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Imaging of Developmental Malformations Affecting Craniocervical Junction

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ABSTRACT

Background Craniovertebral anomalies assessment is challenging and reaching the accurate diagnosis is the corner stone for appropriate management.

Objective: The aim of the study was to demonstrate various imaging features of the developmental anomalies affecting cranio-cervical junction (CCJ) by using CT scan and MRI ~~scans~~.

Patients and methods: In the Radiology Departments in Mansoura University and Mansoura University Children's Hospital, 30 patients (16 males and 14 females) participated in this study at different ages with history of CCJ abnormalities by at least one imaging modality. Those patients underwent careful examination of the previous imaging scan. Some of the patients were fully diagnosed using MRI only or CT scan. Analysis was performed on the MDCT and MRI examination.

Results: Arnold Chiari malformation (ACM) type 1 was the most common diagnosis followed by ACM type 2, tonsillar herniation in 3 cases, MPS, achondroplasia in 2 cases while split atlas, platypesia, Down syndrome and Os odenoottidum in 1 case each.

Conclusion: Careful radiological examination of the CCJ and understanding of its complicated anatomy is required in order to reach the proper diagnosis of its lesions.

Keywords: Craniovertebral junction, Developmental anomalies, CT, MRI, Mansoura University.

INTRODUCTION

The craniocervical (craniovertebral) junction represents the complex transitional zone between the cranium and the spine and comprises a complex balance of different elements including osseous structures articulated with synovial joints, intrinsic ligaments and membranes and muscles [1].

Evaluation and assessment of craniocervical anomalies is challenging and coming to accurate diagnosis plays a crucial role in the management. Congenital, developmental and acquired anomalies cause atlanto-axial instability or it can affect the vertebrobasilar vascular system [2].

Computed tomography scan (CT) and magnetic resonance imaging (MRI) have improved their technological capability, offering a high quality 3-dimensional visualization of the craniovertebral junction. CT evaluates bones Very well, but MRI is absolute necessary in joint, ligament, vascular and soft tissue evaluation [3]. This study aimed to demonstrate various imaging features of the developmental anomalies affecting craniocervical junction by using CT scan and MRI examination.

PATIENTS AND METHODS

This retrospective study was carried out through the period from July 2020 till August 2023 in Mansoura University Hospital and Mansoura University Children's Hospital, Radiology Department. 30 Patients were enrolled in this study (16 males and 14 females). The patients' age ranged from the first day of life up to the age of 18 years. They were referred to Radiology Department from Genetics Clinic in Children's Hospital, Mansoura University and Neurosurgery Department, Mansoura University Hospital.

Inclusion criteria: Both sexes. Patients with accidentally discovered lesion in cranio-cervical junction region by at least one imaging modality.

Exclusion criteria: 1) Patients with contraindication for CT scan (Patients with known allergy to sedation. Patients with movement disorder that cannot be controlled by sedation). 2) General contraindication for MRI scan [(General contraindications as previously mentioned in CT scan, patients with significant deformity not fit for MRI machine (un fit for examination by head coil and the gantry), claustrophobic patients and patients who had artificial implants (cardiac implants, cochlear implants, etc.)].

Techniques and methods:

- **Computed tomography:**
  MDCT is the primary modality used for screening patients with suspected carnio-cervical bony anomalies. All scans are performed on a 128 multi-detector, 128 channel CT scanner GE systems). Cervical spine scan is obtained at 0.625 mm slice thickness with axial, coronal, sagittal reformats and 3D reconstruction.

- **Magnetic resonance image:**
  Images are acquired using a 1.5 T magnet Philips system with 16 channels. The Cervical spine protocol includes sagittal T1WI, T2WI, STIR and axial T1WI, T2WI and 3D GRE sequences. Sagittal images were obtained using FOV 20cm, slice thickness 3mm with 0.3mm slice gap, while axial images were obtained using 18cm FOV, slice thickness 3mm with slice gap 1mm. **Sedation** [chloral hydrate 5% was used (0.5mg/kg single dose)].

