A 13 years old male patient presented clinically with atrophy of the small muscles of the hands, bulbar cranial nerve manifestations and cerebellar manifestations. Physical examination revealed a short neck, low hairline, painful torticollis, restricted neck motion, and a loss of the normal cervical spine lordosis. Neurologic examination showed downbeat nystagmus, bulbar cranial nerve impairment, bilateral corticospinal tract signs, ataxia, and weakness and sensory loss in the hands.

**Figure 1.** Sagittal CT scan reconstructed image (A). Plain x ray image (B) and Precontrast MRI T1 image (C). Notice that the odontoid process is abnormally large, subluxated, and posteriorly located. Also notice basilar invagination, The tip of the odontoid process is in touch of the lower pons. A syringomyelic cavity is seen opposite the second vertebra and extends for only one spinal segment. The cerebellar tonsils are herniated below the level of the foramen magnum. The brain stem is abnormally elongated with herniation of the medullar below the level of the foramen magnum (Type II Chiari malformation). Notice malalignment of the cervical vertebrae.
Figure 3. Precontrast MRI T1 images. Notice that the odontoid process is abnormally large, subluxated, and posteriorly located. Also notice basilar invagination. The tip of the odontoid process is in touch of the lower pons. A syringomyelic cavity is seen opposite the second vertebra and extends for only one spinal segment. The cerebellar tonsils are herniated below the level of the foramen magnum. The brain stem is abnormally elongated with herniation of the medullar below the level of the foramen magnum (Type II Chiari malformation). Interestingly the foramen of magendi (which is situated at the caudal end of the 4th ventricle and can easily be appreciated on the T1 MRI sagittal images) is appreciated as being markedly diminished in volume and transformed into a long slit that opened below the level of the foramen magnum. Notice the extradural space occupying lesion compressing and anteriorly displacing the upper cervical sinal cord.

Figure 2. The atlas is abnormally high, almost completely invaginated intracranially (basilar invagination) with complete fusion of the atlas with the occipital bone (atlanto-occipital fusion or assimilation of atlas)
Figure 4. Precontrast MRI T1 images. Notice that the odontoid process is abnormally large, subluxated, and posteriorly located. Also notice basilar invagination, The tip of the odontoid process is in touch of the lower pons. A syringomyelic cavity is seen opposite the second vertebra and extends for only one spinal segment. The cerebellar tonsils are herniated below the level of the foramen magnum. The brain stem is abnormally elongated with herniation of the medullar below the level of the foramen magnum (Type II Chiari malformation). Interestingly the foramen of magendi (which is situated at the caudal end of the 4th ventricle and can easily be appreciated on the T1 MRI sagittal images) is appreciated as being markedly diminished in volume and transformed into a long slit that opened below the level of the foramen magnum. Notice the extradural space occupying lesion compressing and anteriorly displacing the upper cervical spinal cord.

Figure 5. MRI T2,T1 images. Notice that the odontoid process is abnormally large, subluxated, and posteriorly located. Also notice basilar invagination, The tip of the odontoid process is in touch of the lower pons. A syringomyelic cavity is seen opposite the second vertebra and extends for only one spinal segment. The cerebellar tonsils are herniated below the level of the foramen magnum. The brain stem is abnormally elongated with herniation of the medullar below the level of the foramen magnum (Type II Chiari malformation). Interestingly the foramen of magendi (which is situated at the caudal end of the 4th ventricle and can easily be appreciated on the T1 MRI sagittal images) is appreciated as being markedly diminished in volume and transformed into a long slit that opened below the level of the foramen magnum. Notice the extradural space occupying lesion compressing and
anteriorly displacing the upper cervical spinal cord.

Figure 6. MRI T2 images. Notice that the odontoid process is abnormally large, subluxated, and posteriorly located. Also notice basilar invagination. The tip of the odontoid process is in touch of the lower pons. A syringomyelic cavity is seen opposite the second vertebra and extends for only one spinal segment. The cerebellar tonsils are herniated below the level of the foramen magnum. The brain stem is abnormally elongated with herniation of the medullar below the level of the foramen magnum (Type II Chiari malformation). Interestingly the foramen of magendi (which is situated at the caudal end of the 4th ventricle and can easily be appreciated on the T1 MRI sagittal images) is appreciated as being markedly diminished in volume and transformed into a long slit that opened below the level of the foramen magnum. Notice the extradural space occupying lesion compressing and anteriorly displacing the upper cervical spinal cord.

