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A Case Series of Fused Vertebrae and their Clinical Relevance

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ABSTRACT

Introduction: vertebral synostosis is a condition where in there is a fusion of vertebrae at various levels of vertebral column i.e. cervical, lumbar and thoracic in the order of frequency, which may be congenital or acquired. Various types of morphological anomalies, such as spinal synostosis, occipitalization, sacralization, are reported by researchers and these sequentially fused vertebral segments are referred as block vertebrae. The acquired fusion of vertebrae is secondary to trauma, tuberculosis or any other infections and may be due to juvenile rheumatoid arthritis. On the long-term, such segmental fusion can increase osteoarthritis at levels below and above the fused segment due to excessive wear and tear on these joints. These conditions may be asymptomatic or found incidentally or patients may present with back and radicular pain. Hence the comprehensive knowledge of these conditions among clinicians, surgeons, neurologists, neurosurgeons and orthopaedic surgeons are of utmost importance for the early diagnosis and to prevent long term complications.

Keywords: Blocked vertebrae; Congenital vertebral synostosis; Vertebral synostosis.

Introduction

The vertebral column is a curved linkage of individual bones or vertebrae and is also also called as backbone, spine, spinal column. The adult vertebral column usually consists of 33 vertebral segments. It extends from neck region to tail, as in vertebrate animals. In humans, it is named at different regions as cervical, thoracic, lumbar, sacral and coccyx. At the level of cervical there are 7 vertebrae, thoracic 12, lumbar 5, sacral 5 (all fused together to form 1 sacrum) and coccyx 1, composed of 3-5 vertebral segments[1].

At each region, these vertebrae are classified as typical and atypical. Each typical vertebrae consists of vertebral arch on the posterior aspect with a foramen, and a body in the anterior aspect. From the vertebral arch, two processes extend as two transverse processes and a single spinous process. It also has two superior and two inferior articular processes that make contact with the inferior and superior articular processes of neighbouring vertebrae, respectively[2].

Many researchers have reported a number of anatomical defects like occipitalization, sacralization, lumbarization, lack of the posterior vertebral arch, and spinal synostosis. Vertebral fusion at single or multiple levels is also referred to as block vertebrae or spinal fusion or vertebral synostosis.

The fusion of two or more vertebrae can be complete or partial, congenital or acquired. The congenitally fused vertebrae is due to failure of segmentation of sclerotomes at certain levels at the time of organogenesis, leading to Klippel-feil syndrome or other associated spinal deformities such as scolosis[3]. The acquired type of fusion of vertebrae is secondary to infections like tuberculosis or any other infections, due to trauma or may be due to rheumatoid arthritis[4]. Congenital fusion of vertebrae most commonly involves at the level of cervical regions[4,5].

Occipitalization represents the most cephalic 'blocked' vertebra encountered in the spinal column[6]. It is characterized by complete or partial fusion of the atlas to the base of the occipital bone[7]. Fusion of thoracic vertebrae, less frequent than cervical vertebrae, is due to ossification of anterior longitudinal ligaments [8]. Lumbosacral transitional vertebrae (LSTV) occur as a congenital anomaly in the segmentation of lumbosacral spine [9]. In LSTV, either the fifth lumbar vertebra may show assimilation to the sacrum called as sacralization of lumbar vertebrae or the first sacral vertebra may be fused with lumbar vertebra referred as lumbarization of sacral vertebra[10].

The surgical fusion of 2 vertebrae is known as spondylodesis or spondylosyndesis. The spinal fusion may lead to restricted movements, premature degenerative changes and neurological deficits. The symptoms varies according to the extent and level of vertebral fusion[5,11].

In the present study, we report a series of vertebral synostosis collected from the department of Anatomy of our college.

The comprehensive knowledge and awareness of the presence of skeletal abnormalities of spine are of great importance to the clinicians and surgeons in the planning of medical, surgical approaches and to avoid long term complications.

