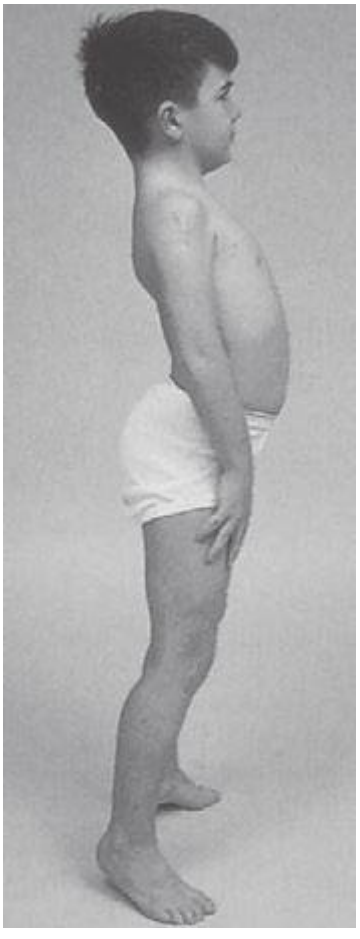

**FIGURE 462e-5** **Lordotic posture,** exaggerated by standing on toes, associated with trunk and hip weakness.

and anoctaminopathies (LGMD2L), there is a predilection for early atrophy of the gastrocnemius muscles, particularly the medial aspect. Atrophy of the humeral muscles is characteristic of FSHD.

## LABORATORY EVALUATION

A limited battery of tests can be used to evaluate a suspected myopathy. Nearly all patients require serum enzyme level measurements and electrodiagnostic studies as screening tools to differentiate muscle disorders from other motor unit diseases. The other tests described—DNA studies, the forearm exercise test, and muscle biopsy—are used to diagnose specific types of myopathies.

**Serum Enzymes** CK is the preferred muscle enzyme to measure in the evaluation of myopathies. Damage to muscle causes the CK to leak from the muscle fiber to the serum. The MM isoenzyme predominates in skeletal muscle, whereas creatine kinase-myocardial bound (CK-MB) is the marker for cardiac muscle. Serum CK can be elevated in normal individuals without provocation, presumably on a genetic basis or after strenuous activity, minor trauma (including the EMG needle), a prolonged muscle cramp, or a generalized seizure. Aspartate aminotransferase (AST), alanine aminotransferase (ALT), aldolase, and lactic dehydrogenase (LDH) are enzymes sharing an origin in both muscle and liver. Problems arise when the levels of these enzymes are found to be elevated in a routine screening battery, leading to the erroneous assumption that liver disease is present when in fact muscle could be the cause. An elevated γ-glutamyl transferase (GGT) helps to establish a liver origin because this enzyme is not found in muscle.

**Electrodiagnostic Studies** EMG, repetitive nerve stimulation, and nerve conduction studies (Chap. 442e) are essential methods for evaluation of the patient with suspected muscle disease. In combination, they provide the information necessary to differentiate myopathies from neuropathies and neuromuscular junction diseases. Routine nerve conduction studies are typically normal in myopathies but reduced amplitudes of compound muscle action potentials may be seen in atrophied muscles. The needle EMG may reveal irritability on needle placement suggestive of a necrotizing myopathy (inflammatory myopathies, dystrophies, toxic myopathies, myotonic myopathies), whereas a lack of irritability is characteristic of long-standing myopathic disorders (muscular dystrophies, endocrine myopathies, disuse atrophy, and many of the metabolic myopathies). In addition, the EMG may demonstrate myotonic discharges that will narrow the differential diagnosis (Table 462e-4). Another important EMG finding is the presence of short-duration, small-amplitude, polyphasic motor unit action potentials (MUAPs). Such MUAPs can be seen in both myopathic and neuropathic disorders; however, the recruitment or firing pattern is different. In myopathies, the MUAPs fire early but at a normal rate to compensate for the loss of individual muscle fibers, whereas in neurogenic disorders the MUAPs fire faster. The EMG is usually normal in steroid or disuse myopathy, both of which are associated with type 2 fiber atrophy; this is because the EMG preferentially assesses the physiologic function of type 1 fibers. The EMG can also be invaluable in helping to choose an appropriately affected muscle to sample for biopsy.

**DNA Analysis** This serves as an important tool for the definitive diagnosis of many muscle disorders. Nevertheless, there are a number of limitations in currently available molecular diagnostics. For example, in Duchenne and Becker dystrophies, two-thirds of patients have deletion or duplication mutations in the dystrophin gene that are easy to detect, while the remainder have point mutations that are much more difficult to find. For patients without identifiable gene defects, the muscle biopsy remains the main diagnostic tool.

**TABLE 462e-3 DRUGS THAT CAUSE TRUE MYALGIA**

| |
|---|
| Cimetidine |
| Cocaine |
| Cyclosporine |
| Danazol |
| Emetine |
| Gold |
| Heroin |
| Labetalol |
| Methadone |
| D-Penicillamine |
| Statins and other cholesterol-lowering agents |
| L-Tryptophan |
| Zidovudine |

**TABLE 462e-4 MYOTONIC DISORDERS**

| |
|---|
| Myotonic dystrophy type 1 |
| Myotonic dystrophy type 2/proximal myotonic myopathy |
| Myotonia congenita |
| Paramyotonia congenita |
| Hyperkalemic periodic paralysis |
| Chondrodystrophic myotonia (Schwartz-Jampel syndrome) |
| Centronuclear/myotubular myopathy[a] |
| Drug-induced |
| Cholesterol-lowering agents (statin medications, fibrates) |
| Cyclosporine |
| Chloroquine |
| Glycogen storage disorders[a] (Pompe's disease, branching enzyme deficiency, debranching enzyme deficiency) |
| Myofibrillar myopathies (MFM)[a] |

[a]Associated with myotonic discharges on electromyography but no clinical myotonia.