Anomalies of the Craniovertebral junction - a rare case report
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Abstract: A rare case of anomalies of the craniovertebral junction (CVJ) is reported owing to its clinical significance and rarity. A 15 year old male presented with cervical compressive myelopathy whose MRI revealed Hypoplasia of dens and atlanto-axial dislocation. The treatment goals for persons with atlanto-axial instability (AAI) are to protect the spinal cord, stabilize the spinal column, decompress neural tissue, and reduce any deformity. He was treated surgically with lateral mass screw fixation and he is improving post operatively.

Keyword: craniovertebral junction - hypoplasia of dens - atlanto-axial instability - lateral mass screw fixation

CASE REPORT:
A 15 year old male admitted with complaints of neck pain, weakness of all four limbs and numbness below the level of neck of 5 months duration & difficulty in breathing for 15 days. Complaints started as neck pain radiating to right upper limb, followed by insidious and progressive weakness of right upper limb. It was followed by weakness of right lower limb, left lower limb and left upper limb in succession (U pattern). History suggestive of both proximal & distal muscle weakness. History of decreased sensation of all modalities below the level of neck. History of trivial trauma to neck 5 months back & 15 days back. Complaints aggravated for 15 days with breathing difficulty. No history of thinning of limbs / involuntairy movements / LOC. No history suggestive of cranial nerve palsy. No history of bowel or bladder disturbance. No history of electrical injury / irradiation / vaccination. No history of fever / respiratory tract infection. No history of similar illness in the past. No history of multiple joint pain or deformities. No history of similar illness in the family. Born out of non consanguinous marriage. No history of mental retardation. Attained all developmental milestones normally.

ON EXAMINATION:
conscious & oriented, afebrile, dyspnoeic
no pallor/ icterus/ cyanosis/ clubbing/ generalised lymphadenopathy/ pedal edema
no neurocutaneous markers or congenital anomalies
no markers of HIV/ TB/ Syphilis no peripheral nerve thickening,

no spine angulation
height / neck ratio = 10.3; no low hair line / webbing of neck
Pulse = 82/min, regular BP = 110/70 mmHg RR = 28/min, regular, thoracoabdominal

CNS EXAMINATION:
• Higher mental functions = normal
• Examination of cranial nerves : no cranial nerve palsy
• Examination of Motor system:
  • Bulk= no wasting / atrophy
  • Tone = hypertonia in all 4 limbs
  • Power = both upper limbs : 4/5 both lower limbs : 4/5
  • Reflexes:
    • Superficial – corneal, conjunctival, palatal, pharyngeal, abdominal & cremasteric reflexes – Present
    • Plantar – Bilaterally extensor
    • Deep – Jaw jerk-not present
Biceps, Triceps, Supinato, Knee jerk & Ankle jerk – Brisk bilaterally
No patellar / ankle clonus
• coordination – normal; no involuntary movements
• gait – Romberg’s sign positive
• Examination of Sensory system:
  • Superficial – pain, touch & temperature – impaired below C3 dermatome
  • Deep – pressure, deep pain, position sense impaired. Vibration sense decreased below C3 level
  • cortical sensation – could not be assessed
• Romberg’s sign – positive
• No signs of meningeal irritation; No autonomic dysfunction
• Skull – normal; No kyphoscoliosis / gibbus
• No peripheral nerve thickening; no trophic changes
• no bruit over orbit, carotid, skull or vertebra
• Other system examination:
  • Cardiovascular system; S1 S2 heard, no murmurs
  • Respiratory System = Vesicular breath sounds heard bilaterally, tachypnoea+, no added sounds
  • Abdomen = no organomegaly

INVESTIGATIONS DONE:
• B. sugar = 92 mg/dl B. urea = 28 mg/dl S. creatinine = 0.7 mg/dl S. Na+ = 136 mEq/L S. K+ = 3.6 mEq/L
• Hb= 10.2 g%, TC= 8200/cu.mm, DC= P76L22E2, Platelets=1.2 Lakhs /cu.mm
• ESR = 10 mm in 1st hour; RA Factor = negative
• Urine = albumin-nil, sugar-nil, deposits= 1-2 pus cells
• ECG= sinus rhythm; ECHO= normal study

MRI cervical spine: Compressive myelopathy, cord compression & narrowing at spinomedullary junction. AP diameter 3 mm. Hypoplasia of dens. Atlanto-axial dislocation. Narrow & steep posterior fossa

MRI

DIAGNOSIS:
CRANIOTOVERTEBRAL JUNCTION ABNORMALITY - HYPOPLASIA OF DENS, ATLANTO AXIAL DISLOCATION. CERVICAL COMPRESSIVE MYELOPATHY

Treatment & Follow up:
Patient was treated initially with supportive measures like cervical immobilization with cervical collar. Later he was taken up for Lateral mass screw fixation surgery by Neurosurgery department. Perioperative and post operative period was uneventful. He was followed up and showing signs of improvement.