Figure 7. Post contrast MRI T1 image (A) Showing a dumb-bell cystic neurofibroma (Confirmed surgically) at C 2 vertebra compressing and displacing the spinal cord. Notice malalignment of the cervical vertebrae and the high odontoid process.
In the above report case we have a primary pathology, a secondary pathology and a tertiary pathology

The primary pathology is basilar invagination (intracranial invagination of atlas and axis)

- Basilar impression/invagination is a congenital skeletal malformation that combines deformity of the osseous structures of the base of the skull with invagination of the foramen magnum and upper cervical spine into the posterior fossa.

- Basilar invagination is almost invariably associated with assimilation of atlas (atlantooccipital fusion). In most of the cases the odontoid process is fixed and not subluxated. In the author experience, hydrosyringomyelia and syringobulbia are more likely to occur when the odontoid process is fixed. Metwally \(^\text{12}\) regarded basilar invagination as a risk factor that increases the probability of a cervical syringomyelic cavity breaking upward into the medulla and forming syringobulbia. Probably the craniocervical junction is more crowded when the odontoid process is fixed than when it is mobile and subluxated, thus increasing the probably of the emergence of the secondary and the tertiary pathology as explained later.

- Complete intracranial invagination of the atlas and axis (basilar impression or invagination) is demonstrated in about 25\% of cases \(^\text{12}\) with congenital syringomyelia. Basilar impression/invagination acts by reducing the volume capacity of the posterior fossa and crowds the cerebellum thus producing cerebellar tonsillar herniation, thereby setting up the substrate for the funneling of the CSF pressure waves into the central canal of the spinal cord thus creating hydrosyringomyelia. \(^\text{12}\)

Secondary pathology is cerebellar tonsillar herniation (Arnold Chiari malformation)

- Basilar impression/invagination acts by reducing the volume capacity of the posterior fossa and crowds the cerebellum thus producing cerebellar tonsillar herniation, thereby setting up the substrate for the funneling of the CSF pressure waves into the central canal of the spinal cord thus creating hydrosyringomyelia. \(^\text{12}\)

Tertiary Pathology is hydrosyringomyelia

- Funneling of the CSF pressure waves into the central canal of the spinal cord thus creating hydrosyringomyelia.

---

**DIAGNOSIS:**

MULTIPLE CONGENITAL BONY AND SOFT TISSUE CRANIOCERVICAL ANOMALIES (BASILAR INVAGINATION, ASSIMILATION OF ATLAS, SUBLUXATION OF THE ODONTOID PROCESS, TYPE II CHIARI MALFORMATION) ASSOCIATED WITH EXTRADURAL CYSTIC NEUROFIBROMA AND A SINGLE SPINAL SEGMENT SYRINGOMYELIC CAVITY AT THE LEVEL OF THE NEUROFIBROMA (C 2 VERTEBRA)

**DISCUSSION**

Three main points must be commented upon while discussing this case. The Basilar impression/invagination, The syringomyelia and the epidural neurofibroma

- Basilar impression/invagination

Basilar impression is a congenital skeletal malformation that combines deformity of the osseous structures of the base of the skull with invagination of the foramen magnum and upper cervical spine into the posterior fossa. The foramen magnum is small and deformed. The floor of the posterior fossa and the odontoid process are displaced upward, further narrowing the foramen magnum and compressing the medulla and upper cervical spinal cord. Metabolic bone disease, such as osteomalacia, osteogenesis imperfecta, congenital hypothyroidism, rickets, and Paget's disease, may result in acquired basilar impression. Basilar impression is also seen in association with mucopolysaccharidoses II (Hurler syndrome) and Down's syndrome. The congenital condition is inherited as an
autosomal dominant trait with variable expression.

Cervical spine (oblique lateral aspect).

