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Evaluation of the Lumbar Spine in Patients with Glycogen Storage Disease: CT Demonstration of Patterns of Paraspinal Muscle Atrophy

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CT studies of the lumbar spine were performed in 19 patients with glycogen storage disease. Nine of 10 patients with McArdle's disease and seven of nine patients with acid maltase deficiency demonstrated posterior paraspinal muscle atrophy out of proportion to their ages. In addition, the psoas muscles were spared in all 10 patients with McArdle's disease and were involved with atrophy in seven of the nine patients with acid maltase deficiency.

We conclude that when patients with low back pain—or asymptomatic patients—demonstrate otherwise unexplained atrophy of the paraspinal muscles the diagnosis of glycogen storage disease should be considered. Furthermore, when the psoas muscles are spared, the specific diagnosis of McArdle's disease is suggested.

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Low back pain is among the most common symptoms encountered by the neuroradiologist. When imaging is performed, lesions such as disk herniations, degenerative osteoarthritis, or spinal stenosis are often demonstrated. Not infrequently, however, no lesions are found to account for the patient's symptoms. Rarely, atrophy of the paraspinal muscles out of proportion to the patient's age is seen. Having encountered this phenomenon several times in patients with glycogen storage diseases who had no other reason for low back pain, we began to study a group of these patients who were being followed at our institution.

Glycogen storage diseases are disorders of carbohydrate metabolism characterized by various enzymatic defects resulting in faulty glycogenolysis [1–9]. In certain disorders, adolescent and even adult patients may present with low back pain or proximal muscle weakness. We used CT to study the lumbar spines of nine patients with glycogen storage disease type II, also known as acid maltase deficiency, and 10 patients with type V, also known as McArdle's disease. In 14 of the 19 patients the studies were done to evaluate either low back pain or proximal muscle weakness; the other five patients had neither muscle weakness nor symptoms referable to the lumbar spine.

Subjects and Methods

Noncontrast CT scans of the lumbar spine were obtained in 10 patients with proved McArdle's disease and in nine other patients with proved acid maltase deficiency. High-resolution scanning was performed on fourth-generation CT scanners (Picker 1200, Cleveland, OH). In 15 of the 19 patients, contiguous 5-mm axial sections were obtained from the L1 to the L3 level, followed by 3-mm axial sections through the L3–L4, L4–L5, and L5–S1 disk spaces, angled to the disks. In three patients, contiguous 5-mm sections were obtained from the L1 through the S1 level. In one of the patients, 3-mm sections were obtained through the disk spaces from L1–L2 through L5–S1, angled to the disks. Images were obtained at soft-tissue windows to evaluate the paraspinal musculature as well as at bone windows to evaluate the osseous structures. In addition, one of the patients with McArdle's disease had

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MR imaging of the lumbar spine during the course of his evaluation. Sagittal images were obtained using spin-echo sequences of 500/35 and 2500/38–76; transaxial images were acquired from the L3 level to the S1 level using a spin-echo pulse sequence of 500/35.

Of the 10 patients with McArdle's disease, four were symptomatic with low back pain; one had severe low back pain and proximal muscle weakness; one was symptomatic with pain across the shoulders but not in the lower back; two were symptomatic with proximal muscle weakness but no symptoms referable to the lumbar spine; two were totally asymptomatic. All 10 patients were able to ambulate without assistance. The ages of these patients ranged from 19 to 65 years (mean, 37 years). Seven of the patients were men; three were women (Table 1).

Four of the patients with acid maltase deficiency had proximal muscle weakness; one had an exaggerated lumbar lordosis; one had low back pain; another had both severe low back pain and marked proximal muscle weakness; two others were asymptomatic. Eight of these nine patients were able to ambulate without assistance; one patient required a cane to walk. Seven of the patients were males and two were females. Their ages ranged from 14 to 52 years (mean, 23 years) (Table 2).

Atrophy of the posterior paraspinal and psoas musculature was evaluated according to guidelines described by Hadar et al. [10], as follows. Grade 0 = homogeneous muscle density with occasional low-density fine striations of fat outlining muscle bundles; grade 1 = slight decrease in muscle mass with coarse, fat striae; grade 2 = marked decrease in muscle mass with a prominent amount of fat; grade 3 = total replacement of muscle by fat (Fig. 1). The muscles were graded according to their inherent radiologic appearance, without regard to age-related changes.

