Head drop and camptocormia

T Umapathi, V Chaudhry, D Cornblath, D Drachman, J Griffin, R Kuncl

The spectrum of bent spine disorders

Head drop (drop) results from weakness of the neck extensor, or increased tone of the flexor muscles. It is characterised by marked anterior curvature or angulation of the cervical spine and is associated with various neuromuscular (table 1) and extrapyramidal disorders.10–15 Camptocormia or the bent spine syndrome was first described in hysterical soldiers in 1915 by the French neurologist Souques.16 Typically there is marked anterior curvature of the thoracolumbar spine. In some patients the spine is angulated forward, the arms propped against the thigh for support. More cases, all among soldiers, were reported during the first and second world wars. These patients responded well to psychotherapy. Recently camptocormia arising as a result of weakness or abnormality in the tone of the paraspinal muscles has been described (table 2). In contrast with other skeletal disorders of the spine such as kyphosis, the deformity in head ptosis and camptocormia is not fixed and is corrected by passive extension or lying in the supine position. It is not possible to straighten the neck or back voluntarily. The evaluation of these disorders can indeed be challenging and often no definitive diagnosis is made, as illustrated by four cases of head ptosis and camptocormia seen by us at the Johns Hopkins Hospital.

CASE A
An 80 year old man developed head ptosis insidiously over a period of few weeks. A week before this he had an upper respiratory tract infection and also experienced transient sharp pain over the left and then the right shoulder. He had no diplopia, dysarthria, dysphagia, limb weakness, or fatigueability. Examination showed severe neck extensor weakness, Medical Research Council (MRC) grade 2. Muscle strength was normal in all other cranial, proximal, and distal limb muscles. Serum creatine kinase (CK) was 362 IU/l (normal 24–195) at presentation. Anti-acetylcholine receptor antibody was not detected. Repetitive nerve stimulation of right biceps, right nasalis and right trapezius was normal. A therapeutic trial of pyridostigmine failed. Cervical spine MRI did not show significant compression of the cord, roots, or any specific changes in the paraspinal muscles. An EMG disclosed positive sharp waves restricted to lower cervical paraspinal muscles. To reduce confounding from neuropathic changes secondary to coexistent age related spondylotic changes, the thoracic paraspinal muscle was chosen for biopsy. The pathological findings were mixed. There were neuropathic changes such as angular atrophic fibres; occasional nuclear sacs, target and targetoid fibres as well as myopathic features such as the presence of hypertrophic and split muscle fibres; a few necrotic, degenerating and regenerating fibres; increased internalised nuclei, and mild endomysial fibrosis. No type grouping was noted, but there was massive type I predominance. No specific treatment was offered. A few months later, he started to improve. At follow up 2 years later, he was able to keep his head up for prolonged periods, especially in the sitting position. However, he was more symptomatic while standing. He reported an overall 60% subjective improvement in neck strength and weakness had not progressed to other muscles.

CASE B
A 74 year old man with diabetes mellitus and hyperlipidaemia (for which he had been taking a statin for a few years) complained of progressive anterior curvature of the spine associated with proximal limb weakness for few months. On examination, he had moderately severe anterior thoracolumbar curvature that could be extended passively. Diffuse weakness was present; proximal limb strength was MRC grade 4 and distal, 4+. Electrodiagnostic evaluation only showed evidence of an axonal polyneuropathy, consistent with diabetes. Serum CK was persistently raised (367 IU/l). Spine MRI only showed mild degenerative changes. Anti-acetylcholine receptor antibody was not detected. Biopsy of the rectus femoris showed increased fibre size variability, necrosis, mild fibrosis, occasional red rimmed vacuoles, and one fibre which stained positive for ubiquitin, suggestive of inclusion body myositis. However, inflammation or amyloid

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Table 1  Neuromuscular causes for head ptosis

<table>
<thead>
<tr>
<th>Site of pathology</th>
<th>Disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Motor neuron</td>
<td>Amyotrophic lateral sclerosis</td>
</tr>
<tr>
<td>Periphereral nerve</td>
<td>Chronic inflammatory polyneuropathy</td>
</tr>
<tr>
<td>Central nervous system</td>
<td>Myasthenia gravis</td>
</tr>
<tr>
<td>(a) Inflammatory</td>
<td>Polymyositis</td>
</tr>
<tr>
<td>(b) Congenital/heritary</td>
<td>Inclusion body myositis</td>
</tr>
<tr>
<td>(c) Endocrine/metabolic</td>
<td>Facioscapulohumeral dystrophy</td>
</tr>
<tr>
<td>Muscle</td>
<td>Cushing syndrome</td>
</tr>
</tbody>
</table>

Table 2  Causes of camptocormia

<table>
<thead>
<tr>
<th>Disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Motor neuron</td>
</tr>
<tr>
<td>Muscle</td>
</tr>
<tr>
<td>Parkinsonism</td>
</tr>
<tr>
<td>Idiopathic</td>
</tr>
</tbody>
</table>

Abbreviations: CK, creatine kinase; MSA, multisystem atrophy; ALS, amyotrophic lateral sclerosis.
deposition were not seen. Despite stop-
ing statin, his weakness continues to pro-
gress and CK has remained raised (239 IU/L).

CASE C
A 59 year old woman presented with a 4
year history of progressive, painless
anterior curvature of the spine. She had
a family history of facioscapulohumeral
muscular dystrophy but not of bent
spine. Examination showed campto-
cornia along with scapular winging
and mild facial and proximal weakness.
Her CK was at the upper limit of normal, 186
IU/L. An EMG showed short duration,
small amplitude motor units with early
recruitment in the proximal muscles.
Abnormal spontaneous activity was seen
in the thoracic paraspinal muscles. No
decremental response to repetitive nerve
stimulation was present. Spine MRI was
unremarkable. 4q35 gene deletion, con-
sistent with facioscapulohumeral mus-
cular dystrophy, was detected.