1. Temporal bone
2. Head of mandible
3. Mandible
4. Occipital bone
5. Atlas
6. Axis
7. Third cervical vertebra
8. Spinous process of fourth cervical vertebra
9. Transverse process of fifth cervical vertebra with groove for spinal nerve
10. Intervertebral foramen
11. Vertebra prominens (C7)
12. First rib
13. Sternum (manubrium sterni)
14. Atlantoaxial joint
15. Lateral atlantodental joint
16. Body of axis
17. Body of fourth cervical vertebra
18. Body of fifth cervical vertebra
20. Body of seventh cervical vertebra

Figure 8. Normal craniocervical junction
Basilar impression (or invagination) is the deformity of the bones of the base of the skull at the margin of the foramen magnum. The floor of the skull appears to be indented by the upper cervical spine; therefore, the tip of the odontoid is more cephalad. This increases the risk of neurologic damage from injury, circulatory embarrassment, or impairment of cerebrospinal fluid (CSF) flow. Two types of basilar impression exist: (1) primary, a congenital abnormality often associated with other anomalies such as atlanto-occipital fusion, hypoplasia of the atlas, bifid posterior arch of atlas, odontoid abnormalities, Klippel-Feil syndrome, and Goldenhar syndrome; and (2) secondary, a developmental condition usually attributed to softening of the bone, in which the deformity develops later in life.

**Table 1. Characteristics of basilar invagination**

- Elevation of the floor of the posterior fossa (the floor the posterior fossa is convex upward)
- The upper cervical spine (atlas and axis) encroaches on the brainstem and spinal cord (the tip of the odontoid is more cephalad) as the base of the skull is displaced toward the cranial vault
- Partial to complete atlanto-occipital fusion (assimilation of atlas)
- The odontoid process might be partially of completely subluxated
- Compression of the cervico-medullary junction by the odontoid process, fibrous band, or tight dura at the level of foramen magnum
- Chiari malformation, and congenital syringomyelia might be associated with basilar invagination

With basilar impression, the upper cervical spine encroaches on the brainstem and spinal cord as the base of the
skull is displaced toward the cranial vault. Motor and sensory disturbances are noted in 85% of individuals who are symptomatic. However, most affected patients remain asymptomatic until the second or third decade of life, when they may present with headache, neck ache, and neurologic compromise (prevalence of symptoms, 15%).

Figure 10. Basilar invagination. The base of the skull is elevated and convex upward with high odontoid process.

Figure 11. A,B Plain X ray of the cervical spine in two cases with congenital syringomyelia. Notice widening of the cervical canal (A,B). Also notice the associated Klippel-Feil anomaly and basilar impression with high odontoid process (B).

Complete intracranial invagination of the atlas and axis (basilar impression or invagination) is demonstrated in about 25% of cases with congenital syringomyelia. Basilar impression acts by reducing the volume capacity of the posterior fossa and crowds the cerebellum thus producing cerebellar tonsillar herniation, thereby sitting up the substrate for the funneling of the CSF pressure waves into the central canal of the spinal cord thus creating hydrosyringomyelia. In most cases with basilar impression the odontoid process was fixed with no evidence of
subluxation. Congenital syringomyelia, when associated with basilar invagination, is more likely to be associated with extension of the cervical intramedullary cavitation into the lower brain stem (syringobulbia). Metwally regarded basilar invagination as a risk factor that increases the probability of a cervical syringomyelic cavity breaking upward into the medulla and forming syringobulbia.

Assimilation of the atlas (complete fusion between the atlas and the occipital condyles) occur in about 5% of cases of congenital syringomyelia, however it might be present as an isolated phenomenon. In atlanto-occipital fusion, the odontoid process is abnormally high, subluxated and compressing the cervico-medullary junction posteriorly. The most significant finding in assimilation of the atlas with neurological symptoms is an odontoid process with abnormal size, in abnormal position and with abnormal mobility. Assimilation of the atlas is invariably present with basilar invagination and some degree of atlanto-occipital fusion is invariably present in basilar invagination.  

When the atlas is fused with the occiput, flexion of the head results in partial forward subluxation of the fused atlas on the axis. Posterior displacement of the odontoid process then occurs, resulting in compression of the cervico-medullary junction. As the posterior luxation of the odontoid process is intermittent (only during head flexion), so intermittent compression of the cervico-medullary junction might result, initially, in intermittent neurological manifestations.

Figure 12. CT myelography showing basilar invagination with atlanto-occipital fusion. The odontoid process is more
cephalad than normal and subluxated and compressing the cervicomedullary junction posteriorly upon head flexion.

