Paraspinal neuromuscular syndromes

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Introduction

This article includes discussion of paraspinal neuromuscular syndromes, bent spine syndrome, floppy head syndrome, isolated neck extensor myopathy, and primary camptocormia. The foregoing terms may include synonyms, similar disorders, variations in usage, and abbreviations.

Overview

Neuromuscular syndromes of the paraspinal muscles comprise the dropped head syndrome and the bent spine syndrome (camptocormia). Although these phenotypes also occur in conjunction with dystonia or Parkinson disease, large series suggest that the majority of cases are due to an isolated paraspinal myopathy or are a prominent part of a more widespread myopathy, neuropathy, or motor neuron disease. Even camptocormia and dropped head syndrome in Parkinson disease are often due to myopathic changes in the paraspinal muscles. In this article, the author discusses advantages and disadvantages of paraspinal muscle biopsies for diagnostic purposes. In any event, histology has to be interpreted with caution because these particular muscles show a variety of neuromuscular abnormalities even in healthy subjects.

Key points

- Weakness of the paraspinal muscles may lead to dropped head syndrome or bent spine syndrome (camptocormia).
- The phenotypes of the dropped head and bent spine syndromes may be caused by many underlying etiologies, but the majority of cases seem to be due to an isolated paraspinal myopathy or provide a prominent sign of a more widespread myopathy.
- Clarifying the etiology in every single case has an impact on therapy because dystonic forms may respond to sensory tricks or botulinum toxin therapy, and parkinsonian forms may respond well to deep brain stimulation.

Historical note and terminology

In 1986, Lange and colleagues described severe neck extensor weakness, using the term “floppy head syndrome” (Lange et al 1986). The floppy head syndrome occurred as part of a generalized neuromuscular disorder, such as myasthenia gravis, polymyositis, or motor neuron disease, in 9 of 12 patients; in the other 3 there was no apparent cause for the neck weakness. In 1992, Suarez and Kelly elaborated on the same clinical manifestations, now favoring the words “dropped head syndrome” (Suarez and Kelly 1992). They described 4 patients with a noninflammatory myopathy primarily affecting the lower cervical and upper thoracic paraspinal muscles. The patients had mild limb girdle weakness in addition to severe neck weakness. A subsequent report suggested that an important cause of the head drop is an isolated myopathy of the neck extensor muscles (Katz et al 1996). These patients did not have extremity weakness, and using the term “isolated neck extensor myopathy,” a specific disorder was distinguished from other neuromuscular conditions that also cause neck weakness. In contrast, the terms “dropped head syndrome” or “dropped head sign” are best used to describe the phenomenology regardless of the underlying cause.

A related disorder of paraspinal muscles has been referred to as the “bent spine syndrome” or “camptocormia.” The latter is derived from the Greek: kamptos for bent and kormos for trunk. The French neurologist Souques first recognized this syndrome during World War I in soldiers who acutely developed an anteriorly flexed trunk (Karbowski 1999). That condition was thought to be psychogenic and lasted only a few months. In the last decades, however, camptocormia has been described in association with Parkinson disease (Djaldetti et al 1999), multiple system atrophy (Askmark et al 2001), dystonias (Reichel et al 2001), and other neuromuscular disorders marked by weakness of the
thoracic, lumbar, or sacral paraspinal muscles (Serratrice 1996; Umapathi et al 2002). As with the dropped head syndrome, the differential diagnosis in patients with a bent spine includes other generalized neuromuscular disorders and an isolated focal paraspinal myopathy.

