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Magnetic resonance imaging of children with Duchenne muscular dystrophy

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Abstract. Eight children representing a spectrum of clinical states of biopsy-proven Duchenne muscular dystrophy (DMD) underwent magnetic resonance (MR) scans to assess the degree of muscular involvement and disease progression. Five muscle groups (neck, shoulder girdle, pelvic girdle, thigh and calf) were evaluated. In each case, involved muscles were clearly demarcated. Image estimates of disease severity by degree of muscle involvement correlated well with clinical staging. In our experience MR is useful for assessment of disease stage, selection of appropriate muscles for biopsy and planning for courses of physical and rehabilitation therapy.

The muscular dystrophies, a heterogenous group of genetic diseases characterized by progressive muscular weakness, are a relatively common childhood disorder with significant morbidity and mortality. Duchenne type muscular dystrophy (DMD), the most severe, and the most frequently occurring of the muscular dystrophies, has an estimated incidence ranging from one in 1700 to one in 7700 live-born male births [4, 10]. The gene is transmitted with X-linked recessive inheritance.

Motor development is characteristically delayed, with progressive weakness initially involving the pelvic girdle and neck flexors, followed by weakness in the shoulder girdle and proximal limb muscles. Classic clinical features include pseudohypertrophy of the calves, a lumbar lordosis, Achilles tendon contractures, and a characteristic waddling gait. The disease is generally fatal between years 12 and 25, frequently from pulmonary complications owing to involvement of the respiratory muscles. Affected children may demonstrate elevations of serum creatine phosphokinase (CPK) from birth. Muscle biopsy demonstrates histologic alteration including muscular atrophy, patchy areas of necrosis and regeneration of muscle fibers.

There are few precise, non-invasive, objective methods to evaluate the progression of DMD. Definitive diagnosis and a baseline to evaluate disease progression are currently established on initial muscle biopsy.

Consideration of the expense and morbidity of this procedure limits the usefulness of repeated biopsies as a means of following disease progression.

Clinical parameters to assess progression are not precise, and measure only gross decremental changes in muscle function.

Methods of radiographic evaluation of the muscular dystrophies have included plain films, radionuclide scans [1], ultrasound [3, 6] and computed tomography [2, 5, 9]. All of these, with the possible exception of computed tomography, are relatively imprecise for the evaluation of disease progression. We report the use of magnetic resonance imaging (MRI) in the assessment of myopathy in children with Duchenne muscular dystrophy.

Materials and methods

Eight children with biopsy-proven DMD underwent MR scans to assess disease in at least three of five separate muscle groups: the neck shoulder girdle, pelvic girdle, thigh and calf. Ages ranged from 2.5 to 11.5 years. Clinical states encompassed most of the clinical spectrum, ranging from stage II to stage VIII. Patients were further divided into two clinical groups: those less severly affected (clinical stages I–IV) and children with more severe disease (clinical stages V–VIII). There were four children in each group.

All studies were performed on a Picker International 0.5 tesla superconducting magnet. Several pulse sequences were employed in the early phases of the study. The parameters that we typically used, TE = 40 ms and TR = 550, were the minimum our system allowed during a 4-slice acquisition.

In all patients transverse scans were obtained in 3–5 muscle groups, as noted above. Coronal scans of the pelvis and thigh were additionally obtained in three of the subjects better able to tolerate the longer acquisition times.