TABLE 1: Findings in 10 Patients with McArdle's Disease (Type V Glycogen Storage Disease)

Case No.	Age (yr)	Sex	Symptoms	Posterior Paraspinal Atrophy	Psoas Atrophy	Other Findings
1	33	M	Asymptomatic	1	0	Disk bulges L3–L4, L4–L5
2	65	M	Severe PMW, LBP	3	0	Disk bulges L3–L4, L4–L5
3	48	M	Severe LBP	2	0	None
4	20	F	Interscapular pain	1	0	Disk bulge L5–S1
5	56	M	LBP	2	0	Disk bulge L4–L5
6	43	F	PMW	1	0	Disk bulges L1–S1
7	36	M	PMW	1	0	Posterior fusion L3–S1
8	27	F	LBP	1	0	Disk bulge L5–S1
9	19	M	Asymptomatic	0	0	Disk bulge L5–S1
10	25	M	LBP	1	0	Small HNP L5–S1

Note.—PMW = proximal muscle weakness. LBP = low back pain, HNP = herniated nucleus pulposus.

TABLE 2: Findings in Nine Patients with Acid Maltase Deficiency (Type II Glycogen Storage Disease)

Case No.	Age (yr)	Sex	Symptoms	Posterior Paraspinal Atrophy	Psoas Atrophy	Other Findings
1	17	F	LBP	2	2	None
2	52	M	Severe LBP, PMW	3	1	Disk bulge L5–S1
3	14	M	Severe PMW	1	2	None
4	31	M	PMW	2	3	None
5	14	M	Asymptomatic	0	0	None
6	14	M	Lordosis	1	1	None
7	16	F	Asymptomatic	0	0	None
8	20	M	PMW	1	2	Disk bulge L5–S1
9	31	M	PMW	3	3	None

Note.—LBP = low back pain, PMW = proximal muscle weakness.

Results

Lumbar spine examinations in the patients with McArdle's disease revealed that three of the five who suffered from low back pain had moderate to severe atrophy of the posterior paraspinal muscles (erector spinae and multifidus). The remaining two patients with low back pain as well as four of the other five patients who were less symptomatic (including one who was asymptomatic) demonstrated mild fatty infiltration, but even this was consistently out of proportion to the patients' ages. One of the patients who was totally asymptomatic had no atrophy at all. Of note is the fact that the psoas muscles were completely spared in all 10 patients with McArdle's disease (Figs. 2 and 3). Seven of these patients were additionally noted to have annular disk bulges at various lumbar levels but no other positive findings; one demonstrated a small central disk herniation at L5–S1; one patient had previously undergone posterior fusion of L3 through S1; the scan of the 10th patient was unremarkable except for the muscle atrophy.

Of the patients with acid maltase deficiency, seven demonstrated prominent fatty infiltration and atrophy of both the posterior paraspinal muscle group and the psoas muscles (Fig. 4). The atrophy ranged from mild to severe, but again was consistently noted to be abnormal for the patients' ages. Five of these seven patients had no other demonstrable abnormalities on their scans, while two were noted to have a disk bulge at L5–S1. In two patients there was no convincing evidence of muscle atrophy. These patients were both asymptomatic.

Discussion

The erector spinae and multifidus muscle groups comprise the deep paraspinal muscles in the lumbar spine, posterior to the lumbar transverse processes [11]. Atrophy of these muscles has been noted in conjunction with advanced age, neuromuscular disorders, and disuse [10, 12, 13], as well as with ankylosing spondylitis and spinal osteoid osteoma, likely as a result of the disuse that occurs in these conditions [14–16]. In addition to this posterior paraspinal muscle atrophy, psoas muscle atrophy was noted in three of the 14 patients with ankylosing spondylitis studied by Gordon et al. [15] and in the patient with spinal osteoid osteoma studied by McConnell and Daneman [16]. This also was thought to be related to disuse.

In our study, we describe posterior paraspinal muscle atrophy in seven of nine patients with acid maltase deficiency and in nine of 10 patients with McArdle's disease. Additionally, all seven of the patients with acid maltase deficiency who demonstrated posterior paraspinal muscle atrophy also had psoas atrophy. However, psoas involvement was not noted in any of the patients with McArdle's disease.