CASE D
A 63 year old woman developed slowly
progressive, painless thoracolumbar
stump of about 30 degrees over 8 to 9
years. The deformity was not fixed and
was worse in the standing position,
better sitting, and minimal while lying
down. There was no family history of
similar weakness. She had been taking
lipid lowering drugs for 10 years and
symptoms did not improve on their
withdrawal. Weakness was demonstra-
ble in the thoracic paraspinal and proxi-
mal limb muscles. Her CK was persist-
ently raised at 606 IU/l (after withdrawal
of atorvastatin). No acetylcholine receptor antibody was not detected. Spinal MRI showed mild
generative changes in the cervical spine
and normal thoracic spine. No specific
changes were seen in the paraspinal
muscles. Spontaneous activity consisting
of positive sharp waves, fibrillation poten-
tials, and complex repetitive dis-
charges were only seen in the right lower
cervical paraspinal muscles. Biopsy of
the rectus femoris muscle disclosed rare
nuclei and minimal fibre type grouping.
No specific diagnosis on the type of
myopathy could be made. Although she
subjectively felt slightly subjectively
worse at a review 9 months later there
was little change to her condition.

HEAD DROP
Table 1 shows the various neuromuscular
disorders reported to cause head ptosis.
Myasthenia gravis, especially in elderly
people and amyotrophic lateral sclerosis
are the commonest causes. The predilec-
tion of these diseases for focal or
segmental onset may explain this. Severe
isolated antecollis causing forced anter-
oexion of the neck occurs in parkinso-
nian syndromes. Rivest et al have docu-
mented antecollis in four of their
nerve biopsy cases of multisystem
atrophy (MSA). The abnormality de-
vvelops, sometimes in a subacute manner
over a period of weeks, in the late or
middle stages of the disease. The neck
can only be passively and forcibly ex-
tended to its normal position with diffi-
culty. None of the patients had convinc-
ding dystonic spasms of the anterior neck
muscles, although deeper muscles may be
involved. Speech, swallowing, and
upgaze deficits are often associated.
Trials of botulinum toxin into both ster-
noclidomastoid muscles are ineffective
and response to levodopa generally
disappointing.

Yoshiyama et al12 published a series of
seven patients with parkinsonism exhib-
iting head drop, four of whom carried a
diagnosis of probable MSA. Although at
rest there was no anterior neck spasm,
they attempted to extend the head voluntarily
or passively was accompanied by con-
thracis of the sternocleidomastoid mus-
cles on surface EMG in all of these
patients. There was no correlation be-
tween the severity of the head ptosis and
parkinsonism. Three patients had im-
provement of neck symptoms with treat-
ment of parkinsonism, whereas one
worsened. A recent report described
head ptosis in seven out of 459 patients
evaluated for parkinsonism. All seven
had features of MSA. Interestingly these
patients were also documented to have
myopathic changes in the paraspinal
muscles. In nine and positive sharp waves in one
three patients with parkinsonism who also
reported in the neck extensor muscles of
patients with parkinsonism. Okamiya et al
reported a patient with vascular parkinso-
nism and neck extension weakness who had myopathic changes in the
extensor muscles. One of our patients
(listed JHH case 2 in table 3), who has some clinical evidence of MSA, showed
mixed myopathic and neurogenic
features in the paraspinal muscles.

IDIOPATHIC HEAD DROP
In some cases, no obvious aetiology for
the head drop is apparent even after
extensive evaluation and on prolonged
follow up.13-15 Combining the data from
various reports of idiopathic head ptosis
in the literature and patients seen at our
institution (table 3), the following profile
of idiopathic head ptosis emerges. The
female to male ratio is 3:2. Mean and
median ages at presentation are 74.5 and
73 respectively. The onset is often sub-
acute, over days and weeks rather than
months. Two of our patients and one that
was reported by Lerman14 had a history
of significant weight loss. Discomfort at
the back of the neck is reported in some
patients but other sensory abnormalities
are not prominent. The weakness is pro-
found in the neck extensors, and involve-
ment of contiguous proximal muscles is
at most modest. The CK was normal in
all but two patients. Katz et al16 reported
oedema-like changes and atrophy of the
neck extensor muscles on MRI. An EMG
of the paraspinal muscles was remark-
able for the increased reports of sponta-
neous activity. Fibrillations were present
in nine and positive sharp waves in one
out of the 15 patients for whom EMG
data are available. Interestingly, such
abnormalities were pronounced in the
lower cervical segments; and in one case
reported by Katz et al17 extended to the
midthoracic region. The motor unit
action potentials in most cases are
described as having short duration and
small amplitude with early recruitment.
However, this has to be interpreted with
cautions as paraspinal motor unit action
potentials are generally small and may
have multiple turns even in normal indi-
viduals. EMG of the limb muscles is
mostly unremarkable. The histology of
limb muscles is normal or reveals mild
non-specific myopathic changes such as
increased fibre size variability. In two of
our cases there is some evidence of den-
ervation. The paraspinal muscle biopsies
show less subtle but again non-specific
changes such as fibrosis, increased fibre
size variability, “moth eaten” fibres, and
myofibrillar disarray. Degeneration, re-
generation, and necrosis were reported
by Jaster et al18 and also seen in one of our
patients. Interestingly, as mentioned
above, similar biopsy findings have been
reported in the neck extensor muscles of
patients with parkinsonism. Patients who also
have head drop,15 19 suggesting that these
changes may not be specific for a neuro-
muscular disorder.

The weakness remains localised and
in most cases stabilises after a short
period of progression. In some patients
(including case A) there is some
documented recovery. Secondary skel-
etal deformity can result in disability
even after good recovery of neck
strength,19 highlighting the importance
of maintaining good neck posture during
the period of weakness. Immunosup-
pressive agents have been tried for vary-
 ing periods with disappointing results
overall. Rose et al10 reported a positive
effect with prolonged corticosteroids, but
their patient had generalised weakness
and inflammatory infiltrates (albeit
scanty) in one biopsy. There may be a
role for cervical spine fusion in allowing
patients with disabling and non-
recovering head drop to maintain a
functionally useful head position.