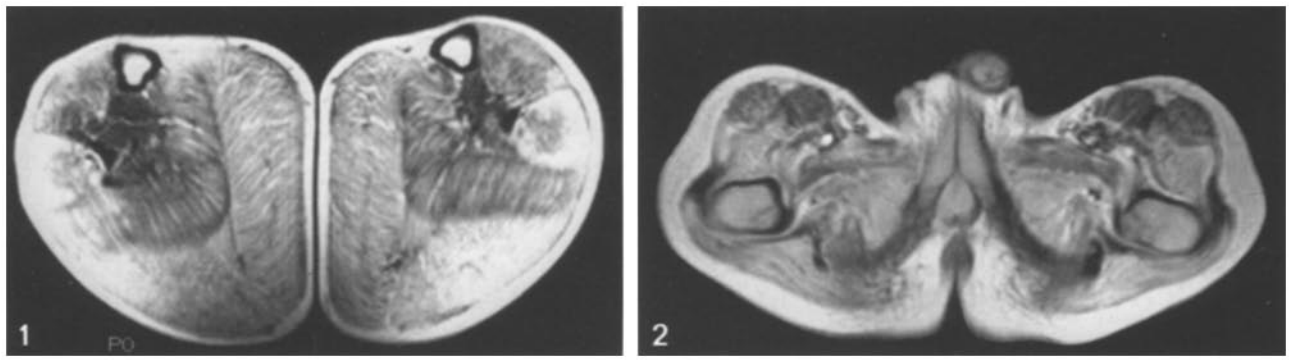


Fig. 1. Transverse section through the mid-calf of a 10.5-year-old boy with Duchenne muscular dystrophy. Clinical stage 6. Note massive „pseudohypertrophy" of both the soleus and gastrocnemius muscles

Fig. 2. Axial section through the pelvic floor of a 12-year-old with DMD, clinical stage 5. Involved muscles show increased signal intensity. There is characteristic sparing of the rectus femoris and sartorius muscles

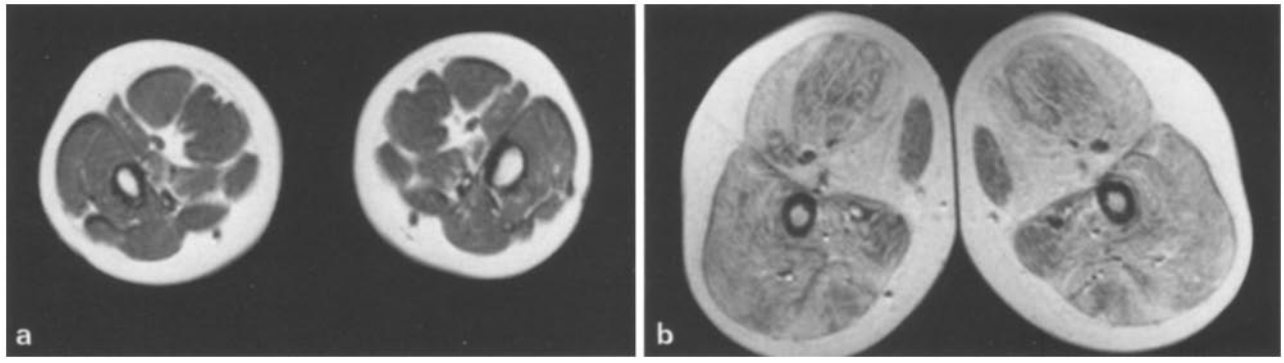


Fig. 3a and 3b. Comparison sections through the thigh in two patients with DMD. **a** was clinical stage 1-2. **b** was clinical stage 5 at time of MR image acquisition. Note the selective muscle pathology and lesser degree of fatty infiltration in the earlier stage

Results

The musculoskeletal system is well imaged by magnetic resonance, with clear delineation between cutis, sub-cutis, muscle and bone. In both the early and late phase children with DMD, those muscles replaced by fat were clearly demarcated on the MRI scans. Involvement was characterized by a non-homogenous increased intensity signal in affected muscles as compared to the normal. Image estimates of disease based on the degree of fat replacement of muscle correlated very well with the clinical stage of the disease.

In general T1 weighted spin echo images with a short TE and a short TR were found to be the most satisfactory. Although images taken with TR up to 900 ms demonstrated all the abnormal areas in those patients, shorter TR images appeared to improve the discrimination between involved and uninvolved muscles. Longer TR sequences subjectively appeared not to offer any advantage, and prolonged the scanning time.