All patients underwent the following:

(1) Thorough history taking [The main complaint with

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analysis including (onset, duration, etc.)].
(2) Clinical examination.
(3) Reviewing any previous radiological studies.
(4) MRI examination and CT scan was done as follows: Both MRI & CT scans were done for 20 cases. CT scan wasn't performed in 8 cases because MRI was diagnostic and further CT scan was considered to add no value to the patient with undue radiation dose. MRI wasn't performed in 10 cases, which were examined in the main hospital, because they were not adequately sedated by chloral hydrate (pediatric general anesthesia was available only in the children hospital) and CT scan was considered diagnostic. Those investigations were done using: a. MRI was done by using Philips Ingenia 1.5 Tesla device (Philips Medical Systems) using head and neck surface coil. b. CT scan was done using 128-multidetector CT Philips Medical system.

Imaging interpretation:
Interpretation of MRI:
Detection of any abnormality in craniocervical junction. Identification of the anomaly as well as its impact on spinal cord.

Interpretation of MDCT: Detection of any bony abnormalities in the cranio-cervical region.

CASES

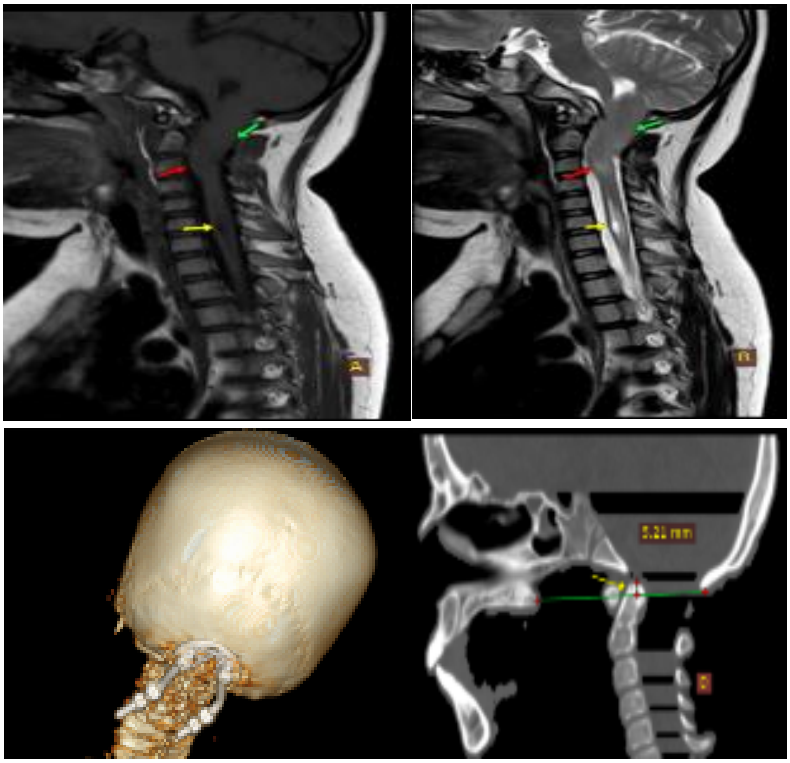

Figure (1): 11 years old male with mental retardation and disturbed gate. Non contrast MRI of the cervical spine in sagittal cuts (T1 (A) & T2 (B) weighted images) showed downward herniation of the cerebellar tonsils (green arrow), spinal cord edema (red arrow) and syrinx [low SI in T1 & high SI in T2 (yellow arrow)] opposite C5, 6 & 7vertebral level. Non contrast CT scan sagittal cut showed elevation of the odontoid process (yellow arrow) above Chamberlain line by 5.21 mm (Normal up to5mm). 3D reconstruction image showed post-operative decompression surgery of the skull base. Diagnosis: Arnold Chiari malformation with type 1 basilar invagination underwent decompression surgery of skull base.

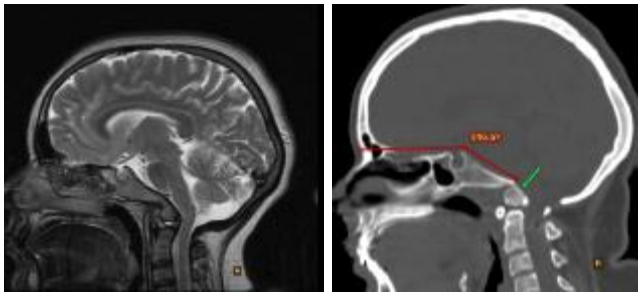

Figure (2): 17 years old female with migraine. Non contrast MRI of the brain sagittal cut (A) in T2 weighted image revealed abnormal flatting of the skull base with kinking of the spinal cord. Non contrast CT scan of the brain and cervical spine (B) showed increased Welcher basal angle (146 ^{o}). Green arrow: OS odontidum. Diagnosis: Platybasia & Os odentidum.