Camptocormia
The causes of camptocormia are summa-
rised in table 2. As in head drop, several
<table>
<thead>
<tr>
<th>Report</th>
<th>Sex</th>
<th>Onset age</th>
<th>Weakness</th>
<th>CK IU/l</th>
<th>Paraspinal mus EMG</th>
<th>Limb EMG</th>
<th>Cx (cervical) paraspinal bx</th>
<th>Limb mus bx</th>
<th>Radiological examination Cx (cervical spine)</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lange1 (3 patients)</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>*Non-diagnostic</td>
<td>*Non-diagnostic</td>
<td>-</td>
<td>Unchanged 3–5 y</td>
</tr>
<tr>
<td>Suarez1 case 1</td>
<td>M</td>
<td>71</td>
<td>2 months</td>
<td>Mild shoulder weakness</td>
<td>N</td>
<td>Fibs, small amp, short duration MUAP, early rec. at lower Cx</td>
<td>N</td>
<td>Biceps: ↑ fibre size variability</td>
<td>Degen</td>
<td>Unchanged 5 months</td>
</tr>
<tr>
<td>2</td>
<td>F</td>
<td>81</td>
<td>3 months</td>
<td>Mild deltoid, neck flexor weakness</td>
<td>N</td>
<td>Fibs, small amp, short duration polyphasic MUAP at Cx</td>
<td>-</td>
<td>-</td>
<td>Deltoid: type II atrophy</td>
<td>N</td>
</tr>
<tr>
<td>3</td>
<td>F</td>
<td>68</td>
<td>3 months</td>
<td>Mild weakness</td>
<td>N</td>
<td>Fibs, small MUAP, early rec at lower Cx</td>
<td>Early rec, sternomastoid mus</td>
<td>Fibre size variability; NADH: moth-eaten; GT: 2 fibres with vacuoles</td>
<td>-</td>
<td>N</td>
</tr>
<tr>
<td>4</td>
<td>F</td>
<td>63</td>
<td>several months</td>
<td>Mild deltoid weakness</td>
<td>N</td>
<td>Fibs, short duration MUAP, early rec at Cx on Th</td>
<td>-</td>
<td>-</td>
<td>Biceps: type IIatrophy</td>
<td>N</td>
</tr>
<tr>
<td>Katz2 case 1</td>
<td>F</td>
<td>85</td>
<td>6 weeks</td>
<td>Mild neck flexion and deltoid weakness</td>
<td>N</td>
<td>Fibs, short duration MUAP, early rec at lower Cx to mid-Th</td>
<td>N</td>
<td>Fibre size variability, ↑ internal nuclei and fibrosis</td>
<td>Deltoid: N</td>
<td>Mild spondylosis, no neural impingement</td>
</tr>
<tr>
<td>2</td>
<td>M</td>
<td>64</td>
<td>1 week</td>
<td>Nil</td>
<td>N</td>
<td>Fibs, short duration MUAP, early rec. at lower Cx to Th</td>
<td>N</td>
<td>Fibre size variability, split fibres; ↑ internal nuclei and fibrosis, myotubular disarray in hypertrophic fibres</td>
<td>Biceps: N</td>
<td>&quot;</td>
</tr>
<tr>
<td>3</td>
<td>F</td>
<td>84</td>
<td>3 months</td>
<td>Nil</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>Fibre size variability, central NADH staining</td>
<td>Deltoid: N</td>
<td>&quot;</td>
</tr>
<tr>
<td>4</td>
<td>M</td>
<td>65</td>
<td>2 months</td>
<td>Nil</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>Fibre size variability, ↑ internal nuclei and fibrosis</td>
<td>Deltoid: N</td>
<td>&quot;</td>
</tr>
<tr>
<td>Khella3</td>
<td>F</td>
<td>84</td>
<td>-</td>
<td>Nil</td>
<td>N</td>
<td>Short duration, polyphasic MUAP, early rec</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>Stable after 5 y</td>
</tr>
<tr>
<td>Jaster4</td>
<td>M</td>
<td>69</td>
<td>4 h</td>
<td>Nil</td>
<td>N</td>
<td>Small amp, short duration MUAP at lower Cx</td>
<td>N</td>
<td>Degen, regen, and necrotic fibres</td>
<td>-</td>
<td>N</td>
</tr>
<tr>
<td>JHH, case 1</td>
<td>M</td>
<td>80</td>
<td>3 weeks</td>
<td>Nil</td>
<td>N</td>
<td>PSW in Cx 5 and Cx 8</td>
<td>N</td>
<td>Neurogenic and some myopathic changes</td>
<td>-</td>
<td>N</td>
</tr>
<tr>
<td>2</td>
<td>M</td>
<td>79</td>
<td>1–2 days</td>
<td>Mild weakness of neck flexion and shoulder abduction</td>
<td>N</td>
<td>Small amp, short duration MUAP, early rec at Cx 7–Cx 8</td>
<td>Small amp, short duration MUAP</td>
<td>Neurogenic changes</td>
<td>Biceps: neurogenic changes</td>
<td>Multilevel degen</td>
</tr>
<tr>
<td>3</td>
<td>F</td>
<td>81</td>
<td>“Days”</td>
<td>Mild ptosis</td>
<td>N</td>
<td>1+ fibs, PSW</td>
<td>N</td>
<td>-</td>
<td>Deltoid: N</td>
<td>Mild degen</td>
</tr>
<tr>
<td>4</td>
<td>F</td>
<td>70</td>
<td>“Days”; pain at onset</td>
<td>Mild proximal weakness</td>
<td>N</td>
<td>N</td>
<td>Few small amp, short duration MUAP</td>
<td>Non-specific myopathic changes</td>
<td>Quadriiceps: mild neurogenic changes, type II atrophy</td>
<td>N</td>
</tr>
<tr>
<td>5</td>
<td>F</td>
<td>73</td>
<td>“Weeks”</td>
<td>Nil</td>
<td>N</td>
<td>Small amp, short duration MUAP, early rec</td>
<td>N</td>
<td>-</td>
<td>Trapezius: type II atrophy</td>
<td>-</td>
</tr>
</tbody>
</table>

CK, creatine kinase; mus, muscle; bx, biopsy; EMG, electromyogram; PSW, positive sharp waves; Fibs, fibrillation; amp, amplitude; MUAP, motor unit action potential; rec, recruitment; regen, regenerative changes; degen, degenerative changes; Cx, cervical segment; Th, thoracic segment; NADH, nicotinamide adenine dehydrogenase; GT, gomori-trichome; *site not indicated; "–", not done/information not available; JHH, Johns Hopkins Hospital.
neuromuscular diseases can present with segmental involvement of the postural muscles, resulting in camptocormia.