Atlanto-occipital fusion, also termed "occipitalization of the atlas," is the most commonly recognized anomaly involving the craniovertebral junction, encountered in 0.14% to 0.25% of the population. It is characterized by complete or partial fusion of the atlas to the occiput and likely results from failure of segmentation of the last occipital and first cervical sclerotomes and the hypochordal bow. Affected patients often have low hairline, short neck, and restricted neck movements, similar to the Klippel-Feil syndrome. Associated congenital anomalies, present in 20% of patients, include incomplete clefting of the nasal cartilage, cleft palate, congenital external ear deformities, cervical ribs, hypospadias, and urinary tract anomalies. Atlanto-occipital fusion invariably results in basilar invagination, less severe when only the anterior arch is fused to the basion and most pronounced when there is complete assimilation, including fusion of the lateral masses to the occipital condyles which, themselves, may be hypoplastic. Seventy percent of patients additionally have associated congenital nonsegmentation of C2-3 (partial or complete). This places greater demands on the atlantoaxial joint leading to atlantoaxial instability in 50% of patients. Sudden death in patients with atlanto-occipital assimilation has been reported.

![Figure 13. Atlanto-occipital assimilation with atlantoaxial subluxation. A, Reconstructed midsagittal CT scan reveals complete assimilation of the anterior and posterior atlas arches to the occipital bone. The odontoid process (O) lies at the level of the foramen magnum. A = anterior arch; P = posterior arch; B = basion. B, Midsagittal T1-weighted MR image reveals elongation and a "comma" configuration to the anterior lip of the foramen magnum produced by incorporation of the anterior atlas arch to the basion (dot). A similar finding is identified at the posterior margin of the foramen magnum. There is an increase in the width of the anterior atlantodental interval (dotted line), signifying incompetence of the transverse ligament, such that the displaced odontoid process (O) is producing marked compression of the cervicomedullary junction. Also note congenital incomplete segmentation of C2 and C3.](image)

As opposed to atlanto-occipital fusions, atlantoaxial fusions are rare. Fusion of the anterior atlas arch to the odontoid process is frequently accompanied by hypoplasia of the posterior atlas arch and congenital block vertebrae in the lower cervical spine. The odontoid process itself may be relatively hypoplastic. This fusion anomaly does not predispose to any instability. There is an increased incidence of atlantoaxial fusion anomalies associated with the Chiari I malformation, in addition to other malformations of the skull base and cervical vertebrae. An unusual case of anterior atlas arch fusion to the basion and posterior atlas arch fusion to the posterior arch of C2, associated with insufficiency of the transverse ligament, has been reported.
Figure 14. Chiari I malformation with basilar invagination. A, Midsagittal T1-weighted MR scan demonstrates caudal displacement of the cerebellar tonsils (solid arrow), almost to the C3-4 level. Marked basiocciput hypoplasia is present resulting in shortening of the clivus and basilar imagination with all of the anterior Cl arch, the odontoid process, and much of the axis body, positioned above Chamberlain’s line (dotted line). Note how the basion (open arrow) is located at the midspons level such that, essentially, all of the medulla lies within the upper cervical spinal canal. Congenital nonsegmentation of C2-3 is also identified. B, Midsagittal T1-weighted MR image of the thoracic spine demonstrates associated hydromyelia with multiple loculations.

- **Clinical Characteristics**

Neurologic symptoms most often begin after age 10 years. Neck stiffness, progressive spasticity and weakness of the legs, occipital headaches, and difficulty walking are common complaints. Basilar impression should be considered in cases of progressive cerebellar ataxia or spastic paraparesis and brain-stem or cervical cord syndromes, which might be mistaken for multiple sclerosis, spinal cord tumors, Arnold-Chiari malformation, or syringomyelia. Basilar impression may be associated with the Arnold-Chiari malformation and aqueductal stenosis; therefore, hydrocephalus may occur. The progressive symptoms are attributed to pressure on the medulla and upper cervical spinal cord, similar to that of other craniocervical junction defects, such as Arnold-Chiari malformation. Interference with the local blood supply, as well as the direct compression of neural structures, has been postulated as a pathogenic mechanism.