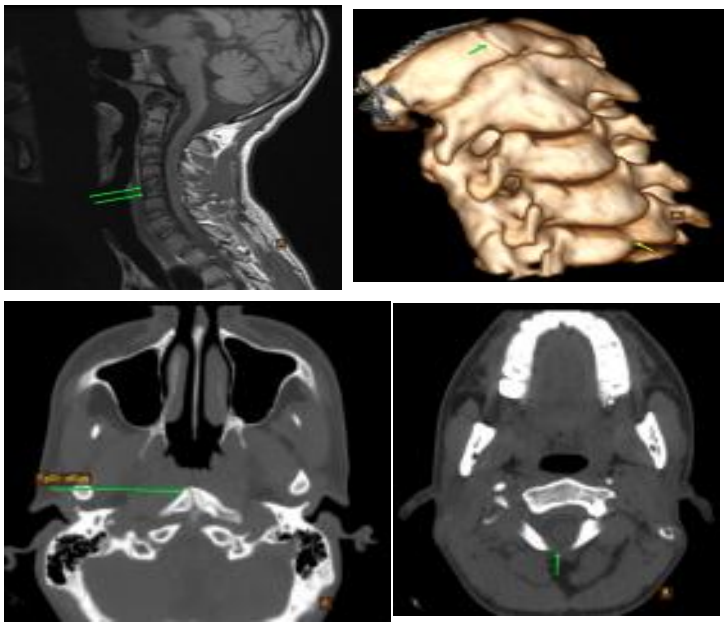

Figure (3): 12 years old male with cervical pain. MRI sagittal T1WI (Green arrow) showed a normal shape f C5 vertebral body. 3D image showed split atlas (green arrow) and butterfly vertebra (yellow arrow). Non contrast CT of the cervical spine in axial (A & b) images showed non fusion of the anterior and posterior arch of atlas vertebrae (green arrow). Diagnosis: Split atlas associated with non-fusion C5 vertebral body (butterfly vertebrae).

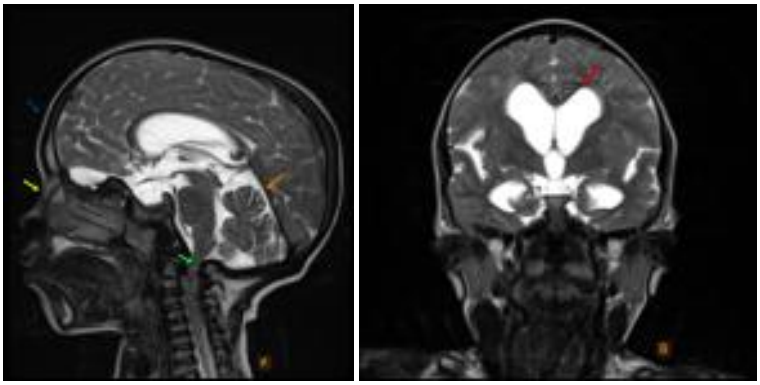

Figure (4): 11 years old male ~~boy~~ of achondroplasia presented with hydrocephalus. Non-contrast MRI of the brain in T2 weighted image sagittal (A), coronal (B) revealed abnormal shape of the skull vault (blue arrow), depressed nasal bridge (yellow arrow), vertical straight sinus (orange arrow), cervico-medullary kink with stenosis of foramen magnum (green arrow). Symmetrical dilatation of the lateral ventricle (red arrow).

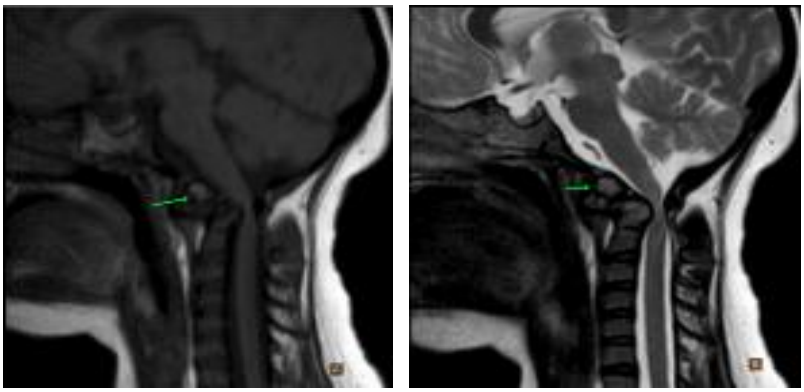

Figure (5): 11 years old female case of Down syndrome presented with spasticity in both lower limbs, hypertonia and hyperreflexia in both LLs. Non-contrast MRI scan of the cervical spine sagittal cuts on T1 (A) and T2 (B) weighted images reveled abnormal bone present in front of the odontoid process (green arrow). It was seen compressing the spinal cord associated with spinal cord edema (low T1 and high T2 signal intensity). Diagnosis: os odontiduim.