In a large retrospective series of 63 patients with bent spine syndrome, 40 had isolated paraspinal myopathy with fatty infiltration and lobular endomysial fibrosis; 19 had more widespread signs of myopathy (including 8 cases of limb girdle muscular dystrophy, 3 myotonic dystrophy type I, 2 facioscapulohumeral dystrophy, and 2 inclusion body myositis); and 4 had Parkinson disease without paraspinal myopathy (Laroche and Cintas 2010). On the other hand, there is some evidence that camptocormia and head drop syndrome in Parkinson disease are predominantly myopathic. In a series of 17 Parkinson patients with camptocormia or head drop, histopathology revealed chronic myopathic changes in 14 of 17 biopsies consisting of abnormal variation in fiber size; increase in internal nuclei; and increase in connective tissue, myofibrillar disarray, and similarities to protein surplus myopathies (Spuler et al 2010). The authors speculate that aberrant protein aggregation may link Parkinson disease and camptocormia. It is not clear whether the classic “stooped” appearance of parkinsonian patients represents mild forms of antecollis/camptocormia. This uncertainty reflects the lack of a clear clinical definition and the different thresholds that physicians use for diagnosis. Most authors propose that a marked (minimum 45°) flexion in the sagittal plane should be required to diagnose antecollis and camptocormia, respectively. It seems likely, however, that the postural deformities in Parkinson disease have a multifactorial pathophysiology. Contributing factors include muscular rigidity, axial dystonia, weakness caused by myopathy, body scheme defects, and structural changes in the spine. Clarification of the relative contribution of these different factors may help to define clear diagnostic criteria and ultimately lead to improved treatment approaches (Doherty et al 2011).

**Clinical manifestations**

**Presentation and course**

**Isolated neck extensor myopathy.** This disorder typically occurs in individuals in the seventh decade or older. There is no predilection for men or women. In mild cases, the patient notices difficulty keeping the head erect, whereas in extreme cases, the neck extensors become so weak that the chin rests firmly against the chest wall when the patient is standing. Careful inspection shows that the head drop results from weakness that is maximal over the mid and lower cervical and the upper thoracic regions. Symptoms and signs usually develop relatively quickly, over a period of days to a few weeks, but they can progress for months. Patients may complain of dull or burning neck pain that accompanies the progressive phase. Others note difficulty walking or holding a conversation because of the inability to look ahead. Dysphagia is another common complaint that probably reflects interference with the swallowing mechanism by the flexed neck posture (Katz et al 1996). This difficulty is relieved by passive elevation of the head. There may be mild deltoid weakness (Suarez and Kelly 1992; Umapathi et al 2002), but deltoid muscle biopsies have been unrevealing and the significance of mild proximal limb girdle weakness in elderly people is uncertain. Obvious weakness outside the paraspinal muscles should raise suspicion that a neuromuscular condition other than isolated neck extensor myopathy is present.

**Primary camptocormia.** This also tends to affect elderly people (Serratrice et al 1996). Women are more commonly affected than men, and an affected family member (by some reports) can be identified in up to 60% of cases (Laroche et al 1995; Serratrice et al 1996). Weakness develops over 1 to 8 years and back pain is frequent (Reinsel 1995). The thoracic, lumbar, or sacral regions may be involved alone; in some cases wide regions of the spine, including cervical muscles, are involved. Some authors believe that isolated neck extensor myopathy and primary camptocormia are variants of the same disease (Oerlemans and de Visser 1998; Swash 1998). The weakness results in an inability to stand upright. This is exaggerated by activities that require back extension, such as walking up an incline or taking off a shirt when the skin is damp. There may be proximal limb weakness, more commonly in the pelvic than the scapular region (Laroche 1995). However, as with isolated neck extensor myopathy, a generalized phenotype should lead to consideration of some other neuromuscular disease. The clinical course is usually benign and patients remain ambulatory with the use of a cane.

**Prognosis and complications**

Isolated neck extensor myopathy and primary camptocormia are essentially static conditions. Rest-related improvement has been observed in a case report of isolated neck extensor myopathy (Oishi et al 2000).
Clinical vignette

Isolated neck extensor myopathy. A 77-year-old man developed insidiously progressive difficulty breathing because of congestive heart failure. As a result, he began to sleep sitting in an easy chair. Within weeks he noticed a constant dull pain in the back of his neck. A few days later he had difficulty keeping his head in the upright position. Muscle biopsy of the cervical paraspinal muscles revealed increased connective tissue, necrotic myofibers, split fibers, and increased internal nuclei without inflammation or vacuoles.