**Amyotrophic lateral sclerosis (ALS)**
The kyphoscoliotic posture along with head drop is typical in patients with moderately advanced ALS. Involvement of paraspinal muscles was described by Gower in his account of patients with ALS. In most cases, the wasting invades the muscles of the back, and it sometimes begins in them. Our group had previously reported a patient with ALS who developed severe camptocormia, requiring him to support himself with hands on his thighs. Limb muscles were less involved in this patient.

**Myopathy**
Most generalised myopathies, because of pelvic girdle weakness, cause lumbar lordosis rather than kyphosis. The exception may be inclusion body myositis (IBM), which was reported as a cause of pelvic girdle weakness by Hund et al. A 70 year old man developed back and later leg weakness over a period of months. The CK was slightly raised. An EMG showed spontaneous activity and polyphasic motor units with reduced recruitment in the paraspinal muscles. A biopsy of a relatively non-atrophic part of the paraspinal muscles was reported to show morphology typical of IBM. The predisposition for focal muscle weakness may explain the occurrence of camptocormia in IBM. Alternatively it may occur as a secondary phenomenon. The preferential weakness of the quadriceps over the iliopsoas often forces the patient to lock the knee in extension and predisposes to a bent over posture for better balance. This may stretch the back muscles excessively, putting them in a position of mechanical disadvantage and in turn lead to further bending. Katz et al has proposed an important role for mechanical factors in the evolution of head drop. A similar pathophysiology may cause camptocormia in IBM. Nemaline myopathy involves the paravertebral muscles and can cause kyphoscoliosis or lordosis. This congenital myopathy can rarely present in adult life. A case of a 62 year old woman with adult onset nemaline myopathy who presented with severe camptocormia has been reported. Furthermore, nearly half the cases of adult onset nemaline myopathy in the literature have been associated with head drop. We think that facioscapulohumeral muscular dystrophy has not been previously reported as a cause of camptocormia (case C). Interestingly in the series of Lange et al on head ptosis there was one patient with facioscapulohumeral muscular dystrophy. In the series of Serratrice et al one patient had type I predominance (in quadriceps/deltoid muscle biopsy) and fingerprint morphology on ultrastructure, suggestive of a congenital myopathy.

**Extrapyramidal disorders**
A mild stooped posture is a hallmark of parkinsonism, but severe anterior curvatura is not common. Djaladetti et al described eight patients with presumed idiopathic Parkinson’s disease, at Hoehn and Yahr stage 2 to 4, mean age of 66±5, and symptom-duration of 13.1±5.1 years who had severe forward flexion of the thoracolumbar spine. This was apparent on standing, less obvious while sitting, but disappeared in the supine position. There was no mention of the nature of onset. The EMGs of the paraspinal muscles done in five patients were normal. Three patients apparently had immediate and reversible aggravation of the bent spine with levodopa therapy whereas the other two had improvement. The authors postulated that camptocormia in parkinsonian’s disease may represent a type of dystonia.

Postencephalitic parkinsonism has also been reported to cause curvature of the spine. Onuaguluchi reported five cases in 1964, followed a year later by a series consisting of 14 cases from Martin. The latter included seven photo illustrated case histories. The spinal curvature in these patients was in both anteroposterior as well as lateral planes and diminished in the supine position. In many patients it was noted to extend “from sacrum to neck”. In one woman the forward flexion of the neck was only seen when she was blindfolded. There was no consistent relation of the spinal curvature to the side of greater muscular rigidity. Three patients underwent stereotactic surgery. One patient received lesions to the ventrolateral thalamus and pallidum and another to the pallidum alone, ipsilateral to the concavity of the spine. A third patient received a lesion to the pallidum contralateral to the side of spinal curvature. In all the cases rigidity and tremor on the opposite side was abolished; but the spinal curvature was corrected only in the cases where the stereotactic lesion was placed ipsilateral to the concavity of the spinal curvature. Animal work by Ferrier, Demas-Mansat, and Hassler had previously shown that unilateral caudal lesions could cause a similar deformity of the spine in animals, which can be corrected by destruction of the contralateral head of the caudate nucleus. Severe pathologic changes have been described in the caudate nucleus in postencephalitic parkinsonism. Martin hypothesised that tonic activity in the caudate nucleus and its efferent pathways to the pallidum might play an important part in maintaining spinal posture.

The slowly progressive and chronic forms of a rare encephalomyelitis that occurs among the Lakut people of Siberia, termed Viliuisk encephalomyelitis, is also characterised by a forward bent posture. Other features include rigidity, bradykinesia, dementia, and dystarthis. Pathological findings include multiple areas of inflammation, necrosis, perivascular leucocytic cuffing, spongiform changes, and fibrosis as well as marked atrophy of the basal ganglia.

Camptocormia was reported as a side effect of sodium valproate toxicity in a 23 year old epileptic patient. Interestingly the deformity resolved when the plasma valproate level dropped to about 300 μmol/l, only to return 4 months later when the plasma concentration rose to about 330–530 μmol/l. This patient had no other extrapyramidal features. Reversible parkinsonism has been well documented as a rare side effect of valproate and an extrapyramidal mechanism might have been responsible for the effect of valproate on spinal posture.