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Materials and methods

The study sample consists of four specimens of adult human dried vertebrae irrespective of age and sex, which were fused at different levels, obtained from the Departmental of Anatomy. These vertebrae are retrieved from bodies which were voluntarily donated for purpose of research and teaching. The specimen had intact bodies and vertebral arches. The specimens were subjected to detailed evaluation and digital vernier calliper was used to take measurements wherever possible, and measurements were recorded with up to two decimal places.

Case Series

Cervical synostosis: this is a case of fusion of two typical cervical vertebrae i.e. C4 and C5. Fusion was seen at the level of body and lamina. Anterior and posterior longitudinal ligaments were ossified. However the spinous process remained unfused. (Fig 1)

Thoracic synostosis: this is a case of extensive fusion of eleven thoracic vertebrae showing evident ossification of anterior longitudinal ligament. Fusion was evident at the level of anterolateral aspects of body. Intervertebral disc region and posterior aspects of body remained unfused. Articular process, transverse process and spinous process were unfused (fig 2a, 2b).

A case of fusion of fusion of seven thoracic vertebrae showing evident ossification of anterior longitudinal ligament. Anterolateral aspects of body, transverse processes and spinous process remain unfused (fig 2c).

Lumbar synostosis: this is a fusion of five lumbar vertebrae, all around the bodies, laminae, superior and inferior articular facets and spinous processes (Fig 3).

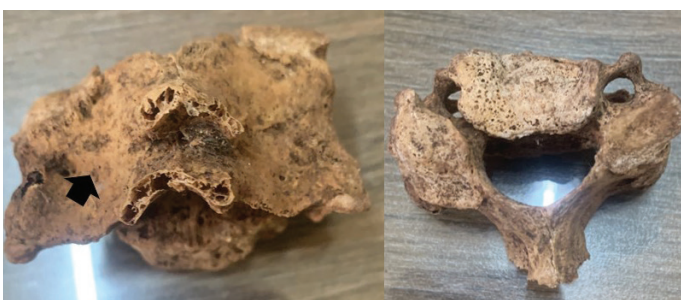

Figure 1. Posterior and superior view of fused cervical vertebrae.

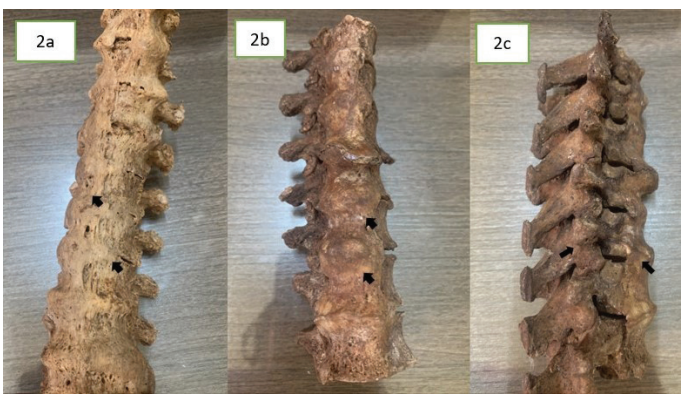

Figure 2. Lateral view of fused thoracic vertebrae.