No medical therapy was prescribed. The patient complained of discomfort when he wore either a firm or soft cervical collar because they rubbed against his chin when he talked, ate, or turned his head. Similarly, he found several braces uncomfortable. He also tried to avoid spending time in the upright position where the head would fall forward and, once his cardiac problems were treated, he slept in a reclined position rather than seated. He improved slightly over the following year.

Primary camptocormia. For 3 years, a 49-year-old woman noted progressive anterior curvature of the spine, most evident over the lumbar region. She had mild low back pain that increased with prolonged standing. Her mother had had the same symptoms, but beginning much later in life. Examination showed camptocormia but no definite limb weakness. Her serum creatine kinase and acetylcholine receptor antibody levels were normal. EMG showed short duration, small amplitude motor units without abnormal spontaneous activity limited to the lumbar paraspinal muscles and sparing the limbs. No decremental response to low frequency repetitive nerve stimulation was present. Spinal MRI showed increased T1 and T2 signal suggestive of fatty changes in the lumbar paraspinal muscles. An aluminum walker helped her gait. There has been no obvious change in the degree of weakness over the past 2 years.

Biological basis

Etiology and pathogenesis

The causes of isolated neck extensor myopathy and primary camptocormia are not known. It has been postulated that, in susceptible individuals, a combination of postural changes and aging may place excessive loads on the paraspinal musculature, eventually leading to the focal myopathy (Katz et al 1996; Oishi et al 2000). For example, the gravitational forces pulling the head downward should increase as the head begins to fall forward. As the neck muscles are stretched they contract inefficiently and the head moves further into a flexed posture. This vicious cycle continues until the head rests firmly against the chest. The fact that patients with extrapyramidal disorders, with their related postural changes, are susceptible to paraspinal extensor myopathies supports this hypothesis (Askmark et al 2001). Others have pointed to evidence favoring a focal inflammatory myopathy (Hilliquin et al 1992; Biran et al 1999), focal mitochondrial disease (Baquis et al 1997), or a focal dystrophy (Laroche et al 1995; Serratrice et al 1996; Yoshimura et al 1996) as the etiology.

For both isolated axial extensor paraspinal myopathies, muscle histology of the paraspinal muscles reveals a nonspecific myopathic process including increased connective tissue, split fibers, and numerous fibers undergoing myonecrosis. Biopsies of limb muscles are normal (Katz et al 1996). No biochemical or genetic abnormality has been identified. Sakiyama and colleagues reported on a family with maternal inheritance of axial myopathy and encephalopathy associated with ragged-red fibers and cytochrome c oxidase-negative fibers in muscle biopsy and a novel heteroplasmic mutation (m.602C>T) in the tRNA(Phe) gene of the mitochondrial DNA (Sakiyama et al 2011). Paraspinal muscles appear to be particularly susceptible to age-related mitochondrial respiratory chain defects. Campbell and colleagues observed an age-related increased in cytochrome oxidase deficient (respiration-deficient) myofibers in paraspinal muscle in association clonal expansion of mtDNA deletions and depletion (Campbell et al 2013).

Epidemiology

The incidence and prevalence of these disorders are not known. Isolated neck extensor myopathy occurs more commonly than primary camptocormia. Both conditions tend to come to medical attention when the onset is rapid or the degree of involvement is particularly severe. However, abnormal posture is common in elderly people, and we suspect this is attributable to axial extensor muscle weakness in at least some individuals.
Prevention

Risk factors include any condition that affects the posture (aging, Parkinson disease), activities that cause the individual to remain upright for excessive periods (sleeping in a chair), or diseases that weaken the paraspinal muscles (neuromuscular conditions, radiculopathy). Why a severe degree of weakness occurs in some individuals but not others is unknown. There are no obvious strategies for prevention.

Differential diagnosis

Axial paraspinal extensor myopathies may occur in a wide range of myopathic, neuromuscular junction, motor neuron, and neuropathic disorders or in Parkinson disease. Table 1 gives an overview on the ever growing number of myopathies potentially manifesting or even presenting with camptocormia.