**IDIOPATHIC CAMPTOCORMIA**
In some cases of camptocormia, such as head ptosis, the clinical abnormality remains confined to the back and no apparent cause is found even on prolonged follow up. Is idiopathic camptocormia a distinct entity? Table 4 summarises the various reports on this condition. The data from these studies were combined with our cases B and D to develop a profile of idiopathic camptocormia. The incidence of camptocormia, like head drop, increases with advancing age. All but eight patients developed it at an age older than 60. The earliest age of onset is 49 years. The male to female ratio is about 3:1. The weakness is mainly confined to the extensor muscles of the spine. Onset and progression are more chronic compared with head drop, often extending over a period of months to years. Mild back discomfort was present in a few. Sensory symptoms were absent. The presence of a family history in some patients suggests that investigations for genetic and congenital neuromuscular conditions such as facioscapulohumeral muscular dystrophy and nemaline myopathy might result in better characterisation of the idiopathic cases. The CK is raised in cases B and D and in some of the cases described by Hilliquin et al and Serratrice et al. The paraspinal muscles have been reported to show atrophy and a heterogeneous appearance on radiological examination (CT or MRI). The EMG findings are not uniform, with reports of both myogenic and neurogenic features. In case D, spontaneous activity consisting of positive sharp waves, fibrillation potentials, and complex repetitive discharges is seen only in the right lower cervical paraspinal muscles, probably an incidental finding.
Table 4  Case reports and series of idiopathic camptocormia

<table>
<thead>
<tr>
<th>Report (first author)</th>
<th>Sex</th>
<th>Age, mean/ range</th>
<th>Weakness</th>
<th>Onset Pattern</th>
<th>CK IU/l</th>
<th>Paraspinal BMG:</th>
<th>Limb BMG</th>
<th>Lumbar paraspinal mus bx</th>
<th>Other mus bx</th>
<th>Radiological examination, paraspinal mus</th>
<th>Radiological examination, lumbar spine</th>
<th>Miscellaneous</th>
</tr>
</thead>
<tbody>
<tr>
<td>Delisle(^\text{a})</td>
<td>M</td>
<td>57–75</td>
<td>Few weeks–9 y</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>Fibrosis</td>
<td>–</td>
<td>Low density heterogeneous appearance</td>
<td>–</td>
<td>–</td>
</tr>
<tr>
<td>Laroche(^\text{b})</td>
<td>M</td>
<td>63.5/51–81</td>
<td>Mild weakness of other mus in 2</td>
<td>N</td>
<td>Neurogenic in 5, myopathic in 11, &quot;uninterpretable&quot; in 16</td>
<td>Deltoid, gluteal: myopathic in 6, distal leg: neurogenic in 5</td>
<td>Deltoid N in 5</td>
<td>Fibrosis, mod inflam infiltrate in 1</td>
<td>Atrophy, fibrosis; mod inflam infiltrate in 6</td>
<td>Lumbar disc arthrosis, spondylolisthesis in 5</td>
<td>–</td>
<td>Family history of similar deformity in 2</td>
</tr>
<tr>
<td>Hillquin(^\text{c})</td>
<td>M</td>
<td>61–86</td>
<td>No other weakness</td>
<td>N</td>
<td>Neuropathic in 4, –</td>
<td>Neurogenic in 1, –</td>
<td>Atrophy, fibrosis; mod inflam infiltrate in 6, necrotic fibres in 3</td>
<td>–</td>
<td>Atrophy</td>
<td>–</td>
<td>–</td>
<td></td>
</tr>
<tr>
<td>Ehrenstein(^\text{d})</td>
<td>F</td>
<td>75</td>
<td>6 months</td>
<td>N</td>
<td>Small, short MUAP with CRD at lumbar segment</td>
<td>Quadriceps; type II atrophy</td>
<td>–</td>
<td>–</td>
<td>Atrophy and hypodensity</td>
<td>–</td>
<td>–</td>
<td></td>
</tr>
<tr>
<td>Poullin(^\text{e})</td>
<td>F</td>
<td>63–64</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td></td>
</tr>
<tr>
<td>Pennison-Besnier(^\text{f})</td>
<td>F</td>
<td>72</td>
<td>16 months</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>–</td>
<td>Atrophy and hypodensity</td>
<td>–</td>
<td>–</td>
<td></td>
</tr>
<tr>
<td>Serratrice(^\text{g})</td>
<td>M</td>
<td>60/49–75</td>
<td>–</td>
<td>–</td>
<td>Myopathic in 5, neurogenic and neuropathic in 1, N in 2</td>
<td>Fibrosis in 2, necrosis and regeneration in 1, &quot;fingerprint&quot; in 1</td>
<td>–</td>
<td>–</td>
<td>Areas of hypodensity</td>
<td>–</td>
<td>Family history of similar deformity in 2</td>
<td></td>
</tr>
</tbody>
</table>

N, normal; ↑, increased; ↓, decreased; mod, moderate; CK, creatine kinase; mus, muscle; bx, biopsy; BMG, electromyogram; PSW, positive sharp waves; fibs, fibrillation; amp, amplitude; MUAP, motor unit action potential; rec, recruitment; CRD, complex repetitive discharges; degen, degenerative changes; regen, regenerative changes; inflam, inflammatory; “–”, not done/information not available.
increasing age of patients in disease and control biopsies.55 Hence, it may be difficult to make a specific aetiological diagnosis by studying the paraspinal muscles alone.

Probable neuropathic contribution to the pathophysiology of idiopathic head drop and camptocormia

Pennison–Besnier et al56 described a 72 year old woman with a 16 month history of camptocormia, whose lumbar paravertebral muscle showed fibrosis, fatty infiltration of interfascicular septa, groups of angular atrophic fibres, nuclear bags, fibre type grouping on ATPase stain, core, and target fibres. They think that denervation of the paraspinal muscles from impingement of the dorsal rami by osteoarthritic facet joints causes camptocormia. They asserted that the paraspinal muscle biopsies of other reports and two of their patients might not have shown typical neurogenic changes because they were performed late in the course of the illness. However, as already mentioned, neurogenic changes on muscle biopsy are common in elderly people and may not be clinically significant.56 57 This is especially true in the cervical and lumbar paraspinal muscles, where there is a high incidence of degenerative vertebral disease in elderly people. Furthermore, to cause significant weakness of spinal extension large segments of the paraspinal muscles need to be denervated as sectioning of C1-C6 dorsal branches in patients with spasmodic torticollis58 does not cause any functional impairment. In addition, although spondylodiscitis could conceivably damage neural elements by vascular compromise, either from arterial ischaemia at the watershed region of the spinal arteries of Adamkiewicz or venous congestion, direct impingement of spinal roots have not been reported in patients with idiopathic head ptosis and camptocormia (tables 3 and 4). Therefore it is more likely that chronic denervation of the paraspinal muscles is a contributing factor rather than the basis for the development of idiopathic head ptosis and camptocormia.