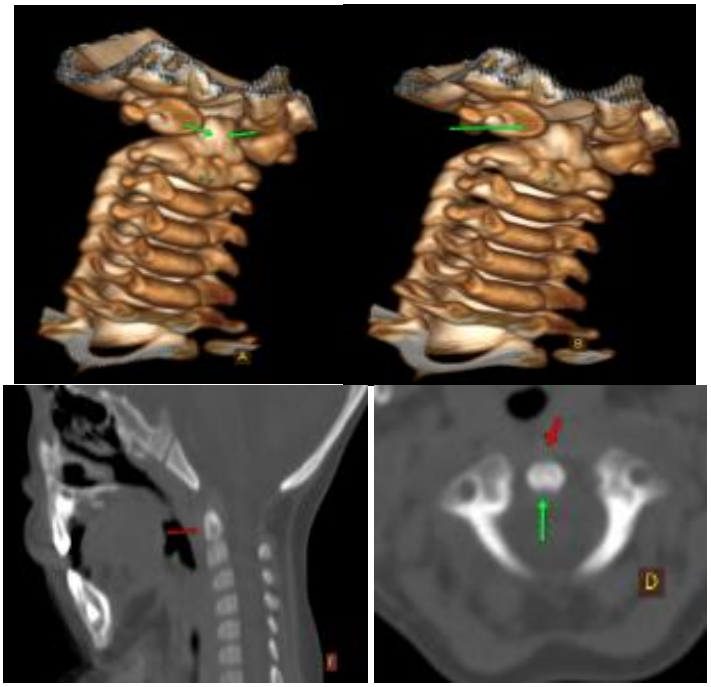

Figure (6): 1 month female patient presented with congenital hydrocephalus underwent VP shunt. 3D reconstruction images revealed non-visualization of the anterior arch of atlas vertebra. (A) Two green arrows pointing to the odontoid process. (B) Green arrow pointing to the anatomical site of the anterior arch. Non-contrast CT scan of the cervical spine sagittal (C), axial (D) images revealed non-visualization of the anterior arch of atlas vertebra. Red arrow pointed to the anatomical site of the anterior arch, while the green one pointed towards the odontoid process. Diagnosis: Absent anterior arch of atlas vertebra.

Ethical approval: The Ethics Committee of Faculty of Medicine, Mansoura University granted the study approval. All participants signed informed consents after a thorough explanation of the goals of the study. The Helsinki Declaration was followed throughout the study's conduct.

Statistical analysis

Results were statistically analyzed using statistical package of social sciences (SPSS 26, IBM/SPSS Inc., Chicago, IL). It included estimates for summarizing the continuous data as mean (X) and standard deviation (SD) for normally distributed data or median (Med) and range for skewed data. Frequency with percentage (%) was used for presenting qualitative data. A significant p-value was considered when it is equal or less than 0.05.

RESULTS

The current study included 30 patients with craniocervical junction lesion discovered by at least one imaging modality. The median age of the cases was 7 years and the age ranged from 1 day (as the minimum age) and 18 years (as the maximum age). There were 9 cases neonates (less than 1 month), 4 cases with age between 1 month and 1 year, while the remaining 17 cases (56.7%) were in age between 1 year and 18 years (figure 7).

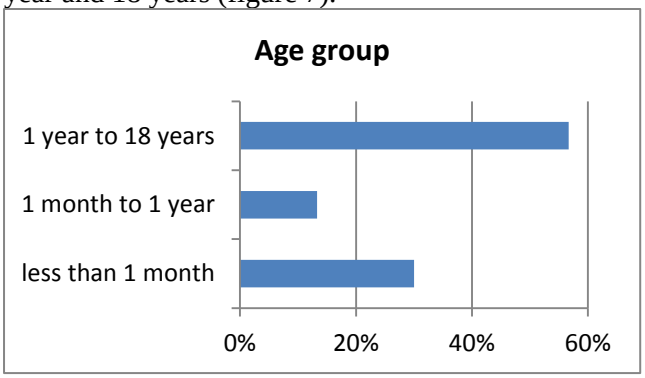

Figure (7): Age group distribution of the cases included in the study.