Table 1. Paraspinal Weakness in Myopathic, Neuromuscular Junction, Motor Neuron, and Neuropathic Disorders

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Reference</th>
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<tbody>
<tr>
<td>Amyotrophic lateral sclerosis</td>
<td>(Wolfe and Bank 1994; Van Gerpen 2001)</td>
</tr>
<tr>
<td>Calpainopathy (carrier)</td>
<td>(Liewluck and Goodman 2012)</td>
</tr>
<tr>
<td>Carnitine deficiency</td>
<td>(Karpati et al 1975)</td>
</tr>
<tr>
<td>Chronic inflammatory demyelinating polyneuropathy</td>
<td>(Hoffman and Gutmann 1994)</td>
</tr>
<tr>
<td>Congenital myopathy</td>
<td>(Riggs et al 1994)</td>
</tr>
<tr>
<td>Duchenne muscular dystrophy (carrier)</td>
<td>(Findlay et al 2013)</td>
</tr>
<tr>
<td>Dysferlinopathy (carrier)</td>
<td>(Gáti et al 2012)</td>
</tr>
<tr>
<td>Focal myositis</td>
<td>(Biran et al 1999)</td>
</tr>
<tr>
<td>FSHD</td>
<td>(Umamathi et al 2002; Jordan et al 2011; Dahlqvist et al 2014)</td>
</tr>
<tr>
<td>Hyperparathyroidism</td>
<td>(Beekman et al 2002)</td>
</tr>
<tr>
<td>Inclusion body myositis</td>
<td>(Goodman et al 2012; Ma et al 2013)</td>
</tr>
<tr>
<td>Licorice induced hypokalemia</td>
<td>(Yoshida and Takayama 2003)</td>
</tr>
<tr>
<td>Mc Ardle disease</td>
<td>(Witting et al 2014)</td>
</tr>
<tr>
<td>Mitochondrial myopathy</td>
<td>(Bacquis et al 1997; Sakiyama et al 2011)</td>
</tr>
<tr>
<td>Motor neuron diseases</td>
<td>(Gourie-Devi et al 2003)</td>
</tr>
<tr>
<td>Myasthenia gravis</td>
<td>(Kataoka et al 2012; Sawa et al 2013)</td>
</tr>
<tr>
<td>Myofibrillar myopathy</td>
<td>(Renard et al 2012)</td>
</tr>
<tr>
<td>Myositis</td>
<td>(Zenone et al 2013)</td>
</tr>
<tr>
<td>Myotonic dystrophy type 1</td>
<td>(Lawson et al 2013)</td>
</tr>
<tr>
<td>Nemaline myopathy</td>
<td>(Lomen-Hoerth et al 1999)</td>
</tr>
<tr>
<td>Polymyositis</td>
<td>(Keating et al 1996)</td>
</tr>
<tr>
<td>Proximal myotonic myopathy</td>
<td>(Evidente and Cook 1997; Serratrice et al 2000)</td>
</tr>
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</table>

Myasthenia. Myasthenia gravis may affect extensor neck muscles along with ocular, bulbar, or limb muscles and is one of the more common causes of dropped head syndrome (Katz et al 1996) or camptocormia (Kataoka et al 2012). Although the dropped head sign as the first or only manifestation of the disease is unusual, a series of 6 patients who presented with dropped head that responded to intravenous edrophonium were confirmed to have myasthenia gravis (Casasnovas et al 2007; Sawa et al 2013).

Myositis. A few cases of dropped head or bent spine syndrome have been attributed to focal myositis of the paraspinal musculature, as shown by MRI and muscle biopsy. Immunosuppressive treatment or intravenous immunoglobulins were effective in some of these cases (Kastrup et al 2008; Zenone et al 2013).

Muscle dystrophy. Dropped head syndrome may also be the dominant feature of a congenital muscular dystrophy, as reported in patients with mutations in the lamin A/C and selenoprotein N genes (D’Amico et al 2005).