Physical examination reveals a short neck, low hairline, painful torticollis, restricted neck motion, and a loss of the normal cervical spine lordosis. Neurologic findings may include downbeat or periodic alternating nystagmus, fixed or intermittent cranial nerve impairment, bilateral corticospinal tract signs, ataxia, and weakness or sensory loss in the hands.

Figure 15. A, Posterior photo of a patient with basilar invagination syndrome and an anomaly of the atlanto-occipital fusion. The image shows an elevated left shoulder, a short webbed neck, and a low hairline. B, This patient has basilar invagination and an anomaly of the occipitocervical junction. The patient's flexion and extension after the atlanto-occipital fusion is demonstrated. His rotation was very limited. C, basilar invagination. Flexion of the
cervical spine in a patient who had an atlanto-occipital fusion.

**Figure 16.** CT myelography showing basilar invagination associated with Arnold Chiari malformation and syringobulbia in (B). Notice the partial to complete atlanto-occipital fusion with no evidence of subluxation of the odontoid process. The odontoid process is more cephalad than normal.

**Figure 17.** MRI T1 precontrast image (A) and CT bone window (B) showing basilar invagination associated with Arnold Chiari malformation and syringomyelia. The hyperintense odontoid process is seen indenting the brain stem (the high MRI T1 contrast of the odontoid process is due to increased fat content). Notice the partial to complete atlanto-occipital fusion with no evidence of subluxation of the odontoid process. The odontoid process is more cephalad than normal.

The diagnosis is confirmed by a lateral roentgenogram of the skull. The odontoid process extends above a line drawn from the hard palate to the posterior edge of the foramen magnum (Chamberlain's line \(^{13}\)). MR imaging and CT clearly define the existence of neural compression.
Management. Surgical decompression of the posterior fossa and the upper cervical cord is the treatment for symptomatic patients with congenital basilar impression. Decompressive surgery should be avoided in children with acquired basilar impression resulting from a metabolic bone disease because surgery may worsen the deformity.  

The associated syringomyelic cavity

The intramedullary syringomyelic cavity demonstrated in this patient is a very shot one (one spinal segment). There are two explanation for the associated syringomyelic cavity.

- The cavity is secondary to the cerebellar tonsilar herniation and represent the starting point that will ultimately extends to involve the cervico-dorsal spinal cord.

Cerebellar tonsilar herniation produces the following anatomical variations as follows

- Stenosis of the foramen magnum and the foramen of Magendie. This is produced either mechanically when the foramen magnum is crowded by the descent of the cerebellar tonsils through it or by the adhesions that were demonstrated between the cerebellar tonsils and the posterior aspect of the cervico-medullary regions. The foramen of Magendie is transformed into an abnormally elongated slit-like channel that opened below the foramen magnum in all patients.
The anatomical aberrations induced by hindbrain herniation will block the normal pathway for the escape of the ventricular fluid arterial pulse wave and redirect it towards the calamus scriptorius. The CSF arterial pulse wave will then be funneled through the calamus scriptorius into the central canal of the spinal cord dilating it and eventually creating hydrosyringomyelia. Accordingly a direct cause-effect relationship could be established between hindbrain herniation and the development of hydrosyringomyelia. 11,12,15

Accordingly hindbrain herniation should constitute the anatomical substrate of hydrosyringomyelia and the cause of syringomyelia should be sought not in the spinal cord but in the herniation of the hindbrain. The diagnosis of congenital syringomyelia is not possible unless this the aetiological factor (hindbrain herniation) is demonstrated radiologically. 11,12,15

However the demonstrated syringomyelic cavity is a very short one and this is not the case for congenital hydrosyringomyelia. In congenital hydrosyringomyelia the syringomyelic cavity is usually a long multisegmental one that involves most of the cervico-dorsal spinal cord. However the demonstrated intramedullary cavity could be just the beginning, the starting point, that will ultimately extends further upward and downwards as the disease progresses. The pathological process, in this patient, was probably caught early enough to demonstrate such a small syringomyelic cavity.

- **2- The second explanation for the syringomyelia cavity is that it is secondary to the epidural neurofibromas (neoplastic syringomyelia)**

The cavity is demonstrated anatomically opposite to an epidural neurofibroma. Spinal cord cavitations are known to coexist with primary spinal cord neoplasms. They could be secondary to CSF flow obstructions and in this way they resemble obstructive hydrocephalus.