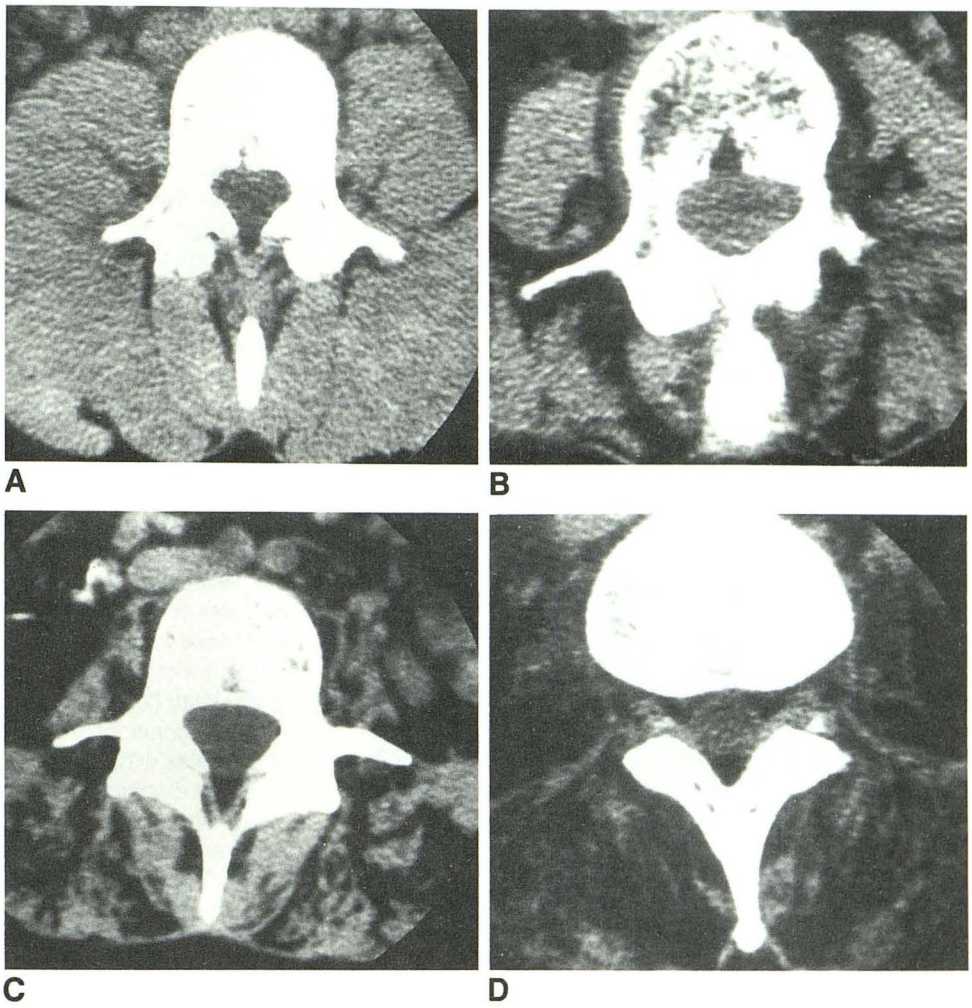

Fig. 1.—Examples of different grades of posterior paraspinal and psoas muscle atrophy as seen on CT scans.

***A*, Grade 0: Homogeneous muscle density with occasional low-density fine striations of fat outlining muscle bundles.**