Motor neuron disease. Motor neuron diseases, including amyotrophic lateral sclerosis, may also cause neck or trunk weakness with about 1% of cases presenting this way (Wolfe and Bank 1994; Van Gerpen 2001; Gourie-Devi et al 2003). Specific neuromuscular disorders can be recognized by subtle findings on examination, laboratory
abnormalities, or histological changes that are not characteristic of isolated neck extensor myopathy or primary camptocormia.

When paraspinal muscle weakness occurs as part of a generalized neuromuscular disorder, it is usually the result of the same underlying pathophysiology that causes weakness in other muscle groups. However, there may be exceptions. For example, Katz and colleagues described a patient with myasthenia gravis who continued to have persistent neck extensor weakness even after limb, ocular, and bulbar weakness responded to immune suppression therapy (Katz et al 1996). Electromyography showed myopathic changes limited to the neck extensors, and we speculated that the head drop was initially caused by the myasthenia, but ultimately the combination of weakness and postural changes resulted in a secondary myopathy of the cervical paraspinal muscles (Katz et al 1996). This observation was corroborated in another report (Rodolico et al 2004). Multilevel nerve root impingement is another gray area that has been postulated as a cause for severe lumbar paraspinal weakness (Penisson-Besnier et al 1994). However, others speculate that nerve root involvement alone cannot easily cause postural changes, and that paraspinal muscles affected by radiculopathy may ultimately become susceptible to myopathic abnormalities (Umapathi et al 2002). The same type of hypothesis may help explain the relatively common occurrence of isolated neck extensor myopathy and camptocormia in patients with extrapyramidal disorders (Okamiya et al 1997; Waclawik et al 1997; Djaldetti et al 1999; Askmark et al 2001; Lava and Factor 2001).

Structural abnormalities of the spine should also be considered in patients presenting with a bent spine (Ehrenstein et al 1996). These are easily excluded if the patient can fully extend the back while supine. Noteworthy, the dropped head syndrome may secondarily lead to cervical spine kyphosis and cervical myelopathy requiring surgery (Nakanishi et al 2007).

Drug-induced camptocormia has been reported with olanzapine (Robert et al 2010) and pramipexole (Nakayama and Miwa 2012).

**Diagnostic workup**

Serum creatine kinase levels are normal in the axial paraspinal extensor myopathies. Tests for acetylcholine receptor antibodies are important to exclude myasthenia gravis. Electromyography reveals myopathic changes limited to the affected paraspinal region. Magnetic resonance imaging rules out structural abnormalities of the spine and demonstrates abnormal signal in damaged paraspinal muscles (Laroche et al 1995; Katz et al 1996). Muscle biopsy of paraspinal muscles may show abnormalities specific to the many possible neuromuscular etiologies listed above. Histology has to be interpreted with caution, however, because the paraspinal muscles show a variety of neuromuscular abnormalities, eg, increased fiber size variability, endomysial fibrosis, adipose tissue, and mitochondrial changes, even in healthy subjects (Zimmermann et al 2014).

**Management**

There are no established effective therapies for patients with primary camptocormia and isolated neck extensor myopathy. Cervical collars or neck braces, along with physical therapy, may be the most practical supportive treatment options. However, it is difficult to find a suitable collar that can support the neck and yet be comfortable. Patients find that soft or hard cervical collars rub against the chin, interfering with speech and rotation of the neck. The best approach Katz and colleagues found is to support the head with a brace, which is connected to a firm padded support that rests against the chest wall (Katz et al 1996). It has an open, lightweight design that provides some flexibility and reduces heat retention. Alternatively, patients have used a custom brace that supports the head from behind, using a rigid backboard attached to a harness worn around the trunk. Both collars require referral to an orthotic specialist. Patients with camptocormia benefit from the use of a walker. This compensates for the trunk weakness by allowing the patient to use the arms for support. With either condition, we usually suggest that patients try to minimize the time spent standing or sitting with the head or trunk unsupported because there is anecdotal evidence that strict bed rest may lead to improvement.

