A SPECTRUM OF VERTEBRAL SYNOSTOSIS

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ABSTRACT

Vertebrae and their intervertebral discs are one of the chief manifestations of body segmentation or metamerism. The differentiation and resegmentation of vertebrae occur at the time of organogenesis. Thus resegmentation is the hallmark of vertebral development .Inappropriate vertebral fusion results in block vertebra or spinal fusion or vertebral synostosis. It can occur at various levels viz., cervical, lumbar and thoracicvertebral levels in the order of frequency. A study was conducted at Dr. B. R. Ambedkar medical college, with vertebral and skull specimens collected from 1st year MBBS students for duration of three years. Three different specimens of fused vertebrae were found; Assimilation of atlas, fused 6th and 7th cervical vertebrae and fused 1st and 2nd thoracic vertebrae. The clinical implications and embryological significance of these three specimens is discussed.

Key Words: Spinal Fusion, Vertebralsynostosis, Block Vertebra

INTRODUCTION

Vertebrae and intervertebral discs are one of the main manifestations of body segmentation or *metamerism* (Standring, 2008). The bodies of vertebrae, thus formed by metamerism can be fused partially or completely. Such fusion of vertebral bodies is called as vertebralsynostosis or Spinal fusion or Block vertebrae. Theaetiology of this abnormality can be congenital / acquired or surgical.

Congenital block vertebrae can result in syndrome manifestations such as KlippelFeil syndrome or may cause spine deformations such as Scoliosis. Acquired fusion of vertebrae is due to diseases such as Tuberculosis, Juvenile rheumatoid arthritis and trauma (Erdil *et al.*, 2003).

Presence of fused vertebrae results in biomechanical stress in the adjoining segments leading to premature degenerative changes at adjoining motion segments (Shankar, 2011).

METHODS

A study of two hundred and seventy vertebrae and one hundred and thirty six skulls was done for a period of three years (2009, 2010 and 2011). The vertebral and skull specimens were collected from students of 1st year MBBS of batch 2009, 2010 and 2011.Ethical committee clearance was obtained.

Inclusion criteria: All intact adult specimens were selected.

Exclusion criteria: Broken, neonatal skulls were excluded.

RESULTS AND DISCUSSIONs

In the present study, three different specimens of fused vertebrae were identified.

Assimilation of Atlas: Fusion of atlas with basiocciput.

Cervical Vertebral Synostosis: Fusion of cervical vertebrae at 6th and 7th level.

Thoracic Vertebral Synostosis: Fusion of thoracic vertebrae at 1st and 2nd level.

Assimilation of Atlas

The occipital condyles of basiocciput were replaced by inferior articular facet of atlas. A small triangular gap was present at the line of fusion of basiocciput and anterior arch of atlas.

Anterior arch and transverse process of atlas was well developed but posterior arch was totally absent. Theantero-posterior length of foramen magnum was 32 mm and transverse diameter was 22 mm. Area of foramen magnum was calculated by formula:

Area of foramen magnum = $\pi * \frac{1}{4} * w(22) * h(32) = 553.14 \text{ mm}^2$

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Where w= transverse diameter of foramen magnum and h = anteroposterior length of foramen magnum. The dimensions of foramen magnum were within normal limits thus osseous malformations causing neurological compression were ruled outSaini (2009).



Figure 1: Base of Skull with Assimilation of atlas, Figure 2: Close up view of assimilation of atlas, Figure 3: Anterobasal view showing gap between basiocciput and atlas. (Aa – Anterior arch of atlas, Ab Pa – Absence of posterior arch of atlas, Bo – Basiocciput, Fm – Foramen magnum, Ft – Foramen tranversarium, Iaf – Inferior articular facet, o – triangular gap between basiocciput and atlas, To – Torus occipitale)

Cervical Vertebral Synostosis

Their bodies and articular facets were fused but laminae and spinous processes were unfused. A definitive