The presence of pain, relatively acute onset, and recovery of head drop over a period of weeks in some patients is reminiscent of the clinical course of brachial neuritis (Parsonage-Turner syndrome). Indeed the clinical diversity of this condition has been well documented,59 60 including involvement of structures as proximal as the roots. Kidron et al had reported C6 root involvement with paraspinal denervation.61 Whether a similar process can selectively involve the cervical plexus and the dorsal spinal nerves can only be confirmed by studying the histology of paraspinal muscles and dorsal spinal nerves early in the course of the illness.

HEAD PTOSIS AND CAMPTOCORMIA: PARTS OF A SPECTRUM?

In most instances head drop and camptocormia are not related. It is rare to find a patient with both disorders except in extrapyramidal disorders such as MSA and postencephalitic parkinsonism.21 However, the similarities between them (table 5) suggest that both head ptosis and camptocormia may occur as a result of similar pathophysiological processes affecting paraspinal muscles of different parts of the spine. In any individual patient local factors such as chronic denervation and loss of tissue elasticity may determine the spinal segment involved.

In summary, head ptosis and camptocormia include a diverse group of disorders that share in common involvement of different parts of the spine resulting in disabling anterior curvature of the spine. In addition to the known causes of flexion deformity of the spine, such as myasthenia gravis, amyotrophic lateral sclerosis, and Parkinson's disease, multiple cumulative age related factors such as loss of muscle tone, tissue elasticity, chronic stretch and denervation of the paraspinal muscles from degenerative spinal disease, may be involved in the pathogenesis. In practice, neuromuscular and extrapyramidal disorders, because of therapeutic and prognostic implications, should be ruled out in all patients. Measures to prevent fixed deformities—for example, passive mobilisation of the spine and the use of supports such as corsets and stiff collars—should be instituted early. At least some patients with idiopathic head drop recover partial function spontaneously. There is no clearly defined role for immunosuppressive or other pharmacotherapy. The case report of valproate induced reversible camptocormia introduces the intriguing possibility of using a GABA agonist in cases with an apparent extrapyramidal aetiology. Patients whose head ptosis does not improve should be considered for cervical fusion, which allows the patient to maintain a functionally useful head position.

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We thank Dr R Kurtzke, Fairfax, Virginia for providing us with information on his patient and valuable comments, and Dr M P T Lunn, Guy's, St Thomas', and King's School of Medicine, London, for contributing important ideas. Competing interests: none declared

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Authors' affiliations
T Umapathi, Department of Neurology, National Neuroscience Institute, Singapore
V Chaudhry, D Cornblath, D Drachman, J Griffin, R Kundi, Department of Neurology, Johns Hopkins University School of Medicine, Baltimore, USA
D Drachman, J Griffin, Department of Neuroscience, Johns Hopkins University School of Medicine, Baltimore, USA
J Griffin, R Kundi, Department of Pathology, Johns Hopkins University School of Medicine, Baltimore, USA

The second to sixth authors contributed equally to this work.

Correspondence to: Dr T Umapathi, Department of Neurology, National Neuroscience Institute, Singapore 308433; tumapathi@yahoo.com

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Head drop and camptocormia

T Umapathi, V Chaudhry, D Cornblath, D Drachman, J Griffin and R Kuncl

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Measuring carotid stenosis

Comparing a new test with a standard involves measuring disagreement. In the case of measuring carotid artery stenosis, some of the disagreement between different tests is because of inherent differences in how the stenosis is demonstrated (test characteristics). This is what we are most interested in when assessing a new technology. However, some of the disagreement simply reflects variability in how we physically make the measurement with the standard technique. Choosing the point of maximum stenosis, choosing the point in the common carotid artery for use as a denominator, measuring from an eyepiece, or measuring from calipers all introduce variation when measuring carotid stenosis. The resulting observer variabili- ty in reporting contributes to disagreement between methods but to some extent is inde- pendent of the method used to generate the angiogram in the first place.

In the medical literature, disagreement between methods is often attributed entirely to test characteristics, with little appreciation of the role of observer variability in reporting. When one method is compared with another and agreements emerge, it is not readily apparent how much of the disagreement is caused by the method used and how much by the process of measurement, unless observer variability data are also presented. The recent paper from Patel et al, interobserver variability data are presented but their signifi- cance in relation to overall agreement does not appear to have been appreciated. Using the data from Patel et al (tables 2 and 4) for symptomatic carotid arteries, it is noted that when 34 carotid digital subtraction angiograms (DSA) are measured by one radiolo- gist, there was disagreement in seven cases when the same films were reported by a second radiologist. Therefore if only DSA was used, seven patients would have had “inap- propriate” surgery according to which radiologist read the angiogram. This is not surprising, and such disagreement is a con- sistent finding in observer variability studies.1 Observer variability in reporting DSA therefore accounted for approximately 20% of disagreement in this particular series of angiograms. This sets a limit on the maximum agreement that any alternative method can demonstrate when compared with DSA. It is clearly not reasonable to expect better agreement from another method than can be obtained by re-reporting the DSA films themselves. In Patel’s table 2, when the same arteries are assessed by computed tomo- graphic angiography (CTA) there was discrep- ancy with DSA in seven cases, while with magnetic resonance angiography (MRA) and ultrasound there was disagreement in six and seven cases, respectively. The three alter- natives thus disagree with DSA to the same extent as can be attributed to observer disagreement in reporting DSA. Put simply, the same number of missed or unnecessary operations would have occurred (roughly 20% in this series) whatever method was used, including DSA alone. Observer variability is

not confined to DSA, and the scatter plots from Patel et al (fig 2) would suggest—in keeping with other studies—that observer variability is greater for MRA and CTA than for DSA.2 It is surprising that this did not translate into more clinically important disa- greements when MRA and CTA are compared with DSA. This is probably accounted for by the fact that in this study, for MRA and CTA, consensus views were taken for any disagreements greater than 10% between observers.

