence of C2 to C3 fusion in class III malocclusion is higher than in other classes (15). Also, posterior arch defects of the upper and middle cervical vertebrae can be associated with posterior arch defects of the thoracic vertebrae, in addition to myelopathy (16).

Some studies have reported the incidence of cervical vertebra congenital anomalies at about 1 in 40,000 births, with a higher incidence in girls (17). Some studies have reported the prevalence of posterior anomalies in cervical vertebrae of about 0.15% (4,5). Anomalies in the first six cervical vertebrae are more likely to occur than other vertebrae in the whole of the spine (18,19). In our report,

the anomaly was observed only in the first three cervical vertebrae. This anomaly included the absence of the posterior arch of C1, absence of the posterior element of C2, along with a mild wedge shape in the body of C3. Simultaneous occurrence of mild wedge-shaped deformity of the body of C3, with the absence of the posterior arch of C1 and posterior element of C2, was a significant case that had not been previously reported. Congenital anomalies of the posterior arch of the atlas are often asymptomatic and diagnosed accidentally (20). Congenital anomalies of the posterior arch of atlas are classified into five types: *type I*, failure of posterior midline fusion of the two hemiarches; *type II*, unilateral defect; *type III*, bilateral defects; *type IV*, absence of the posterior arch, with a persistent posterior tubercle; and *type V*, absence of the entire arch, including the tubercle (21). In the patient whom we studied, the defect of the posterior arch of atlas was type II, and the left half of the posterior arch was not formed. The prevalence of posterior arch of atlas defects is approximately 4% (22,23). Some studies have reported this rate to be 1.5%-5% (24). Any anomaly in the anatomy of atlas should be considered by physicians, surgeons, radiologists, and anatomists to avoid misinterpretation of the patient's anatomic structures and clinical complications (22). Anomalies of the posterior arch of atlas may be confused with atlas fractures. In a postmortem analysis of cadavers, Sanchis-Gimeno and colleagues concluded that it is difficult to distinguish between type II defect of the posterior arch of atlas and fractures (25). The occurrence of odontoid process anomalies is one of the most common cases of C2 anomalies. Defects in the formation of the posterior element of C2 are uncommon. These patients usually develop progressive myelopathy, and rarely neck pain (26,27). In our study, in addition to the absence of the posterior element of C2, this vertebra deviated slightly posteriorly, creating relative stenosis in the foramen magnum, which put pressure on the spinal cord, as was evident on the MRI image. The mild wedge shape in the vertebral body can be due to congenital anomalies or acquired causes. If this condition is seen on radiographs of an adult patient, there is a possibility of osteoporosis, which in these people could be a prelude to vertebral fractures (28). Due to the young age of the patient in our study, the mild wedge shape in the body of C3 seemed more likely to have congenital and evolutionary reasons. Previous studies established a direct relationship between mild wedge shape in upper lumbar vertebrae and the risk of intervertebral disc herniation in this region (29,30). This may be true for cervical vertebrae as well.

Despite the very low prevalence of cervical vertebral congenital anomalies, prompt diagnosis and treatment are very important for these patients because some cervical vertebral anomalies can cause nerve damage, biomechanical instability, nerve compression, or chronic pain (9). Symptoms such as neck pain, headache, or paralysis following vertebral dislocation may occur in cervical vertebral anomalies (31). The diagnosis of these anomalies in patients is mainly determined accidentally by radiological examinations for various reasons such as neck pain, the presence of neck masses, or neck trauma (4). Cervical vertebral anomalies could be detected using plain radiography, but a CT scan is needed to confirm the diagnosis. However, the use of MRI in patients with certain neurological defects to diagnose abnormalities in the spinal cord, intervertebral discs, and nerve roots is practical and can prove to be extremely useful. Therefore, the simultaneous use of plain radiography, CT scan, and MRI is necessary to confirm and accurately diagnose the status of cervical vertebral anomalies (32). In our study, we also used plain radiographic images, CT scan, and MRI to comprehensively examine the structures.

Occipitalization is the most common craniocervical junction anomaly with a prevalence of 0.08%-3% (33). This condition did not occur in our case. As a general rule, fusion and surgery are not required if the anomaly is small and does not impair stability (34). But, usually, in the presence of instability, the option of spinal fusion is explored. In our unstable case, OCF surgery was sufficient to maintain spinal stability. OCF is one of the most effective surgical methods for treating various pathological cases of craniocervical junction that may have been caused by congenital anomalies, trauma, degenerative factors, inflammatory factors, infections, or neoplasms. The most common indications for this surgery are compression myelopathies due to tumors or inflammatory lesions, basilar invagination, and instability in the occipito-atlanto axis (35,36). At 3-year follow-up, no significant problems were identified in terms of stability, and the patient's sensory and motor problems were resolved.

According to urological examinations, low bladder capacity was the cause of urinary frequency in the patient. In this study, we are not able to establish a definitive link between the presence of bladder problems and the development of cervical vertebral defects, but this link may be present because in some studies, such links have been reported (37,38).

Diagnosis of vertebral congenital anomalies is especially important for such patients due to the associated neurological risks and problems. Due to the wide variety of cervical vertebral congenital anomalies,

knowledge of their types and related clinical conditions can help physicians and surgeons in the diagnosis and treating of patients. This is especially important in anomalies that cause spinal instability. We reported a type of unstable cervical vertebral congenital anomaly that had not been reported before. This patient, with the absence of the posterior arch of C1 and posterior element of C2 and with a mild wedge shape in the body of C3, underwent OCF surgery. In this type of anomaly, OCF surgery seems to be an effective choice in relieving the patient's neurological symptoms.

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