Figure 1: Anterior view, Figure 2: Left lateral view, Figure 3: Right lateral view, Figure 4: Superior view of 6th and 7th cervical vertebral synostosis, Figure 5: Lateral Radiograph, Figure 6: Anteroposterior Radiograph of 6th and 7th cervical vertebral synostosis. (Bd – Bodies of fused vertebrae, Fj – Facet joint, Ft – Foramen tranversarium, La – lamina, Sp –Spine, Uafj – Unfused articular facet joint, W/ R- Waist / ridge at the site of fused bodies, *- Partial slit at the site of fused vertebral bodies)

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intervertebral foramen was present between these two fused vertebrae. The height of fused bodies of 6th and 7th vertebrae was 1.1 cm. Articular processes were partially fused on left side and unfused on right side. Anteriorly, bodies were completely fused represented by a waist or ridge. Internally, bodies were partially fused with a partial slit. Spine and laminae remainedunfused.Radiograph of the cervical and thoracic fused vertebral specimens were taken in anteroposterior and lateral views. Radiograph of fused cervical vertebrae showed hypoplastic intervertebral disc.

Thoracic Vertebral Synostosis

There was asymmetric fusion of 1st and 2nd thoracic vertebrae with the bodies and laminae fused more on left side. Their spinous processes remained unfused. Intervertebral foramen was larger on right side and smaller on left side. The costal facet was well defined at the junction of fused bodies on left side. Height of fused vertebral bodies measured 2.1cm on right lateral side, 0.8cm left lateral side. Intervertebral foramen was oval in shape on right side and triangular on left side. Articular facets were completely fused. A slit persisted on right lateral side of bodies indicating incomplete fusion. Lateral radiograph of fused thoracic spine revealed posterior remnant of intervertebral disc.



Figure 1: Superior view, Figure 2: Right lateral view, Figure 3: Left lateral view of 1^{st} and 2^{nd} thoracic vertebral synostosis, Figure 4: Lateral Radiograph, Figure 5: Anteroposterior Radiograph of 1^{st} and 2^{nd} thoracic vertebral synostosis. (Bd – Bodies of fused vertebrae, Fbd – Asymmetrically fused vertebral bodies, Fl- Fused laminae, If – Intervertebral foramen, La – lamina, Pd – pedicle, Sp – Spine, Ubd – Unfused vertebral bodies, T1- 1^{st} thoracic vertebra, T2 – 2^{nd} thoracic vertebra)

Assimilation of Atlas

It refers to abnormal cranial shifting of the occipitocervical border. Assimilation of atlas is the cranial most fused vertebrae. It has to be differentiated from Occipitilisation of atlas which is the caudal shifting of the atlas and Manifestation of the occipital vertebrae which are structures/parts of atlas seen around foramen magnum. Occipitilisation can be partial or complete fusion of atlas with the occipit. If atlas is assimilated, the occipital condyles are replaced by the inferior facets of the atlas Bergman (1993). There is a male preponderance of Assimilation of atlas upto 5:1 ratio with a high incidence of upto 0.75-3%. Partial assimilation of atlas is more common than total assimilation (Altunkas *et al.*, 2009). Based on the etiology, Swjetschnikow classified three types of *atlanto-occipital fusion*;

Acquired type due to Tuberculosis, Syphilis, Arthritis deformans etc.

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S. No.	Author	Incidence
1	James R.Grilliot(1988)	0.08%- 3%
2	Adel Afifi Bergman(1993)	0.5%-1%
3	VineetaSaini(2009), Kassim(,2010)	0.14%- 0.75%, 0.5- 1.5% in Caucasians
4	J.Skrzat(2010)	0.1%-0.8%, 1.09% in Asians

Acquired by fetus in utero by pelvic pressure as known as Basikyphosis of Virchow. Schniffner put forward theory called as DRUCK theory stating that "during delivery, pushing inwards of parts of occipital bone around foramen magnum results in Basikyphosis".

Purely congenital type occurring at the time of Sclerotome differentiation Smith (1907).

