TABLE 122-2 WORK UP FOR A PATIENT WITH SUSPECTED MYOPATHY

FEATURE	DESCRIPTION
HISTORY	
Age of onset	Congenital, childhood, adult
Chronic, progressive, or episodic	Dystrophies are usually progressive; congenital static; metabolic/channelopathies episodic
Triggers	Exercise, foods, temperatures
FAMILY HISTORY	Dominant, recessive, or no family history
WEAKNESS ON EXAM	
Proximal	Difficulty lifting objects, climbing stairs, getting up from chair, scapular winging, waddling gait, Gower's sign
Distal	Difficulty making a tight fist, fastening buttons, opening jars, wrist drop, foot drop
Facial	Difficulty squeezing the eyes shut, transvers smile, inability to pucker or blow out cheeks, inability to whistle
Oculopharyngeal	Ptosis, restricted extra ocular movements, coughing after drinking, and difficulty swallowing
Cardiac	Cardiac conduction defects, cardiomyopathy
Respiratory	Using accessory muscles, difficulty lying flat
LABORATORY	
CK	Dystrophies/inflammatory myopathy increased >10x normal; congenital 3-5x normal; metabolic >10x normal during attacks
Thyroid / Parathyroid	High TSH, low T4, low PTH, Ca^{2+}
ELECTRODIAGNOSTIC STUDIES	Irritated muscle shows fibrillations and positive sharp waves; myopathic motor units are brief, low amplitude, and polyphasic; myotonia spontaneous waxing and waning motor unit amplitude and frequency
MUSCLE BIOPSY	Changes in muscle fiber shape and composition of fiber types, amount of connective tissue, presence of inflammatory cells, necrotic muscle fibers, regenerating fibers, number or morphology of mitochondria, or abnormal deposits of fat or glycogen
GENETIC TESTING	Confirmatory for inherited myopathies

TABLE 122-3 INHERITANCE PATTERN IN GENETIC MYOPATHIES

X-LINKED	Oculopharyngeal muscular dystrophy
Duchenne/Becker muscular dystrophy	Channelopathies
Emory-Dreifuss muscular dystrophy	Central core myopathy
AUTOSOMAL DOMINANT	**AUTOSOMAL RECESSIVE**
Myotonic dystrophy types 1 and 2	Limb-girdle muscular dystrophy (2A-2S)
Facioscapulohumeral muscular dystrophy	Metabolic myopathies
Limb-girdle muscular dystrophy (1A-1H)	Recessive myotonia congenita
	MATERNAL TRANSMISSION
	Mitochondrial myopathies

TABLE 122-4 MODIFIED MEDICAL RESEARCH COUNCIL MOTOR STRENGTH TESTING SCALE

GRADE	DEGREE OF STRENGTH
5	Normal strength through entire range of motion and against resistance
5-	Equivocal, barely detectable weakness
4+	Able to move against gravity and resistance, but examiner can break
4	Able to move against gravity and some resistance
4-	Able to resist gravity but only minimal resistance
3+	Able to overcome gravity and transient resistance, but then quickly gives out
3	Able to overcome gravity but no resistance
3-	Able to resist gravity but not through full range of motion
2	Able to move through range of motion with gravity eliminated
1	Trace muscle contraction
0	No contraction

pattern and degree of involvement of various muscles (Table 122-4). But just as important as isolated strength testing are functional motor tasks, particularly in children. Patients may have difficulty climbing stairs, rising from a low chair or toilet, getting up from the floor, trouble lifting objects over their heads and washing or brushing their hair, or difficulty opening jar tops and fastening buttons. Muscles should be inspected for atrophy or hypertrophy and range of motion around the joints for evidence of tendon contractures. Ten broad patterns of muscle weakness occur in myopathies (Table 122-5). Most myopathies have the proximal, limb-girdle pattern. There are other, highly distinctive, patterns. Weakness that is asymmetric and includes the face, proximal arms and shoulders, and distal lower extremities is characteristic of facioscapulohumeral muscular dystrophy. Weakness that starts in the distal finger flexors (patients cannot curl fingers when making a fist) and proximal lower extremities (the quadriceps) is virtually pathognomonic for sporadic inclusion body myositis. A patient in middle age who presents with ptosis and difficulty swallowing is highly characteristic for oculopharyngeal muscular dystrophy. In all cases these patterns need to be distinguished from other diseases of the nervous system causing similar patterns of weakness (see Chapters 121 and 123.).

DIAGNOSTIC TESTING

The most useful initial laboratory study is the serum creatine kinase (CK), which is commonly elevated in both inherited and acquired myopathies. Despite the obvious localizing value of elevated muscle enzymes it is important to remember not all elevations in serum CK are due to myopathy (Table 122-6).

Electrodiagnostic testing can help distinguish between neurogenic and myopathic causes for weakness. In muscle disease nerve conduction studies are normal. Changes on electromyography characteristic for muscle disease include: chronic changes characterized by small, brief duration, polyphasic motor units; and more acute changes (irritable myopathic changes), which include fibrillations or positive sharp waves.

Muscle biopsies can be an important diagnostic test in patients whose family history and physical examination does not suggest a particular myopathic diagnosis. Characteristic morphological changes are hallmarks of the congenital myopathies (e.g., central core disease, or centronuclear myopathy), inflammatory myopathies (dermatomyositis and polymyositis), and metabolic myopathies (glycogen storage disorders), but most myopathies result in nonspecific muscle changes including rounding of muscle fibers, variation in fiber size, and increased number of internal nuclei.

In patients for whom the family history is suggestive of an inherited myopathy or in which the diagnosis is important to help guide surveillance or treatment, genetic testing is confirmatory.