Isolated camptocormia revealing sporadic late onset nemaline myopathy

Camptocormie isolée révélant une myopathie à bâtonnets tardive

Camptocormia is an abnormal posture with marked flexion of thoracolumbar spine that abates in the recumbent position. Causes of camptocormia include neurological disorders, psychogenic origin and idiopathic camptocormia [1–3]. Sporadic late onset nemaline myopathy (SLONM) is a rare disorder that typically presents subacutely with evolving weakness of the limb-girdle muscles [4]. We describe a patient with isolated camptocormia due to SLONM.

Case report

A 56-year-old woman presented with a 5-month history of progressive camptocormia and right lumbocurvature. There was no family history of neuromuscular disease. She had a history of paranoid schizophrenia that was well controlled with risperidone (8 mg/day). She had chronic low back pain which had evolved over the preceding 10 years. Examination showed significant flexion of the thoracolumbar spine which was most prominent on standing and walking, improved on sitting and was relieved when supine. The patient had scoliosis with right convexity. Muscle strength was normal in the neck, cranial, proximal and distal limb muscles. Sensory examination and tendon reflexes were normal. There was a discrete akesia and symmetrical hypotonia of all four limbs related to neuroleptic drugs. Creatine kinase was 143 IU/L (normal range: 40–225 IU/L). C-reactive protein, erythrocyte sedimentation rate, blood cell counts, thyroid-stimulating hormone level and serum immunoelectrophoresis were normal. Antinuclear antibodies, anti-acetylcholine receptor antibodies, HIV and HTLV serologies were negative. Spine X-ray found a lumbar scoliosis focused on L3 with right convexity, associated with adjacent discopathy. Lumbar spine MRI revealed a significant, asymmetrical atrophy of paraspinal muscles, and a disc protrusion at the L2-L3 level (Figure 1). A thoracoabdominal CT scanner showed hypotrophy of the right spine erector muscles from the T9 to L3 vertebrae, with no atrophy or fatty degeneration in the limbs and abdominal muscles. Electromyography did not show a myopathic or neurogenic pattern, although the patient’s cooperation was limited. Cardiopulmonary investigations were normal. A deltoid muscle biopsy showed nemaline bodies in type 2 fibers which were most apparent using Gomori trichrome staining, as well as lobulated fibers and angulated atrophic fibers that were essentially type 2 fibers and showed random distribution. The percentage of fibers with nemaline bodies was estimated to be 7% (Figure 2). There were no necrotic fibers nor inflammatory infiltrates. Lumbocurvature resolved after two peridural infiltrations. The patient then attended a rehabilitation program which improved the camptocormia. After 3 years’ follow-up she remains stable.

Discussion

Nemaline myopathies (NM) are a genetically diverse group of congenital myopathies, generally of neonatal or childhood onset. However, a study of 143 cases reported six patients with adult-onset NM, presenting at 41 to 59 years of age. The alpha-actin gene (ACTA1) mutations are a common cause of NM [5]. We did not perform a genetic testing for our patient to rule out a genetic origin. Nevertheless, the vast majority of SLONM cases are of acquired origin. To our knowledge, this is the first report of a case in which an isolated camptocormia occurred due to SLONM. An another case of isolated camptocormia in a 77-year-old male due to SLONM was reported but the patient had also a Parkinson’s disease [6]. In one-third of patients, camptocormia occurs secondary to extrapyramidal disorders (Parkinson’s disease and neuroleptic-induced dystonia) or to various neuromuscular disorders (amyotrophic lateral sclerosis, myasthenia gravis, limb-girdle muscular dystrophies, and inclusion body myositis) [1–3]. Two-thirds of patients with camptocormia have delayed-onset paraspinal myopathy. In this idiopathic camptocormia, all patients are older than 60 years at disease onset, and CT and histological findings are nonspecific but remarkably uniform. CT scans show symmetrical atrophy and heterogeneous fatty infiltration of the paraspinal muscles extending along the entire length of the spine [1]. Biopsies of the paraspinal muscles show endomysial fibrosis and fatty infiltration, with no evidence to suggest the presence of another disease. The findings from electromyography of paraspinal muscles are not uniform, exhibiting both myogenic and neurogenic features.

In our patient, camptocormia did not arise secondary to neuroleptic drugs. Biopsy of the deltoid muscle, which was clinically...
unaffected, was the key investigation leading to the diagnosis of NM. SLONM is a rare disorder which may be idiopathic. But at least 50% of SLONM cases are associated with monoclonal gammopathy, and some of them occur in early stage of HIV infection [4,7]. Associated monoclonal gammopathy carries an unfavorable prognosis [4]. The idiopathic form of SLONM typically presents after age 40 and evolves subacutely [4]. Limb-girdle weakness and atrophy are the main symptoms. Distal weakness, head drop, respiratory insufficiency and dysphagia can also occur in association with proximal muscles weakness. Creatinine kinase is normal and EMG findings show features of myopathy [4]. Paraspinal muscle involvement is frequent in SLONM, and dropped head occurs in nearly half of all cases [8]. One case of SLONM has been reported in a 72-year-old man with a 10-year history of progressive isolated head drop [9].

The diagnosis of SLONM is based on muscle biopsy findings of nemaline bodies, or rods, easily identified by Gomori trichrome staining in atrophic or non-atrophic fibers. The frequency of rod-bearing fibers varies between patients, and between different biopsies in the same patient. Associated pathologic features may be necrotic fibers and lobulated fibers in rare cases [4]. Ultrastructurally, the electron-dense nemaline bodies originate from the Z-disc of sarcomeres. However, nemaline bodies are not a specific finding and can be observed as a minor feature in muscle biopsies obtained from patients with a variety of neuromuscular disorders [4]. NM is therefore diagnosed only if nemaline bodies are the unique or predominant abnormality and there are no features to suggest an associated myopathy. Two reported cases of patients with SLONM associated with monoclonal gammopathy demonstrated an impressive clinical response induced with aggressive immunotherapy [10–12]. However, immunotherapy is of uncertain benefit in cases of SLONM that are not associated with monoclonal gammopathy [4]. Physical therapy may be useful.

Faced with an isolated camptocormia, muscle biopsy is therefore an important investigative tool that may lead to an unusual diagnosis such as SLONM.

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References


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