This highlights the important point that combining multiple observations made on the same data will reduce observer variability, and ultimately improve agreement with other methods. Partly for this reason, but also because to some extent the strengths and weaknesses of CTA, MRA, and duplex ultra- sound are complementary, we would suggest that a combination of tests (we use the combi- nation of ultrasound and MRA) should be used in preference to DSA.

What is clear from this study is that most of the disagreement arises from different meth- ods of measuring carotid stenosis can be attributed to observer variability in reporting rather than to the test characteristics of the individual methods themselves. The 10% of patients injured as a result of DSA in this study, and those who continue to be put at risk from catheter angiography in these circumstances, would be quite entitled to ask why they are exposed to a procedure which appears to offer no great advantage over safer alternatives. We suggest that more studies are not required, simply more thorough under- standing of presently available information.

G Young
Middlesbrough General Hospital, Ayresome Green Lane, Middlesbrough TS5 5AZ, UK

P Humphrey
The Walton Centre, NHS Trust, Liverpool, UK

Correspondence to: Dr G Young; gavin.young@sitees.nhs.uk

References

Author’s reply

Doctors Young and Humphrey highlight that differences between tests arise from several factors, some of which are inherent in the test and some of which arise from aspects attributable to observer variation. Some of the aspects to do with observer variation apply to interpretation of all tests and some are specific to certain tests. In our study we were endeavouring to quantify the effect on patient management if non-invasive tests were used instead of intra-arterial angiography to assess carotid stenosis. Our study has several limita- tions, including a relatively small sample size and the fact that we were not able to get all scans read by all observers but rather had to get pairs of observers to concentrate on read- ing only CTA, or MRA, or DSA. A better design would have been to keep the same workers together in pairs but randomly assign the CTA, MRA, or DSA films to each pair. As it is, it is possible that some of the apparent differ- ence between imaging modalities is specific to the pair of observers, not to the modality. However, imaging studies are difficult to fund and expensive to do, and the result and design of our study was a compromise involving all these factors.

We identified that the observer reliability of CT angiography or MR angiography was worse than that for digital subtraction angiogra- phy, as highlighted by Drs Young and Humphrey. Also in general there was more variation between the observers for the reading of asymptomatic stenoses than for sympto- matic stenoses (emphasising the importance of considering patient characteristics, not just the imaging technique). In the determination of the effect that this disagreement might have on patient management, we used homo- grams derived from the European carotid sur- gery trial which were based on intra-arterial angiographic measurement of stenosis. We therefore had to use the comparison of non-invasive test reading with DSA rather than being able to use the individual observ- ers readings of non-invasive tests. Thus as Drs Young and Humphrey point out, the actual effect of using non-invasive tests maybe worse than we have estimated.

Finally, Drs Young and Humphrey suggest that more studies are not required but we are not entirely sure that that is completely true. Non-invasive imaging tests are continually undergoing modifications, may become more accurate, more reliable, less risky and more convenient, and may be improvements in accuracy or practi- cality, but this cannot be assumed to be the case. Much of this tinkering with technology is driven by the manufacturer’s desire to encourage purchase of new machines. Improvements have also occurred in intra-arterial angiography with smaller and more manoeuvrable catheters and greater aware- ness of the risks, which may have helped to reduce the risk of angiography. One “shot” of CTA, MRA, and ultrasound is already out of date because contrast MRA is now increasingly used. While we would hope that non-invasive tests (probably in combination rather than alone) would eventually replace intra-arterial angiography in the majority of patients being considered for carotid inter- vention, we feel it likely that there will always be a need for some intra-arterial angiography for specific cases, or depending on local resources. In any case DSA did not prove less popular than MRA among the patients in our study. There is certainly room for much more in depth examination of existing data but we shouldn’t close the door on the need for further studies.

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Cerebral metastasis after primary renal cell carcinoma

The article by Roser et al., in which it was shown that the treatment of intracranial metastases originating from renal cell carcinoma can on occasion be successful, was most interesting.

We have followed the clinical course of a patient with a renal cell carcinoma with a low mitotic index since 1989. In this patient the course was distinctly more malignant but the disease has also been successfully treated to date. In the last 13 years, this patient has had four metastases surgically removed and a further nine treated with stereotactically guided percutaneous single dose convergent beam irradiation therapy (stereotactic modified linear accelerator, 6–15 MV photons, 18–20 Gy prescribed to the 80% isodose). Apart from slight mnemonic deficits, the patient is in good health.

The following factors which affect the prognosis were all met by our patient:

• The interval between the diagnosis of renal cell carcinoma and the first detected brain metastasis exceeds 17 months (our patient, 18 months; the patient described by Roser et al., 36 months);
• Age below 60 years at the time of initial diagnosis;
• Primary tumour of the left kidney, initial nephrectomy;
• Diameter of primary metastasis < 2 cm;
• Not more than one brain metastasis at the time of initial treatment;
• Solely intracranial metastases;
• Karnofsky > 70%;
• No systemic symptoms such as fever or weight loss at the time of diagnosis;
• Blood sedimentation rate under 50 mm/h at diagnosis of renal cell carcinoma.

Patients in whom prognostic factors predict a good outcome should be treated with intent to cure.