Regarding the gender distribution (figure 8), there was 16 males (53.3%) and 14 females (46.7%)

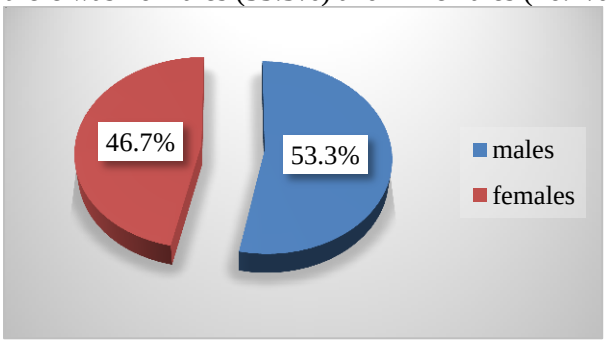

Figure (8): Sex distribution of the cases included in the study.

Regarding presentation, head enlargement was the most common presentation in 9 cases (30%), lumbar swelling in 6 cases (20%), cervical pain in 4 cases (13.3%), headache and neurodevelopmental delay in 3 cases (10%) each, while disturbed gate, migraine, right side weakness, short stature and upper and lower limb spasticity in 1 case each (3.3%) (Table 1).

Table (1): Analysis of the presentation of the cases in the study

Variables	Study cases N = 30
Head enlargement	**9 (30%)**
Lumbar swelling	**6 (20%)**
Cervical pain	**4 (13.3%)**
Headache	**3 (10%)**
Neurodevelopmental delay	**3 (10%)**
Disturbed gate	**1 (3.3%)**
Migraine	**1 (3.3%)**
Right side weakness	**1 (3.3%)**
Short stature	**1 (3.3%)**
Upper and lower limb spasticity	**1 (3.3%)**

Regarding the radiological diagnosis, Chiari 1 was the most common diagnosis in 11 cases (36.7%), Chiari 2 in 7 cases (23.3%), tonsillar herniation in 3 cases (10%), MPS in 3 cases (10%), achondroplasia in 2 cases (6.7%), while split atlas, platypesia, Down syndrome and Os odenoottidum showed 1 case each (3.3%).

Table (2): Analysis of the radiological diagnosis of the cases in the study

Variables	Study cases N = 30
Chiari 1	**11 (36.7%)**
Chiari 2	**7 (23.3%)**
Tonsillar herniation	**3 (10%)**
MPS	**3 (10%)**
Achondroplasia	**2 (6.7%)**
Split atlas	**1 (3.3%)**
Platypesia	**1 (3.3%)**
Down syndrome	**1 (3.3%)**
Os odenoottidum	**1 (3.3%)**

Regarding the radiological findings (table3), basilar invagination, cervico-medullary kinking, OS odontidum in 2 cases (6.7%) each, while absent anterior arch of C1, (Atlanto axial sublaxation + platypesia), odontoid hypoplasia, (odontoid aplasia + atlantoaxial sublaxation + split atlas) and split atlas 1 case each (3.3%). Regarding the intervention in the cases of the study, VP shunt were performed in 8 cases (26.7%) and decompression surgery in 3 cases (10%) (Figure 9).

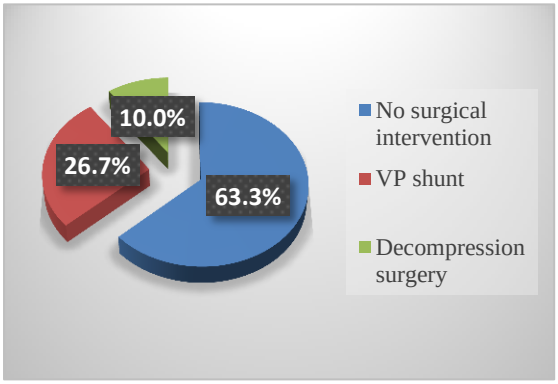

Figure (9): intervention in the cases in the study.

