Institute of Genomic Medicine and Rare Disorders
Semmelweis University

Head of department: Prof. Dr. Mária Judit Molnár
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Muscular dystrophies protocol

The protocol to be used for muscular dystrophies at Semmelweis University, Institute of Genomic Medicine and Rare Disorders follows reference works of:

[1] *Narayanaswami P, Carter G, David W, Weiss M, Amato AA*. **Evidence-based guideline summary: Diagnosis and treatment of limb-girdle and distal dystrophies: Report of the Guideline Development Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine**. Neurology. 2015 Apr 21;84(16):1720-1.
[2] **Bradley's Neurology in Clinical Practice 7e**
[3] **http://www.uptodate.com/contents/limb-girdle-muscular dystrophy?source=search_result&search=lgmd&selectedTitle=1~24 (Accesed: 2016/06/16)**

Definition:

Muscular dystrophies are inherited group of progressive myopathic disorders resulting from defects in genes required for normal muscle function. Muscle weakness is the primary symptom.

Use of the protocol:

1.) When a patient presents with muscle weakness first the anatomic site of the weakness has to be clarified (i.e. confirm that the origin of weakness is due to skeletal muscle disorder). Pattern of weakness, neurologic examination, laboratory studies (particularly creatine kinase), and electrophysiologic studies point to a primary skeletal muscle disease.

2.) Primary skeletal muscle diseases may be genetic in origin, or be acquired. Follow **"Evaluation of patient with muscle weakness"** and consider possibly acquired causes of skeletal muscle disease: "**Table: major causes of myopathy**"

3.) If muscular dystrophy is the suspected cause of the muscle weakness follow the protocol "**Conceptual approach to a patient with suspected limb-girdle muscular dystrophy**"

4.) If clinical features, ethnicity, inheritance are characteristic consider genetic testing without muscle biopsy. Use flowcharts: **"Suspected muscular dystrophy with autosomal dominant / autosomal recessive / X-linked recessive inheritance pattern"**

5.) If clinical features are not characteristic consider muscle biopsy followed by genetic testing.

6.) If metabolic myopathy is suspected clinically follow "**Diagnostic approach to patients with suspected metabolic myopathy"**

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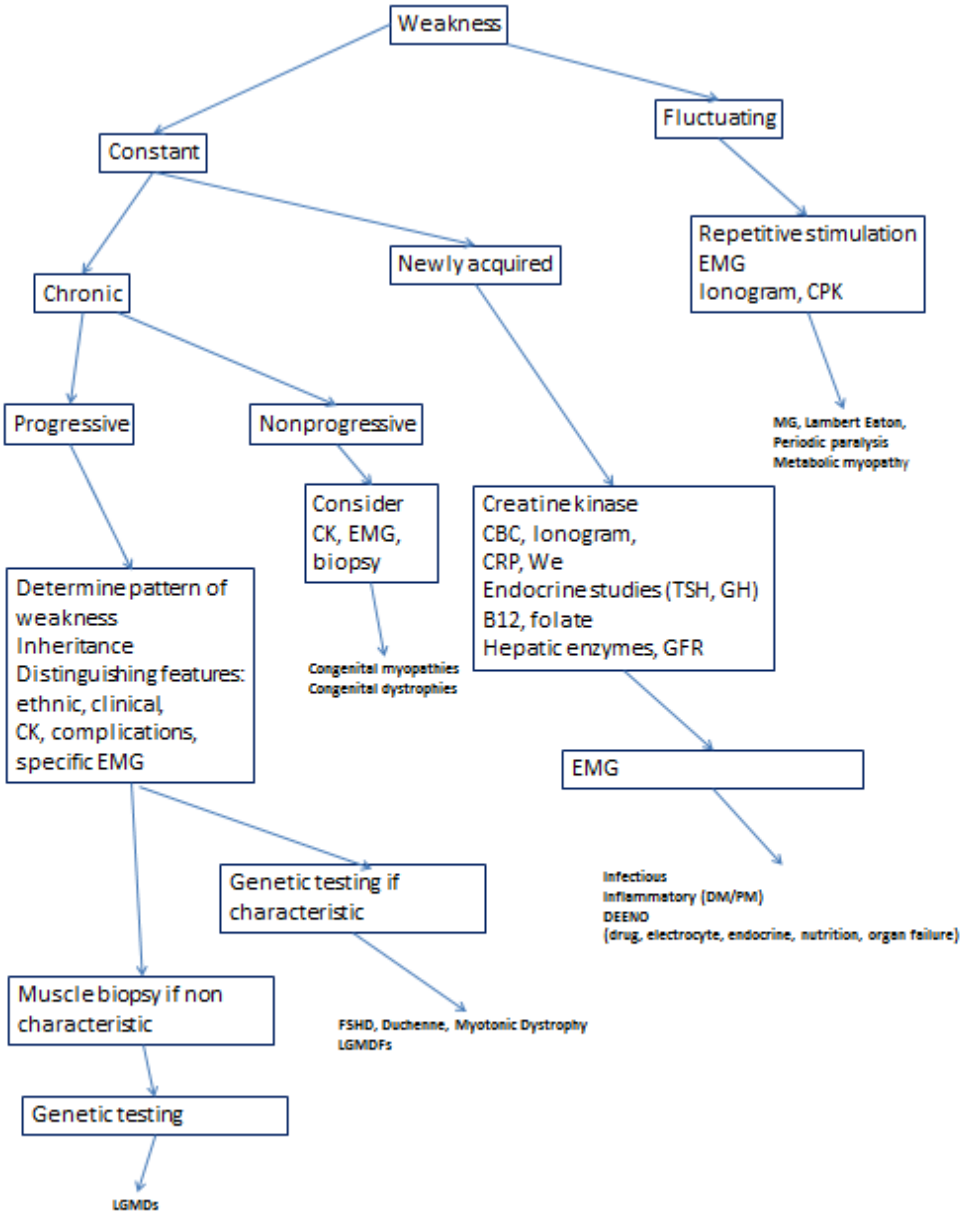
Evaluation of patient with muscle weakness (adapted from Bradley's Neurology 7e):