***B*, Grade 1: Slight decrease in muscle mass with coarse fat striae.**

***C*, Grade 2: Marked decrease in muscle mass with a prominent amount of fat.**

***D*, Grade 3: Total fatty replacement of the muscles.**

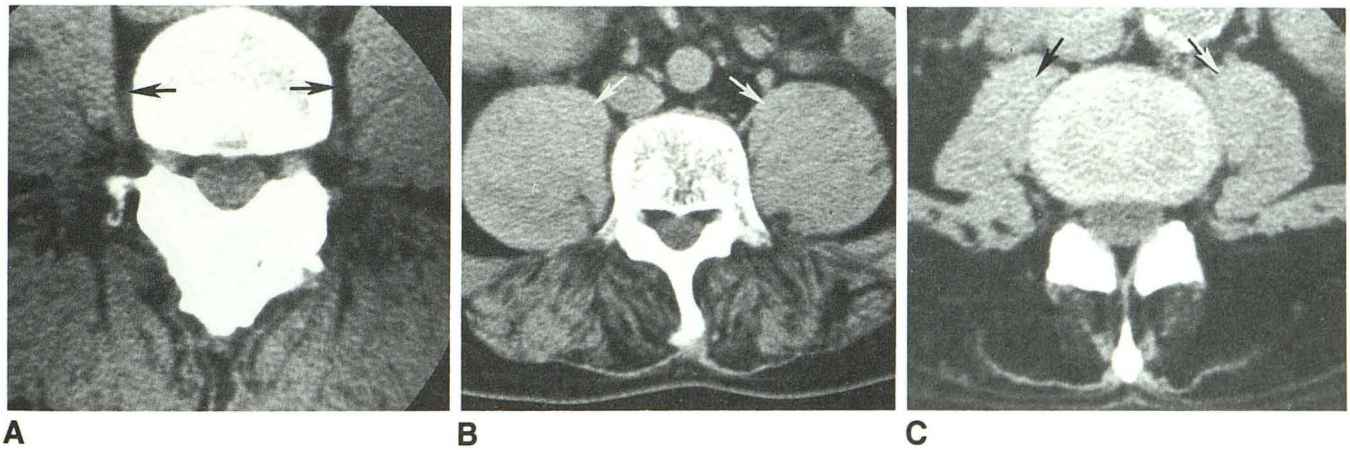

Fig. 2.—CT scans in patients with McArdle's disease. Note sparing of psoas muscles in all three patients (*arrows*) associated with various grades of posterior paraspinal muscle atrophy.

***A*, 36-year-old man with posterior paraspinal muscle atrophy grade 1.**

***B*, 56-year-old man with posterior paraspinal muscle atrophy grade 2.**

***C*, 65-year-old man with posterior paraspinal muscle atrophy grade 3.**

Five of the 10 patients with McArdle's disease had no symptomatology referable to the lumbar spine. Nonetheless, four of the five had positive CT scans. Among the patients with acid maltase deficiency, the only ones to have normal scans were the two who were asymptomatic.

Type II glycogen storage disease, acid maltase deficiency, is caused by a defect of α-glucosidase activity and leads to progressive accumulation of glycogen, mainly in the lysosomes [1–7]. Three forms of this disease are currently identified: an infantile form (classical Pompe disease), an early childhood form, and an adult form. In the infantile form, the usual presenting symptoms are hypotonia, muscle weakness, and congestive heart failure. These symptoms usually appear between birth and 6 months of age. Most patients die by 1 year of age from cardiorespiratory failure, pneumonia, or aspiration. These symptoms are due to the abnormal accumulation of glycogen in all tissues, but particularly in the heart, skeletal muscle, Schwann cells of the peripheral nerves, and within the CNS in the anterior horn cells and neurons of the brainstem nuclei [2]. In the childhood and adult forms, the disease is essentially limited to the musculoskeletal system and patients develop proximal muscle and often respiratory muscle weakness simulating muscular dystrophy or polymyositis. Cardiac abnormalities are either absent or minimal in these patients [1, 2, 4, 6, 7], and the abnormal accumulation of glycogen occurs almost exclusively in muscle fibers.

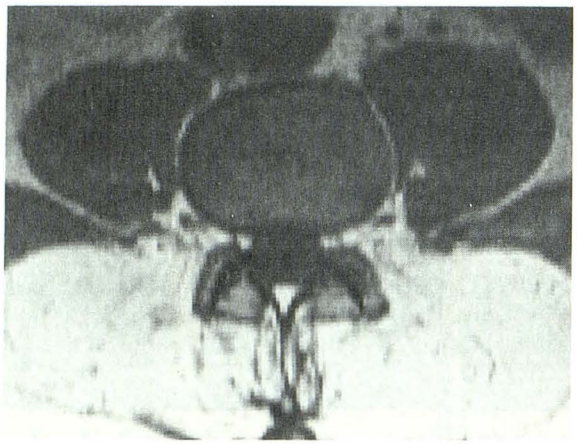

Fig. 3.—Transaxial T1-weighted MR image (500/35) through lumbar spine of patient with McArdle's disease at L3–L4 level shows grade 3 atrophy of posterior paraspinal muscles with sparing of psoas muscles.