- **The dumbbell neurofibroma**

Neurofibromas are known to coexist with craniocervical anomalies and hydrosyringomyelia. Some even regarded the craniocervical anomalies and the associated syringomyelia cavity (when associated with optic nerve gliomas or neurofibromas for example) as a part of neurofibromatosis type 1. 18
SUMMARY

Occipitalization of the atlas or atlanto-occipital fusion is one of the most common osseous anomalies of the craniovertebral junction. Occipitalization represents the most cephalic ‘blocked’ vertebra encountered in the spine. It is characterized by complete or partial fusion of the bony ring of the atlas to the base of the occipital bone. The patients with craniovertebral joint anomalies exhibit the first neurological signs and symptoms usually no sooner than the second decade of life.

In patients with the atlanto-occipital fusion, the clinical findings suggest that the major neurological compression is due to the odontoid projection into the foramen magnum. The signs and symptoms of pyramidal tract, anterior bulbar and cranial nerve involvement may be present.

Assimilation of atlas is an osseous abnormality, which occurs in the base of skull in the region of foramen magnum. The union of the atlas with the occipital bone constitutes the anomaly. There may be partial or complete union. The occipital bone is derived from basioccipital, exoccipital and supraoccipital portions, all of which surround the foramen magnum. The basiocciput goes on to develop into four occipital somites. The caudal portion of the fourth occipital somite goes onto fuse with the cranial portion of the first cervical somite to form the proatlas; the proatlas is assimilated into the occiput to form the articular condyles and the tip of the odontoid process. The caudal half of the first cervical somite along with the cranial part of the second cervical somite goes on to form the atlas and the odontoid process of the axis. A paracondylar process represents vestiges of the cranial half of the first cervical sclerotome. This formation is referred to as a caudal shifting (a vertebra taking on the characteristics of its caudal neighbor) where the occipital vertebra separates from the occiput.

The symptoms and signs of pyramidal tract, anterior bulbar and cranial nerves involvement may be present. Less commonly, if the compression occurred posteriorly by the posterior lip of the foramen magnum, are the symptoms and signs related to the involvement of the posterior column of spinal cord. Patients with occipitalization of the atlas have short neck and restricted neck movements. Symptoms referable to the vertebral artery compression, such as dizziness, seizures, mental deterioration, and syncope may occur alone or in combination with those of the spinal cord compression. Any morphological and structural alteration of the cervical spine may lead to stenosis or substenosis of the vertebral arterial circulation and hence to brain stem anoxia. Fusion between occiput and atlas occurs anteriorly between the arch and the rim of the foramen with some segment of the posterior arch of atlas present in some instances. This fragment can frequently constrict the spinal canal causing intermittent symptoms depending on the position of the head.

Although atlanto-occipital fusion is a congenital condition, many patients do not develop the symptoms until the second decade of life. This may be due to a gradual increasing degree of ligamentous laxity and instability with aging. The onset of clinical symptoms can be sudden and precipitated by relatively minor trauma, the most common course is a progressive, but sudden onset or instant death has also been reported.

- **Addendum**
  - A new version of this PDF file (with a new case) is uploaded in my web site every week (every Saturday and remains available till Friday.)
  - To download the current version follow the link "$http://pdf.yassermetwally.com/case.pdf$".
  - You can also download the current version from my web site at "$http://yassermetwally.com$".
  - To download the software version of the publication (crow.exe) follow the link: $http://neurology.yassermetwally.com/crow.zip$
  - The case is also presented as a short case in PDF format, to download the short case follow the link:
At the end of each year, all the publications are compiled on a single CD-ROM, please contact the author to know more details.

Screen resolution is better set at 1024*768 pixel screen area for optimum display

### REFERENCES

#### References


10. Metwally MYM: Imaging of syringomyelia, a comparative study. Read at the Scientific Meeting of the Egyptian Society of neurology, Psychiatry and Neurosurgery October 1992


18. Chakravarty A, Bhargava A, Nandy S. A patient with optic pathway glioma, scoliosis, Chiari type I
malformation and syringomyelia: is it Neurofibromatosis type 1? Neurol India 2002;50:520