In the present case, there is also absence of posterior arch of atlas along with its tubercle which fits into type E. Other types of posterior arch defect of atlas (Kwon *et al.*, 2009) are:

- Failure of posterior midline fusion of the two hemiarches. Sometimes a small separate ossicle is seen within the gap.
- Unilateral cleft, ranging from a small gap to a complete absence of the hemiarch and posterior tubercle.
- Bilateral clefts of the lateral aspects of the arches with preservation of the most dorsal part of the arch.
- Complete absence of the posterior arch with a persistent isolated tubercle. This anomaly is relatively a more extensive form of bilateral clefts. The lateral parts of the posterior arch are absent except for the posterior tubercle.

Assimilation of atlas is commonly associated with type A posterior arch defect. It can be associated with clinical symptoms such as weakness and paresthesia of all four limbs, chronic neck pain and headache (Kwon *et al.*, 2009).

Clinical Significance

Assimilation of atlas may be coexistent with basilar invagination, protrusion of odontoid process into foramen magnum. Thus the size and configuration of foramen magnum must be evaluated to rule out accompanying osseous malformations. Decreased height of atlas at the time of fusion with occiput is the cause of basilar compression resulting in muscular weakness, ataxia, muscular spasticity and hypereflexia (Grilliot *et al.*, 1988). Neurological symptoms of assimilation of atlas resemble that of tumours in the region of foramen magnum, known as Foramen magnum syndrome. It may cause medullo-spinal compression ventrally or vertebrobasilarin sufficiency due to dorsal compression (Iwata *et al.*, 1998).

70% of patients with assimilation of the atlas will have congenital fusion of cervical vertebrae at the level of C2 - C4. It may also be associated with atlanto-occipital subluxation, hypertrophic odontoid process with brain stem compression (Grilliot *et al.*, 1988).

Recent technique to detect assimilation of atlas is Open mouth Tomography (Cone et al., 1981).

Developmentally, occipital condyles and articular facets of atlas, tip of odontoid process are derived from proatlas, the cranial portion of 4^{th} occipital sclerotomes. Thus failure of segmentation of the vertebral column results in the occurrence of assimilation of atlas (Cone *et al.*, 1981).Posterior arch defect of atlas is attributed to failure of development of cartilaginous preformation of atlas. The defect is also associated with syndromes such as KlippelFeil, Arnold Chiarimal formation, Turner and Down's syndromes (Kwon *et al.*, 2009).

Case of congenital vertebral fusion and assimilation of atlas to occipital bone is a further stage of evolutionary process. During transition from fish to amniote, many vertebrae become fused and assimilated to occipital bone. An addition of another vertebra to already absorbed cranium is a feature of amniote cranium. Thus Assimilation of atlas is a progressive modification of vertebrae whereas Occipitilisation of atlas is a regressive type of modification (Smith, 1907).

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Cervical and Thoracic Vertebral Synostosis

Spinal fusion is the hallmark of KlippelFeil syndrome, a triad of short neck, low posterior hair line and restricted neck mobility.

Maurice Kippel and Andre Feil, 1912 described three types of vertebral synostosis;

Massive fusion of cervical and upper thoracic vertebrae.

Fusion of interspace, hemivertebrae, occipitoatlanto fusions.

Cervical, thoracic and lumbar fusions (Samartis et al., 2006).

Wasp Waist sign (Anterior concave indentation at the site of absent or reduced inter space between fused vertebrae in a radiograph) is associated with complete vertebral fusion in KlippelFeil syndrome (Yuksel *et al.*, 2005).

Fusion of vertebrae can occur in sequence of common occurrence in cervical, lumbar and thoracic region. Surgical fusion of vertebrae is known as Spondylodesis or Spondylosyndesis. It can also be a congenital or acquired abnormality.

Cervical spine is invariably an anomalous constitution of clinical cases of Willet – Sprengel shoulder, brevicollis, kyphosis, congenital deafness, renal agenesis, cardiovascular abnormalities etc., (Cave, 1933-1934).

Radiologically, Johan *et al.*, (2007) described three types of vertebral fusion: Single fused cervical segment seen in 25% of patients, multiple, contiguous fused segments seen in 25% patients and multiple, non-contigious fused seen in 50% patients.