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http://www.uptodate.com/contents/search?search=myopathy&sp=0&searchType=PLAIN_TEXT&source=USER_INPUT&searchControl=TOP_PULLDOWN&searchOffset= **Accesed: 2016/06/16**

Major causes of myopathy

Inflammatory
Polymyositis
Dermatomyositis
Inclusion body myositis
Juvenile dermatomyositis
Vasculitis
Overlap syndromes - lupus, scleroderma, rheumatoid arthritis, Sjögren's syndrome
Rheumatoid arthritis, Sjögren's syndrome
Endocrine disorders
Hypothyroidism
Cushing's syndrome (or exogenous steroid administration)
Electrolyte disorders
Hypokalemia
Hypophosphatemia
Hypocalcemia
Hypernatremia or hyponatremia
Metabolic myopathies
Disorders of carbohydrate, lipid, and purine metabolism

Drugs and toxins
Illicit drugs - cocaine, heroin
Alcohol
Corticosteroids
Other - colchicine, antimalarial drugs, HMG-CoA reductase inhibitors, penicillamine, zidovudine
Infections
Viral - influenza, parainfluenza, Coxsackie, HIV, cytomegalovirus, echovirus, adenovirus, Epstein-Barr virus
Bacterial - pyomyositis, lyme myositis
Fungal
Parasitic - trichinosis, toxoplasmosis
Rhabdomyolysis
Crush trauma
Seizures
Alcohol abuse, including hyperkinetic state with delirium tremens
Exertion, especially with environmental heat illness
Vascular surgery
Malignant hyperthermia
Inherited myopathies
Acid maltase deficiency
Muscular dystrophy

HIV: human immunodeficiency virus.

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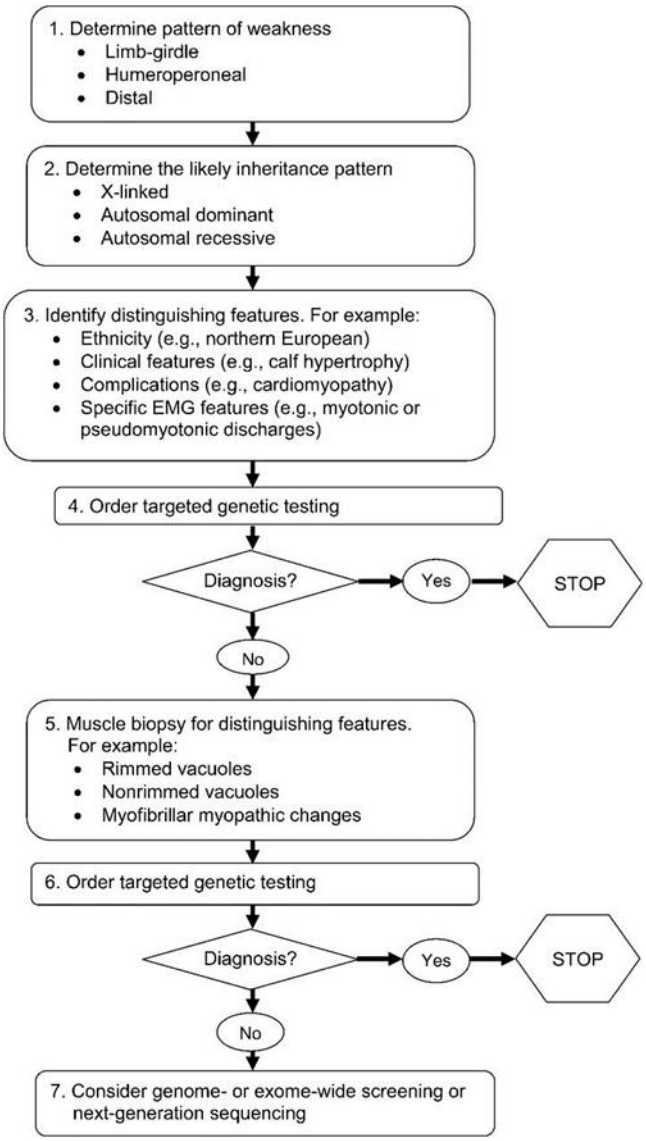
Conceptual approach to a patient with a suspected limb-girdle muscular dystrophy.