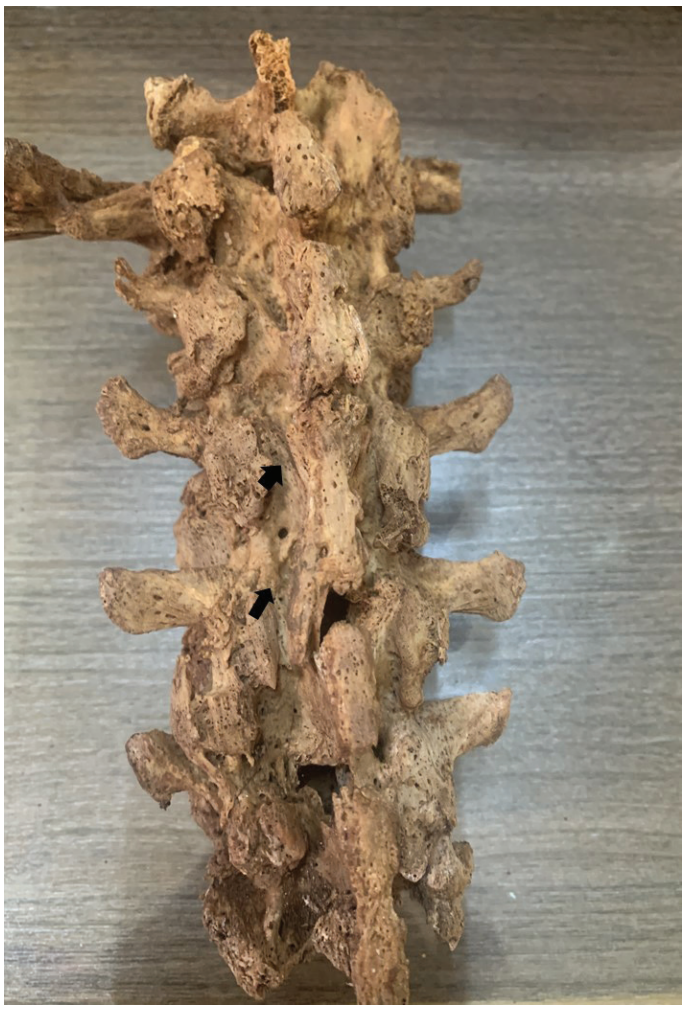

Figure 3. Posterior view of fused lumbar vertebrae.

Discussion

The vertebral or the spinal column begins to develop from the 3rd week of intrauterine life. It develops from sclerotome part of the somites, derived from paraxial mesoderm of intraembryonic mesoderm. Each somite comprises of a sclerotome and a derma-myotome which further can be subdivided into dermatome and a myotome. These somites initially begin in the cervical region and increase in number in the craniocaudal direction. During the 4th week of intrauterine life, there starts a migration of sclerotome cells around the neural tube and notochord. In due course, the sclerotome part of each somite undergoes a process called re-segmentation and leads to the formation of definitive vertebra. When the inferior half of each sclerotome develops and merges with the cephalic half of each subadjacent sclerotome, resegmentation takes place. Thus, the inferior half of one somite and the superior half of its neighbor combine to make each vertebra. The initial sclerotome segment's cephalic and caudal portions include mesenchyme cells that fill the gap between two precartilagenous vertebral centrums without proliferating. They assist in the development of intervertebral discs in this way[4, 12].

Normal segmentation of the sclerotomes is important for the development of a vertebral column. Vertebral fusion anomalies are likely to be associated with disturbance in Pax-1 gene expression in the developing vertebral column[13] and in certain cases may be because of decrease in blood supply during the 3rd - 8th week of intrauterine life[14,15]. So failure of embryological spines to segment normally may result in fused vertebrae or blocked vertebra. Hence it suggests that the causative factor may be due to combination of environmental and genetic factors[16].

Congenital fusion of vertebrae most commonly involves cervical region, followed by thoracic and lumbar regions[17]. The important ones being the fused cervical vertebrae (FCV). Congenital fusion of axis (C2) with the third cervical vertebrae limits the movements between these bones and hence the third vertebrae was given the name as "vertebrae critica" by Cave in the year 1937[18]. The sequence of block vertebrae according to frequency of occurrence is C3-C2, C6-C5, L5-L4, and any region of the thoracic spine[19]. C2-C3 is a frequent location for cervical vertebral fusion, with a frequency of 0.4% to 0.7% and no gender preference. Up to 70% of cervical vertebral synostosis has been linked to C2-C3 fusion with atlanto-axial articulation instability[5].

In FCV, the fusion may be either congenital or acquired type. Acquired FCV is generally associated with diseases like tuberculosis, other infections, juvenile rheumatoid arthritis and trauma 20. Congenital FCV is one of primary malformations of chorda dorsalis and are thought to be due to defects that take place during development of occipital and cervical somites[21].