Sequence of presentation of vertebral synostosis is: C5-C6, C1-C2, C4-C5 followed by C6-C7fusion (Clarke *et al.*, 1995).Prevalence of vertebral synostosisin Lithuanian population is 2.6% of cervicalvertebralfusion, 1.6% of thoracicvertebralfusion and 0.5% of Lumbar vertebral fusion (Masnicova *et al.*, 2003).

Clinical Complications

Vertebral synostosis can be associated with radiculopathy and myelopathy. Various syndrome associations.

Table2: Complications associated with vertebral synostosis with their percentage of incidence (Batra *et al.*, 2008)

S. No	Associated Complications	Percentage of Incidence
1	Neural axis – Diastematomyelia, Tethered cord, Arnold Chiari malformation.	35%
2	Renal – Unilateral horseshoe kidney, Duplicated kidney/ ureters, Hypospadias.	25%-33%
3	Congenital heart disease – Atrial septal defect, Ventricular septal defect, Tetralogy of Fallot, Transposition of great vessels.	10%
4	Musculoskeletal - Clubfeet, Sprengel's deformity, KlippelFeil syndrome Dysplasia of hip, scoliosis	30% 71% 60%
5	Jaw, external ear abnormalities, cleft palate, cervical ribs	20%

of vertebral fusion are Segmentation syndrome1 with laryngeal malformation, VACTERL(S)(Vertebral, Anal, Cardiovascular, Tracheo- Esophageal, Renal and Limb abnormalities+_single umbilical artery), MURC (Mullerian duct aplasia, Renal aplasia, Cervicothoracic somite dysplasia), Hemifacialmacrosomia, Diabetic embryopathy, Alagille, Trisomy 18, Escobar(Autosomal recessive cervical vertebral fusion,

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ptosis, hypertelorism of neck, axillae, genital anomalies and small stature), Joubert, Jarcho - Levin syndrome (Victor, 2010).

Pathological causes of fusion of vertebrae are Fibrodysplasia, Progressive Juvenile Rheumatoid arthritis, Postinfectious, Postsurgical, Ossification of posterior longitudinal ligament of cervical spine, Posttraumatic etc. (Clarke *et al.*, 1995)

The differentiation and resegmentation of vertebrae occurs at the time of organogenesis. This explains the association of vertebral synostosis with cardiac, renal, musculoskeletal and neural abnormalities (Letts*et al.*, 2009).

Embryological Significance

Fused vertebrae or block vertebrae are caused by non-segmentation of the primitive Sclerotome. The anatomy of fused vertebrae indicates the embryological time of occurrence of synostosis: Independent pedicle and transverse process suggests normal initial development followed by fusion.

Ridge on dorsal surface of fused arches indicates that fusion has occurred after initial development and differentiation.

Absence of the joint between articular facets in the fused vertebrae suggests failure of normal development and differentiation of vertebraei.e.Fusion at the precartilaginous stage of vertebral development (Chandraraj *et al.*, 1987).

The sum of height of congenitally fused vertebral bodies is equal to normal height of intervertebral disc if one were present whereas vertebral fusion due to disease has sum of height of fused bodies is less than normal height of intervertebral disc if one were present (Kumar *et al.*, 1988).

Molecular basis

Familial KlippelFeil syndrome is on locus of Chromosome 8q.An inversion of inv(8)(q22.2 q23.3)has been found on 8q chromosome segregating with congenital vertebral fusion(Clarke *et al.*, 1995). Mutation occurs in Pax gene and notch signaling pathway . C2-C3 fusion is an autosomal dominant disorder and C5- C6 fusion is an autosomal recessive disorder.

*Morphology*Addition of vertebra to existing cranium is a feature of progressive variation.

CONCLUSIONS

Block vertebra result due to failure of resegmentation of vertebrae. It is associated with genitourinary, neurological and musculoskeletal abnormalities. Fusion of vertebrae is named as KlippelFeil syndrome in cervical region, Synspondylism in thoracic region, or Block vertebra in lumbar region. Though unrelated, each of these cases is unique due to varied clinical presentations.

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