References


BOOK REVIEWS

Seizures, medical causes and management


This book is unusual among books about seizures because it focuses on acute symptomatic (“situation-related”) seizures, rather than “epilepsy” (although there is inevitably some overlap between the two). It provides definitions and describes the epidemiology and pathophysiology of acute symptomatic seizures in the initial section, which is followed by chapters detailing the specific circumstances in which such seizures are likely to occur, often (although not invariably) including points of management specific to the situation. Subjects covered include seizures occurring in the context of multisystem disease, infection, hypoxic-ischaemic cardiopulmonary conditions, endocrine disorders, cancer, and other conditions. Situation-related seizures occurring as a result of drugs or alcohol misuse are also addressed, as are those occurring in the intensive care situation, and the difficult, but important, differentiation of seizures from syncope. The book ends with a very practical chapter entitled “Anticonvulsants in acute medical illnesses”, in which the considerations affecting the choice of antiepileptic drug in the acute situation are reviewed.

Although situation-related seizures are usually discussed in books about epilepsy, they do appear to constitute a distinct group in a number of respects including prognosis. To a certain extent the topics discussed in the book form a rather disparate group linked in which it was thought that the editors have the energy to produce a third edition, there is (as always) some room for improvement. The series of chapters on infarcts in specific subcortical territories would be enhanced by some figures illustrating the vascular anatomy that is discussed in the text. In addition, the quality of the discussion of study methodology varies considerably between chapters, and some would benefit from a more systematic and accurate approach to statistical and epidemiological concepts.

Medical risks in epilepsy


This is a very useful, reasonably comprehensive yet succinct multiauthor small book on medical risks associated with epilepsy. Areas covered include methodological aspects; accidents and risks in everyday life; traffic accidents; driving regulations; mortality, including SUDEP; psychiatric comorbidity and suicide; fatal adverse drug reactions reporting data (which are rather difficult to interpret); seizure-warning systems and risk prevention; as well as insurance related issues. It also highlights many areas where further research is required. The book generally provides an overview of the more recent research and publications in this area and includes some regulatory issues. Inevitably it has a Nordic emphasis; it includes very useful advice on precautionary measures to minimise risk of injury for people with uncontrolled epilepsy, including climbers and saunas. Some chapters, by necessity, serve purely as available incomplete data. Others are written by key researchers directly involved in the area addressed and provide a very balanced review of current knowledge. On psychiatric comorbidity, while agreeing that “the positive
effects of drug therapy on cognitive and affective functioning because of the reduction in seizure activity are usually far greater than the negative effects”, more information would have been welcome in an otherwise very well balanced chapter. The book would well serve those who wish for whom it is intended, namely, pathologists, neurologists, paediatric neurologists, psychiatrists, and other professionals who deal with patients with epilepsy. The editors rightly stress the “official line” that the majority of patients with epilepsy can achieve good control, with low associated risks.

Lina Nashef

Greenfield’s neuropathology,
7th edition


What can one say. The latest (7th) edition of Greenfield’s Neuropathology has hit the bookshops, and indeed what a resounding thud it makes! The present edition is bigger than ever, again running into two volumes, but now totalling a staggering 2330 pages and costing an equally staggering £395. It comes equipped with a handy CD version of the illustrations, a mere snip at £145.

The 7th edition has undergone considerable changes in content, since the last edition five years ago, reflecting the ever expanding increase in knowledge of diseases of the nervous system and muscle that has come from the exponential growth in neuroscience research over the past decade. Areas of cellular and molecular neurobiology, and the contributions that genetics and neuroimaging have made towards improving our understanding of the causes of disease and our clinical investigative and diagnostic skills, are more strongly featured. Hence, while greater emphasis has been placed on the basic science of disease, the classic descriptive morphology for which Greenfield is renown is well maintained. There are new chapters on “Metabolic and neurodegenerative diseases of childhood” and “Peripheral and mitochondrial diseases”. The chapter on “Pathology of schizophrenia” has been shrewdly expanded to cover “The pathology of psychiatric disorders”. Other chapters have been retained as such, but many have been rewritten with new authors reflecting the pre-eminence of each within their particular subspecialty. There is increased reliance on colour illustrations, line diagrams and tables to illuminate the text, and these are of excellent quality throughout. As to be expected, all chapters are written authoritatively with clarity and style, comprehensively illustrated, and lavishly referenced. Judging by the content of the chapters on ageing and dementia, prion disease, and movement disorders, it is my guess that if anything is not included in each chapter, it’s probably not worth including anyway. The accompanying CD rom is user friendly, and the images are downloadable—a boon to those wishing to produce a ready made lecture or presentation of distinction. The book is a must for practicing and trainee pathologists, but is equally compelling for workers in other clinical neuroscience disciplines and basic researchers interested in the roots of the dysfunctional nervous system. Possession of the 7th edition is guaranteed lasting quality and full value, but before lashing out make sure both your arms and shelves are strong enough to accommodate its presence.

David MA Mann

Smell and taste complaints


Despite the fact that problems with tasting and smelling are common in the general population, few physicians have the knowledge and training to authoritatively deal with them. Christopher Hawke’s Smell and Taste Complaints provides a straightforward guide to the understanding and management of chemosensory disturbances, reflecting the first clinically oriented book of its kind since Ellis Douek’s The Sense of Smell and its Abnormalities (Edinburgh: Churchill Livingstone, 1974). This 180 page pocket sized book provides a cogent overview of the anatomy and physiology of the olfactory and gustatory systems, practical approaches towards their assessment, and suggestions for therapy and management. Importantly, it provides the practitioner with the names and addresses of specialists or has even a casual interest in chemosensation, and should serve to elevate the level of appreciation of these senses within the medical community at large.

Richard L Doty

CORRECTIONS


We regret that an editing error occurred in the correspondence from Jaster JH, Doohan FC, and O’Brien TF. Demyelination in the brain as a paraneoplastic disorder: candidates include some cases of seminoma and central nervous system lymphoma. J Neurol Neurosurg Psychiatry 2002;73:332. The description of a patient expanded altered, in the first line of the fourth paragraph the text should read “...patient who had a non-neurological malignancy, seminoma, and subsequently developed a paraneoplastic syndrome...”.

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