Table (3): Analysis of the radiological findings in the cases in the study

Variables	Study cases N = 30
Basilar invagination	**2 (6.7%)**
Cervico-medullary kinking	**2 (6.7%)**
OS odontidum	**2 (6.7%)**
Absent anterior arch of C1	**1 (3.3%)**
Atlantoaxial sublaxation + platypesia	**1 (3.3%)**
Odontoid hypoplasia	**1 (3.3%)**
Odontoid aplasia + atlantoaxial sublaxation + split atlas	**1 (3.3%)**
Split atlas	**1 (3.3%)**

Regarding the radiological investigations in the current study, CT was performed in 22 cases (73.3%) and MRI was performed in 20 cases (66.7%) (Table 4 & figure 10).

Table (4): Radiological investigations in the cases of the study

Variables	Study cases N = 30
CT	
Not performed	**8 (26.7%)**
Performed	**22 (73.3%)**
MRI	
Not performed	**10 (33.3%)**
Performed	**20 (66.7%)**

Categorical data expressed as Number (%)

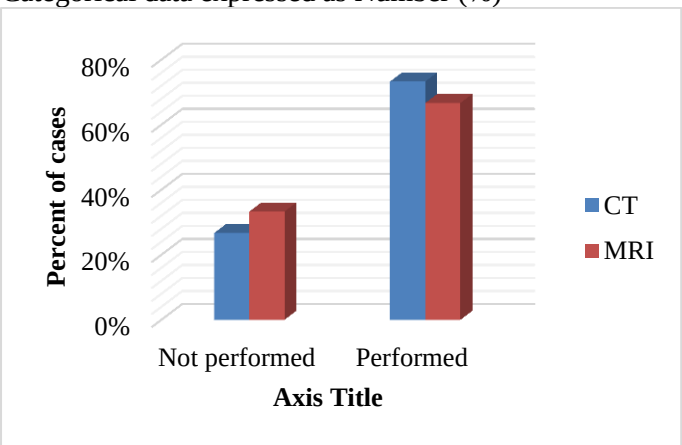

Figure (10): Radiological investigations in the cases of the study.

Regarding the associated findings, hydrocephalus was the most common associated finding in 18 cases (30%), cervical stenosis in 8 cases (26.7%), syrinx in 5 cases (16.7%) and butterfly vertebra in 1 case (3.3%) (Table 5).

Table (5): Analysis of the associated findings in the cases in the study

Variables	Study cases N = 30
Hydrocephalus	**18 (60%)**
Cervical stenosis	**8 (26.7%)**
Syrinx	**5 (16.7%)**
Butterfly vertebra	**1 (3.3%)**

DISCUSSION

Craniovertebral junction (CVJ) has a complicated function and anatomy. It is a common site for many abnormalities (congenital, developmental and acquired). Imaging of this complicated region in order to reach an accurate diagnosis creates a burden on imaging specialists [5].

In the study of **Singh *et al.*** [6], developmental cause of atlanto-axial diseases most commonly presented in 11–20-years age group (18 patients, 40.91%). In our study the mean age was 7 years and the majority of the cases (17cases) presented within the age group ranging from 1 year up to the age of 18 years, which is compatible with the previously mentioned study.

The study of **Talukdar *et al.*** [7] was conducted on 46 cases of CVJ abnormalities, 28 cases (61%) were congenital anomalies, while our study was conducted on 30 patients with developmental anomalies in the craniocervical junction. Developmental etiology was the most common cause of CCJ anomalies in the study conducted by **Dhadve *et al.*** [8] and **Singh *et al.*** [6].

The signs and symptoms of the CVJ pathologies are variable. Typically, they start silently, manifest themselves very late and progress slowly [9]. The most common presenting symptom was neck pain, followed by limb weakness [6, 8].

In our study head enlargement was the most common presentation (as hydrocephalus) in 9 cases (30%) followed by lumbar swelling in 6 cases (20%), while cervical pain was present in 4 cases only (13.3%). In our study the main presentation symptom didn't match the previously mentioned studies of **Dhadve *et al.*** [8] and **Singh *et al.*** [6]. Their studies were conducted on all types of craniocervical junction pathologies with no statistical analysis mentioned as regards different presentation of different group etiology. Headache and neurodevelopmental delay were presented in 3 cases only. Disturbed gate, migraine, right side weakness, short stature as well as upper and lower limb spasticity were presented in one case each.

