

Case Reports**Congenital craniocervical anomaly masquerading as motor neurone disease**

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We wish to publish a case of congenital craniocervical anomaly that had presented initially with clinical features suggestive of motor neurone disease (MND), leading to a critical delay in diagnosis and appropriate treatment. The purpose of this article is to increase awareness of this condition and its capability to masquerade as other conditions, so that delays of this kind could be prevented.

**Case history**

A 28-year-old carpenter was transferred to our unit with inability to move all 4 limbs of 1 month's duration. His illness had started insidiously at age 16, with difficulty in walking. It had progressed relentlessly so that by age 27 he was no longer employed, and was walking with the support of his arms while his legs dragged along. The weakness had first developed in the lower limbs, but had later involved the upper limbs also. Sensory symptoms, sphincter disturbances, brain stem symptoms, or intellectual impairment had not occurred.

He had been evaluated at another hospital 1 year before admission to our unit. A diagnosis card issued from there carried the additional information that there was muscle wasting (distribution not specified), exaggerated reflexes in all 4 limbs, and bilateral Babinski's sign, and also queried a sensory level at L5. Urine full report, blood counts, erythrocyte sedimentation rate, blood glucose, and serum urea and electrolytes had been normal. A diagnosis of MND had been offered, and treatment with physiotherapy and baclofen arranged.

One month before admission to our unit he had sustained a minor fall at home, and had immediately afterwards become completely paralysed in all 4 limbs,

with loss of sensation below the neck, total incontinence, and neck pain.

Physical examination revealed a conscious, alert and orientated man with difficulty in breathing and in talking. He was lying flat in bed without any voluntary movement of the limbs, but occasional extensor spasms involving all 4 limbs were noted. Memory and intellect were unimpaired. Cranial nerve examination revealed mild bilateral ptosis, small but symmetrical and reacting pupils, and sensory loss to pinprick over the face posteriorly. There was no papilloedema or downbeat nystagmus.

The upper limbs were flaccid and areflexic, with severe wasting of supraspinati and infraspinati bilaterally and clawing of both hands. The lower limbs were spastic and hyperreflexic with clonus at knees and ankles. Superficial reflexes, including the plantar responses, were absent. Ill-demarcated sensory levels were noted to light touch at C3-4 and to pinprick in the trigeminal distribution. There was no position sense in the limbs. The analgesia had no sacral sparing. Sweating was not noted over the trunk, limbs or face bilaterally.

All intercostal muscles were paralysed, and there was no discernible breathing movement in the abdomen; he was apparently breathing with the accessory muscles of respiration, with a vital capacity that varied between 400-500 ml at different times. The back and spine were normal clinically. Other systems were normal apart from signs attributable to the low vital capacity. The patient was on an indwelling urethral catheter.

Initial investigation was focused on the possibility of traumatic compression of the cervical cord, and a rigid cervical collar was applied and radiological studies carried out. X-ray skull and cervical spine (lateral view) showed atlanto-axial subluxation. There was also fusion of posterior elements of C2-3 vertebrae with a

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narrow waist and without angulation, suggestive of congenitally fused vertebrae (Figure 1).

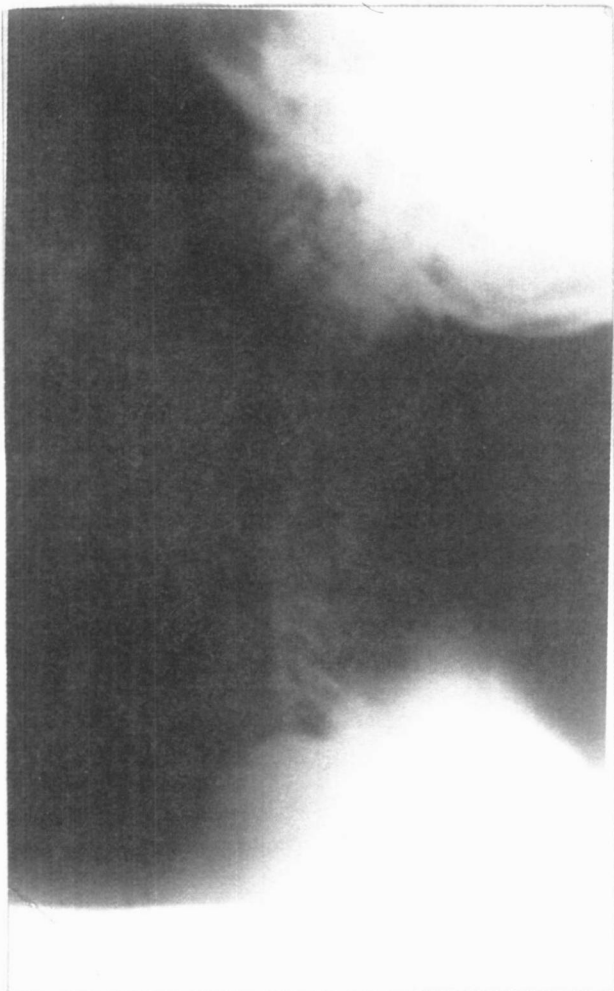


Figure 1

A computed tomographic (CT) contrast myelogram (Figure 2) confirmed the atlanto-axial subluxation, and showed interruption to the contrast column at C2. Defects in the anterior arch and between the lateral mass and the anterior arch of the atlas were also seen; they were well corticated, indicating congenital defects rather than fractures. The odontoid peg was seen protruding into the foramen magnum, indicating basilar invagination. The posterior cranial fossa (including the cerebellum) was normal.