On MR scans, DMD patients have a characteristic pattern of muscular involvement that correlates well with pathologic descriptions. In the eight patients studied, four individual muscles (the gracilis, sartorius, rectus femoris, and semi-tendinosus) were found to be free of abnormality. This was most evident in scans of the pelvis, thigh, and calf (Fig. 1-3). The findings were most obvious in those four patients with the more severe clinical disease, but were suggested in those minimal clinical findings as well.

Evaluation of the calf musculature revealed generalized involvement of all muscle groups, with relatively greater fatty infiltration in the posterior compartment as compared to the anterior muscle group (Fig. 1).

Pseudohypertrophy is attributed to the greatly increased volume of the soleus muscle from fatty deposit within muscle fibers. Less pronounced changes are seen in the gastronemius muscle. All muscles demonstrated a variable degree of muscular atrophy in addition to infiltration.

Discussion

In the past, several radiographic techniques have been used to characterize the muscular dystrophies. Plain films of the lower extremities of children with Duchenne muscular dystrophy reveal characteristic widening of the fibular head (Kaufman's fibular sign) and a small fibula relative to the tibia. Appearance of this sign occurs late in the course of the disease, and serial radiographs are not of use in evaluating severity or progression.

Radionuclide (gallium-citrate) scans [1] have shown a generally increased uptake in affected muscle of children with muscular dystrophy, and increased uptake diffusely in carriers of the genes. These studies suffer from a lack of specificity, and the relative amount of increased uptake cannot directly be correlated with the clinical severity of the disease.

Several investigations using ultrasonography [3, 6] to assess the muscular dystrophies have demonstrated increased fatty infiltration in affected muscles, but quantitation, necessary for accurate asessment of the disease progression has been difficult.

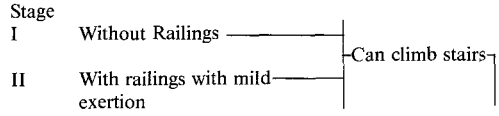
Radda et al. [11] have done biochemical MR studies of muscle in several disease states. It is difficult to compare their spectroscopic results using ATP peaks to our data using proton imaging.

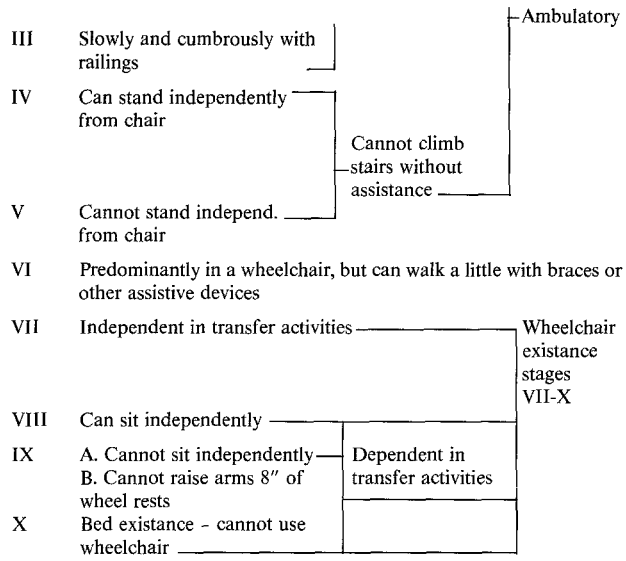
To date, the most precise radiographic imaging method for the evaluation of the disease course in the muscular dystrophies has been computed tomography (CT) [2, 5, 7, 9]. As previously noted, those muscle groups involved in the disease process are clearly demarcated on CT scans and recent investigations have attempted to quantify the amount of fatty infiltration in various muscle groups, allowing objective analysis of the rate and severity of disease progression in serial scans.

Our results show that MRI of the extremities of children with DMD affords excellent definition of the involved muscle groups owing in part to the ability to highlight the fat/muscle delineation. Sagittal and coronal images can be obtained and are helpful in determining the extent of disease.

The degree of involvement as estimated from the amount of fatty infiltation correlates well with the clinical staging. We believe that MRI is of potential value for the diagnosis, staging, and serial evaluation of patients with DMD.

Appendix. Clinical staging of Duchenne's muscular dystrophy





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