In the study of **Talukdar *et al.*** [7] males were more common than females (3:1). However, the study of **Singh *et al.*** [6] found that male to female ratio was

1.47:1. Regarding the gender distribution in our study, it was prevalent in both sexes with ratio reaching 1.14:1 male: female ratio which correlated with the above-mentioned studies

In our study hydrocephalus was the most common associated finding in 18 cases (30%) followed by cervical canal stenosis in 8 cases (26.7%), which was one of the typical findings in patients with MPS (Maroteaux-Lamy syndrome) in the study conducted by **Nessej *et al.*** [10].

In the study conducted by **Talukdar *et al.*** [7] syrinx was found in 9 cases out of 46 cases, while in our study it was found in 5 cases only. However, this study included different pathologies (congenital causes, 28 cases, while others were traumatic, rheumatoid and tuberculous). In **Talukdar *et al.*** [7] study, the commonest congenital CVJ anomalies were atlantoaxial dislocation and Arnold Chiari malformation. While, the study of **Araújo *et al.*** [11] stated that Arnold Chiari malformation is always associated with craniovertebral junction anomalies such as basilar invagination (BI), platypesia, hydrocephaly & meningocele.

Chiari malformations represented the majority of the cases in our study representing 18 cases (11 cases of type 1 & 7 cases of type 2). Basilar invagination was only found in two cases as well as atlantoaxial subluxation. Among the different treatment modalities of Arnold Chiari malformation, the surgical procedures are the most recommended, one example is the ventricle peritoneal shunt (VP-shunt), which is among the most used options for the correction of hydrocephalus, very often associated with this malformation [11].

In our study 8 cases needed VP-shunt (figure9) as result of hydrocephalus representing 72% of total cases of Arnold Chiari malformation & 3 cases needed decompression surgery of the skull base. In the study conducted by **Singh *et al.*** [6], Os odontidum was found in 8 (14%) cases, while in our study Os odontidum was found in two cases(table3) only (7%). The above-mentioned study was conducted on 57 patients, while ours was conducted on 30 patients.

Down syndrome (DS) patients can present with instability of the atlanto-axial articulation which may occurs secondary to either bony anomalies or ligamentous defects. Os odontidum has been reported in about 6% of DS children leading to canal stenosis [12]. In our study one patient with Down syndrome presented with spasticity in both upper and lower limbs. MRI scan showed the presence of Os odontidum, which was causing significant spinal canal stenosis with subsequent spinal cord compression.

MRI is superior to CT in achondroplasia patients for the evaluation of the craniocervical junction. It provides imaging of the brainstem and cervical cord, including cord compression and T2 signal changes, as well as visualization of the posterior fossa structures [13]. In our study two patients were included with achondroplasia both of them underwent MRI, which showed cervico-medullary kinking & spinal cord compression.

Congenital atlas abnormalities are rare malformations. They include aplasia/ hypoplasia and arch clefts and may be asymptomatic [14]. In the study conducted by **Meddeb *et al.*** [15], isolated hypoplasia of the anterior arch has been reported up to 0.7%, while in our study only one case had anterior arch atlas detect, however it was in a case of Chiari malformation and the patient suffered from hydrocephalus

Bipartite atlas (Split atlas) is a rare malformation consisting of partial/ complete defect between the posterior and the anterior arches of C1 vertebrae body (figure3). Several authors considered it as "anatomical variant," without clinical impact, and indeed almost all of the cases reported in the literature were discovered incidentally [15]. In our study only one case was discovered accidently of bipartiate atlas with no neurological symptoms or signs on neurological assessment.

Plain radiographs have limitations in diagnosis and anatomical delineation of CVJ because it is very difficult to identify the CVJ due to structures overlapping. So, cross-sectional imaging (MRI and CT) is used for evaluation. MRI is ideal for nerves, soft tissue and ligaments' evaluation, while multi-Slice CT describes the bone anatomy and pathology of the CVJ well [9, 6].

Radiological investigations in our current study, CT was performed in 22 cases (73.3%) and MRI was performed in 20 cases (66.7%). In 20 cases both CT and MRI scans were performed. Combination of both scans is highly recommended for accurate assessment of the anomalies.

CONCLUSION

Careful radiological examination of the CCJ and understanding of its complicated anatomy is required in order to reach the proper diagnosis of its lesions. Both CT scan and MRI are required to be done for accurate diagnosis. Larger sample to assess the detailed changes in CCJ developmental causes is advised. Fully equipped radiological departments may be needed in case of anesthesia.

- **Conflict of interest:** The authors declared no conflict of interest.
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