We arrived at a diagnosis of congenital craniocervical anomaly with cervical synostosis and basilar invagination, leading to atlanto-axial instability (producing the insidious, progressive spastic quadriparesis since age 16) and subluxation with cervical cord compression following minor trauma (producing quadriplegia with sensory and autonomic levels, and incontinence). The patient requested the removal of the cervical collar, since this made his breathing more difficult.

Following deliberations with neurosurgical and orthopaedic surgical teams and discussions with the patient and his family, an attempt at decompression of the lesion was made. Unfortunately, application of skull calipers for this purpose was complicated by extra-dural haemorrhage, leading to coning, respiratory arrest and death.

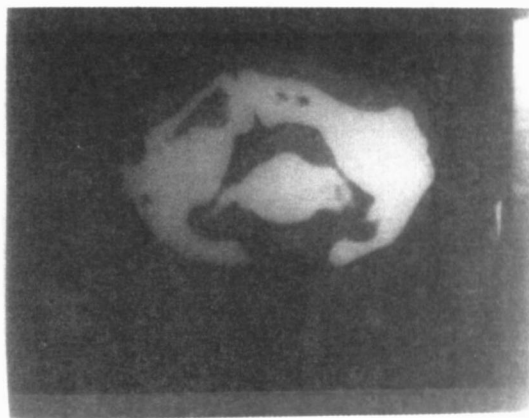


Figure 2

#### Discussion

Our patient's presentation must have looked reasonably compatible with MND to the team that had evaluated him initially. This is especially so in view of the combined upper and lower motor neurone signs, especially with wasting of small muscles of the hands combined with hyperreflexia in the upper limbs. Sensory signs too were not definite then. However, this initial diagnosis seemed untenable to us much later, in view of the physical signs that by then clearly indicated an upper cervical cord lesion. Also not favouring a diagnosis of MND were the age of onset (usually > 40 years), the total duration of illness (usually < 7 years), the query of sensory signs, and the positive Babinski's signs. Radiological investigation then led to the final diagnosis.

Congenital craniocervical anomalies are a group of congenital disorders of the cervical spine cord, some of which have a potential to produce neurological deficit<sup>1,2,3</sup>. They may be associated with congenital anomalies of the rest of the spine, as well as being associated with certain external markers, such as short neck, low posterior hairline, torticollis, and limitation of neck movement. features. Our patient lacked these features.

Basilar invagination, one of the craniocervical anomalies affecting the cranio-vertebral junction, is characterised by invagination or upturning of the margins of the foramen magnum into the base of the skull. Several radiological signs using plain X-rays (4) have been devised for its recognition (see Table 1). Its im-

portance lies in its ability to produce atlanto-axial instability, as was probably the case in our patient. Such instability may be complicated by subluxation following minor trauma. Our patient most probably had a chronic cervical myelopathy since age 16. It led to subluxation following a minor fall.

It is possible that an atlanto-axial subluxation can follow trauma even in a normal, non-fractured spine.

However, this requires a fairly significant fall, and is especially likely to occur if the patient lands on the chin. Our patient recalled a gentle fall while at home doing walking exercises (as part of physiotherapy), and although he could not recall exactly on what he landed, there was no external chin injury. He did not suffer from rheumatoid disease or any disorder of the connective tissues that might predispose to atlanto-axial subluxation.

**Table 1. Some radiological features of basilar invagination**

Radiological sign	Clarification
The tip of the odontoid process is above Chamberlain's line.	Chamberlain's line joins the posterior end of the hard palate to the posterior end of the foramen magnum in a X-ray skull and cervical spine (lateral view).
The tip of the odontoid process is more than 6 mm above McGregor's modification of Chamberlain's line.	McGregor's modification of Chamberlain's line joins the posterior end of the hard palate to the lowest point of the occiput in X-ray skull and cervical spine (lateral view).
The plane of the hard palate is not parallel to the plane of the axis vertebra.	X-ray skull and cervical spine (lateral view).
The atlanto-occipital joint is above	Fischgold's bimaoid line joins the tips of the mastoid processes in an X-ray skull (antero-posterior view).
The atlanto-occipital joint is within 1 cm of Fischgold's digastric line.	Fischgold's digastric line joins the digastric grooves in an X-ray skull and cervical spine (antero-posterior view).
The tip of the odontoid process is within the foramen magnum in a CT.	CT of foramen magnum (axial view or sagittal reconstruction.)

Several interesting though unexplained physical signs have been reported in association with these anomalies (see Reference 1 for a review). These include wasting of small muscles of the hands, loss of position sense in the upper limbs more than in the lower limbs, upper limb mirror movements, and downbeat nystagmus. Our patient had the first of these, while downbeat nystagmus was not seen. There is experimental evidence to support the hypothesis that lesions in the foramen magnum region can produce false-localising signs suggestive of a lower cervical cord lesion, by a mechanism that involves compromise of the venous drainage of the lower cervical cord grey matter (5). An alternative possibility in our patient is a concurrent syrinx, a condition that cannot be confidently ruled out with a CT myelogram; magnetic resonance imaging was not available to us.

Wasting of small muscles of the hands is a misleading sign for a patient with this condition to possess, since in the presence of brisk upper limb reflexes it would strongly suggest a wrong diagnosis of MND. Therefore, in view of these reasons, we suggest that radiological studies of the cervical spine be conducted routinely in all patients on whom a diagno-

sis of MND is considered clinically, so that this rare but treatable condition is not missed.

Other clinical syndromes that have been reported in association with congenital craniocervical anomalies<sup>1,2</sup> include cervical radiculopathy, spastic quadriparesis due to both compressive and non-compressive cervical myelopathy, syringomyelia, brain stem ischaemic episodes, occipital headache, and hydrocephalus. Radiological studies of the cervical spine would be valuable in these situations as well.

#### References

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