Schroeteler and colleagues used a high-frame walker (HFW) with forearm support in 3 patients with parkinsonian camptocormia (Schroeteler et al 2011). Posture improved in all patients while standing and walking with the HFW. Walking distance in the upright position increased, and the occurrence of back pain was reduced. These authors observed sustained therapeutic effects with a HFW in a total of 20 patients and suggest a controlled study over a longer period.
Dystonic forms of camptocormia may respond to sensory tricks. A Parkinson patient with camptocormia, for example, had consistent and sustained benefit by wearing a backpack (Gerton et al. 2010). In idiopathic and parkinsonian camptocormia, several publications advocate the use of deep brain stimulation (DBS). Twenty-six patients (in 9 reports) undergoing DBS have been reported. The targets of stimulation were the subthalamic nucleus (STN, 24 cases) and the globus pallidus internus (GPI, 2 cases). Sixteen patients (66.7%) with subthalamic nucleus stimulation and both patients with GPI stimulation exhibited improvement of camptocormia (Sakas et al. 2010; Umemura et al. 2010; Asahi et al. 2011). Marked degeneration of the paraspinal muscles may predict negative outcome (Asahi et al. 2011). The pooled results of these single cases and small case series warrant a randomized clinical trial of DBS in parkinsonian camptocormia.

Corticosteroids generally have no therapeutic role if there is strong diagnostic evidence of paraspinal axial myopathy (Hilliquin et al. 1992; Katz et al. 1996). Note that responsiveness to corticosteroids has been described in cases that clinically correspond to isolated neck extensor myopathy, but these had histological evidence of paraspinal inflammation or high serum creatine kinase levels (Biran et al. 1999; Rose et al. 1999). Empirical therapy with corticosteroids or pyridostigmine may be tried if there is sufficient concern that myasthenia gravis is present. One patient with isolated neck weakness was suspected of having a mitochondrial myopathy with ragged red fibers and responded to a combination of coenzyme Q and vitamins (Baquis et al. 1997).

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**References especially recommended by the author or editor for general reading.

Former authors

Jonathan Katz MD and Richard J Barohn MD (original authors)

ICD and OMIM codes

ICD codes

ICD-9:
Isolated neck extensor myopathy: 723.9
Camptocormia: 300.11

ICD-10:
Dorsopathy, unspecified: M53.9
Camptocormia: F44.8

Profile

Age range of presentation

0-01 month
01-23 months
02-05 years
06-12 years
13-18 years
19-44 years
45-64 years
65+ years

**Sex preponderance**

Primary camptocormia
female>male, 3:1
female>male, 2:1
female>male, 1:1
Isolated neck extensor myopathy
male=female

**Family history**

Isolated neck extensor myopathy
No family history for isolated
Primary camptocormia
Family history may be obtained

**Heredity**

Isolated neck extensor myopathy
none
Primary camptocormia
heredity may be factor
autosomal dominant

**Population groups selectively affected**

None selectively affected

**Occupation groups selectively affected**

None selectively affected

**Differential diagnosis list**

amyotrophic lateral sclerosis
calpainopathy (carrier)
carnitine deficiency
chronic inflammatory demyelinating polyneuropathy
congenital myopathy
Duchenne muscular dystrophy (carrier)
dysferlinopathy (carrier)
focal myositis
FSHD
hyperparathyroidism
inclusion body myopathy
inclusion body myositis
licorice induced hypokalemia
mitochondrial myopathy
motor neuron diseases
myasthenia gravis
myofibrillar myopathy
myositis
nemaline myopathy
polymyositis
proximal myotonic myopathy
muscle dystrophy
structural abnormalities of the spine
drug use (olanzapine and pramipexole)

**Associated disorders**

Dropped head syndrome

**Other topics to consider**

Minicore myopathy
Myopathies associated with parathyroid disorders
Myopathies associated with thyroid disease
Neuromuscular pathology: overview
Stiff-person syndrome

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