DISCUSSION:
The term craniovertebral junction refers to the occipital bone that surrounds the foramen magnum and the atlas and axis vertebrae. A wide variety of congenital, developmental, and acquired abnormalities can occur at the craniovertebral junction. There may be single or multiple abnormalities in the same individual. Once the stage is set by congenital cv anomalies and produce atlantoaxial instability & subsequently basilar invagination. This is much more common in developing countries where heavy loads are carried on the head from childhood.

The signs and symptoms of CVJ abnormalities are varied, typically begin insidiously, and arise fairly late, progress slowly, remain stationary and rarely relapse. Congential and developmental osseous abnormalities and anomalies affecting the craniovertebral junction complex can result in neural compression and vascular compromise and may manifest with abnormal cerebrospinal fluid dynamics. In patients with the myriad anomalies at CVJ, the natural history is not understood clearly.

many patients are asymptomatic and probably remain so all their lives. Clinical manifestations develop only with advanced age or after a traumatic event.

The causes of Atlanto-axial instability (AAI) are varied. AAI sometimes results from trauma. Other cases occur secondary to an upper respiratory infection or infection following head and neck surgery. Another cause is rheumatoid arthritis (RA), with its predilection for the upper cervical spine. In addition, congenital anomalies, syndromes, or metabolic diseases can increase the risk of AAI. AAI is defined as an atlantodens interval (ADI, distance between odontoid process and the posterior border of the anterior arch of the atlas) of greater than 3 mm in adults and of greater than 5mm in children as measured on plain radiography.

AAI is very rare in patients without predisposing factors. No data exist regarding prevalence.

CLASSIFICATION OF CRANIOCERVICAL ABNORMALITIES:
Congenital anomalies & malformations of cv junction:
Maformations of Occipital bone-
§ Manifestations of occipital vertebra
§ Basilar invagination
§ Condylar hypoplasia
§ Assimilation of atlas
Malformations of Atlas – Assimilation of atlas; Atlantoaxial fusion; Aplasia of atlas arches
Malformations of Axis – Irregular atlantoaxial segmentation; Dens dysplasias; Ossiculum terminale persistens; Os odontoideum; Hypoplasia – aplasia
§ Segmentation failure of C2 / C3
Developmental & acquired abnormalities of cv junction:
Abnormalities of Foramen magnum
§ Secondary basilar invagination (eg., Paget’s disease, osteomalacia, rheumatoid cranial settling, renal & resistant nicks)
§ Foraminal stenosis (eg., achondroplasia)
Atlantoaxial instability
§ errors of metabolism (eg, Morquio’s syndrome)
§ Down syndrome
§ Infections (eg, Grisel’s syndrome)
§ Inflammatory (eg, rheumatoid arthritis)
§ Traumatic occipitoatlantal & atlantoaxial dislocation, os odontoideum
§ Tumours (eg, neurofibromas, syringomyelia)
§ Miscellaneous (eg, fetal warfarin syndrome, Conrad’s syndrome)

SIGNS & SYMPTOMS :
Head tilt
Short neck, low hairline, limitation of neck motion
Webbed neck
Scoliosis; Features of skeletal dysplasias
Neck pain & posterior occipital headache
Basilar migraine
Hand or foot isolated weakness
Quadriparesis / paraparesis / monoparesis
Sensory abnormalities
Nystagmus – usually downbeat & lateral gaze
Sleep apnoea
Repeated aspiration pneumonia, dysphagia
Tinnitus & hearing loss; Vertigo
Most frequent symptom is neck pain originating in the suboccipital region with radiation to the cranial vertex.

Most common neurological deficit encountered in affected children is myelopathy.

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NEURODIAGNOSTIC IMAGING:
• Diagnostic procedure of choice is MRI, but plain cervical spine radiographs with lateral flexion & extension views are essential to gain a better understanding of the biomechanics.
• MRI is used to identify neural abnormalities & osseous compression, as well as to recognize any instability if present.
• Magnetic resonance angiography & CT angiography can provide better understanding of the vasculature, as well as any vascular deformities that may occur with motion of cv junction.
• 3D CT is useful for defining abnormality, as well as for facilitating coronal & sagittal sectioning.
• Numerous craniofmetric reference lines were described before the advent of CT & MRI. They were used to locate the position of neural structures with the associated osseous abnormality.

TREATMENT:
• The treatment goals for persons with AAI are to protect the spinal cord, stabilize the spinal column, decompress neural tissue, and reduce any deformity. It has been demonstrated that surgery is unlikely to reverse clinical symptoms when the spinal cord is compressed by more than 60%.

LATERAL MASS SCREW FIXATION:
Jurgen Harms technique of C1 C2 fixation. Can be used in patients with compromised posterior element. Easier than placement of transarticular screws. Posterior plating utilizing lateral mass screw fixation has been widely accepted for treating the unstable cervical spine caused by trauma, neoplasms, significant degenerative conditions, and failed anterior fusions. Clinical studies have shown that posterior cervical plating results in a high rate of fusion.

References:
• Adam's Text book of Neurology
• Youmann's Text book of Neuroradiology