In type V glycogen storage disease, McArdle's disease, there is a deficiency of muscle phosphorylase, an enzyme necessary to generate adenosine triphosphate during exercise [1, 2]. The disease usually manifests itself in childhood or adolescence, with increased fatigability, and progresses over the next 20 to 40 years to exercise intolerance, muscle weakness and cramping, and myoglobinuria [1, 2, 7–9]. However, in some patients the only symptom may be progressive muscle weakness, which develops late in life (sixth or seventh decade) with no history of muscle cramps or myoglobinuria [7]. Histochemical evaluation of muscle biopsy specimens in all patients with McArdle's disease reveals no muscle phosphorylase activity. In some specimens this enzyme is not present while in other patients an inactive form of this enzyme is detected [2].

While some of the glycogen storage diseases manifest themselves within the first few months of life, others may not manifest themselves until adolescence or even adulthood, at which time patients will present with nonspecific symptoms, such as back pain or muscle weakness. McArdle's disease (type V) and two of the three forms of acid maltase deficiency (type II) are examples. As such, it would be prudent to consider these diagnoses in patients with either low back pain or proximal muscle weakness who demonstrate posterior paraspinal muscle atrophy and no other findings on CT scans of the lumbar spine. Psoas muscle atrophy, in the absence of any cause for disuse, should raise the suspicion of acid maltase deficiency. Furthermore, as two of our patients

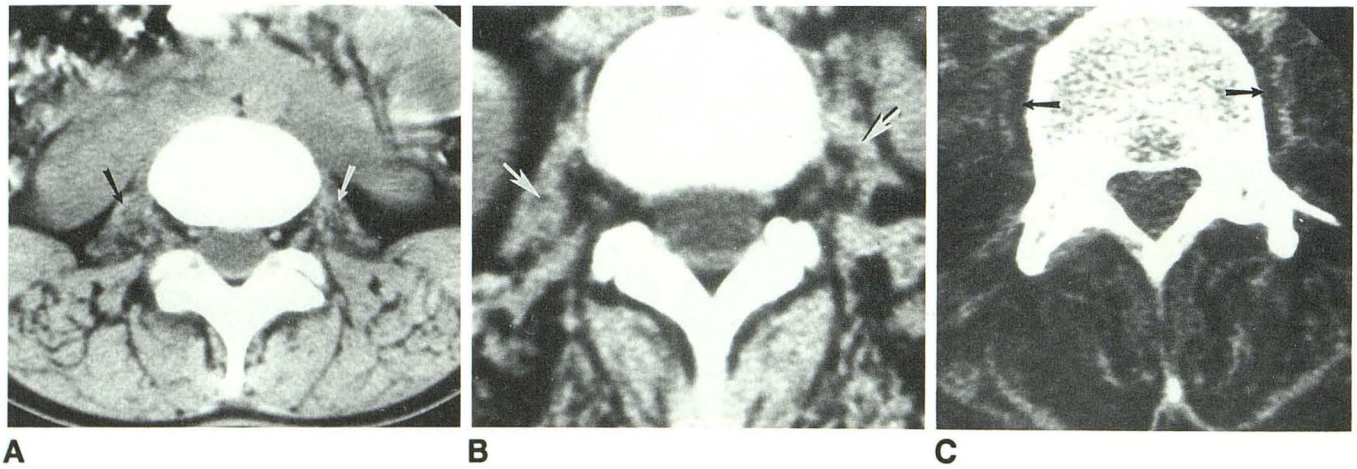

Fig. 4.—CT scans in patients with acid maltase deficiency.
***A*, 14-year-old boy with posterior paraspinal muscle atrophy grade 1 and psoas muscle atrophy grade 2 (*arrows*).**
***B*, 17-year-old girl with posterior paraspinal and psoas muscle atrophy grade 2 (*arrows*).**
***C*, 31-year-old man with posterior paraspinal and psoas muscle atrophy grade 3 (*arrows*).**

with positive CT findings had no symptoms referable to the lower back, it is suggested that the CT findings may precede the development of symptoms.

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