Klippel-Feil syndrome (KFS) is characterized by a triad of congenital fusion of two or more cervical vertebrae presenting as short neck, limited mobility and low hairline at the back[22]. KFS can be classified into three types: Type 1 - 40% cases; Fusion of cervical and upper thoracic vertebrae with synostosis, Type II - 47% cases; Isolated cervical spine fusion and Type III - 13% cases; Cervical vertebrae with lower thoracic or upper vertebral fusion. Recently a new classification has been proposed: (Type I) single-level fusion, (Type II) multiple, noncontiguous fused segments and (Type III) multiple, contiguous fused segments[23].

The fusion of thoracic vertebrae can manifest with clinical signs like congenital scoliosis early in life, shortening of trunk with scoliosis or may be lordosis in older children. These fused vertebrae may cause restricted movements, premature degenerative changes and associated neurological deficits[24].

The incidence of fused thoracic vertebrae varied in literature from 1.6% to 4.16%. A study done on 48 adult dried vertebral columns, the incidence was found to be 4.16% in thoracic region[25]. M Nazeer *et al.* reported a case of fused three typical thoracic vertebrae featuring, bodies were completely fused on right side and partially fused on left side. The articular processes, laminae and spinous processes were unfused. The thoracic vertebral fusion is often seen in association with ossification of anterior longitudinal ligament in Diffuse Idiopathic Skeletal Hyperostosis (DISH), ankylosing spondylitis and osteochondritis[26].

Various other syndromes associated with vertebral fusion are VACTERL (s) which includes "Vertebral, Anal, Cardiovascular, and Tracheo –esophageal, Renal and Limb abnormalities", segmentation syndromes with laryngeal malformations, Jarcho-Levin syndrome, and Joubert syndrome. They may also be associated with aplasia of the mullerian duct, renal aplasia, somite dysplasia in the cervico thoracic region, trisomy 18 and diabetic embryopathy[22].

Lumbosacral transitional vertebrae (LSTV) is a congenital anomaly in the segmentation of lumbosacral spine[27]. In LSTV, the fifth lumbar vertebra may show assimilation to the sacrum known as sacralization of lumbar vertebra, or the first sacral vertebra may get fused with lumbar vertebra known as lumbarisation of sacral vertebra[28].

The thoracic or lumbar vertebral fusion is an autosomal recessive disorder[29]. Lumbosacral transitional vertebrae (LSTV) are most common congenital anomalies of the lumbosacral spine. Besides protecting the spinal cord and spinal nerves, lumbosacral spine plays an important role in posture and locomotion[30].

The origin of LSTV is unclear according to the literature found, but it is likely a product of both genetic predisposition i.e. Hox 10 gene mutation and developmental factors[31]. Wellik *et al.*, showed that in absence of Hox 11 function, sacral vertebrae are not formed and instead these vertebrae assume a lumbar identity. Again in absence of Hox 10 function, no lumbar vertebrae are formed. Thus these studies shows, the normal patterning of lumbar and sacral vertebrae as well as the changes in the axial pattern, such as Lumbosacral transition vertebrae, result from mutations in the Hox10 and Hox11 paralogous genes. LSTV can cause low back pain resulting from the pressure on nerve trunks, ligamentous strain or compression of soft tissues between bony joints[9]. The incidence of disc herniation is also found to be higher in LSTV[32].

The surrounding segments or structures are affected by bio-mechanical strain caused by the block vertebra, which causes early regressive changes at adjacent motion segments. These malformed vertebrae cause movement limitations, early degenerative changes, increased disc prolapse, difficulty in locomotion and accompanying neurological abnormalities.

Conclusion

The comprehensive knowledge and awareness help the clinicians, orthopaedicians and neurosurgeons in the diagnostic and therapeutic procedures. The thorough knowledge of these abnormalities is a prerequisite for a neurosurgeon to prevent complications during and after the spinal manipulations.

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