Pushpa Narayanaswami et al. Neurology 2014;83:1453-1463

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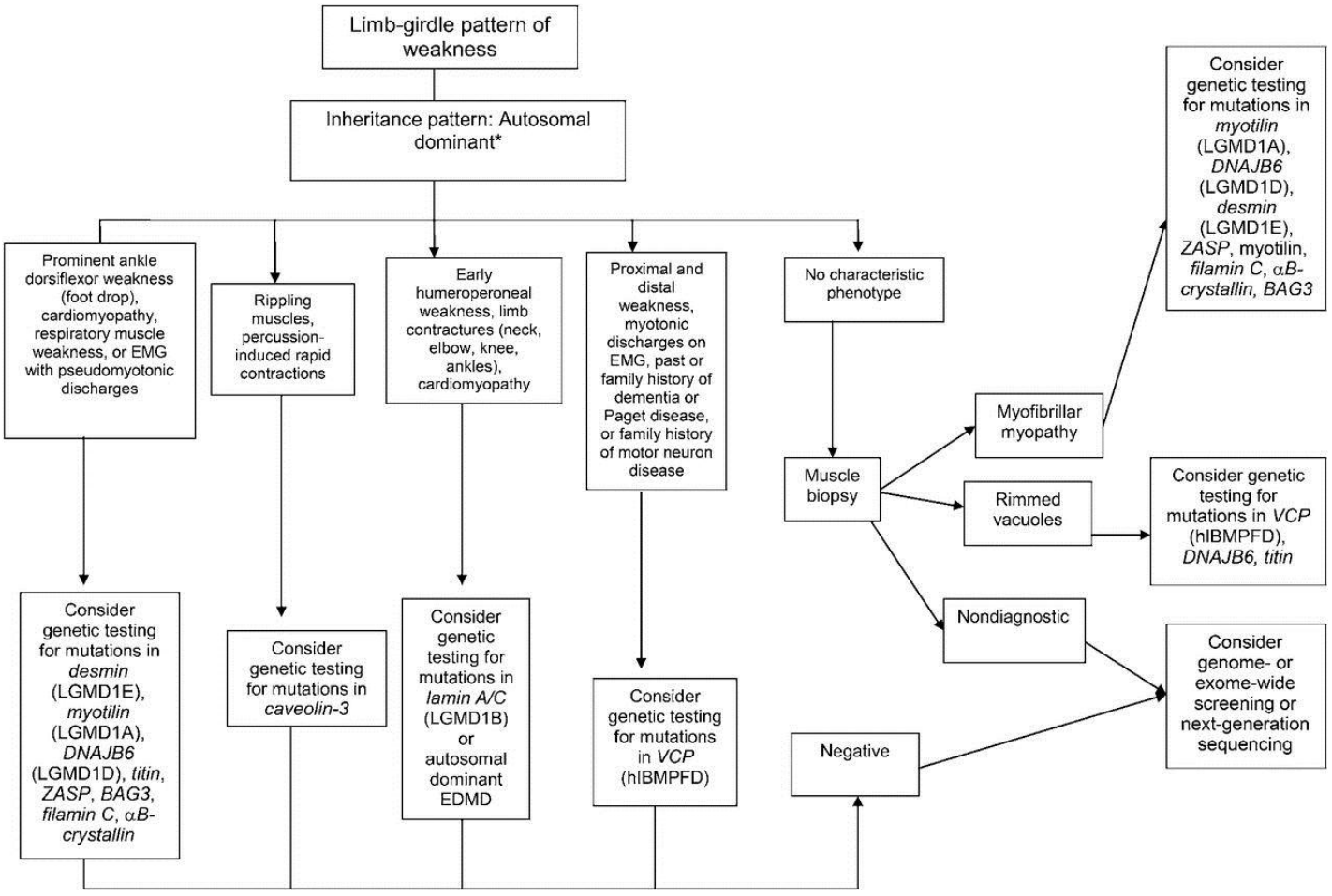
Diagnostic approach to patients with a limb-girdle pattern of weakness and suspected muscular dystrophy with an autosomal dominant inheritance pattern



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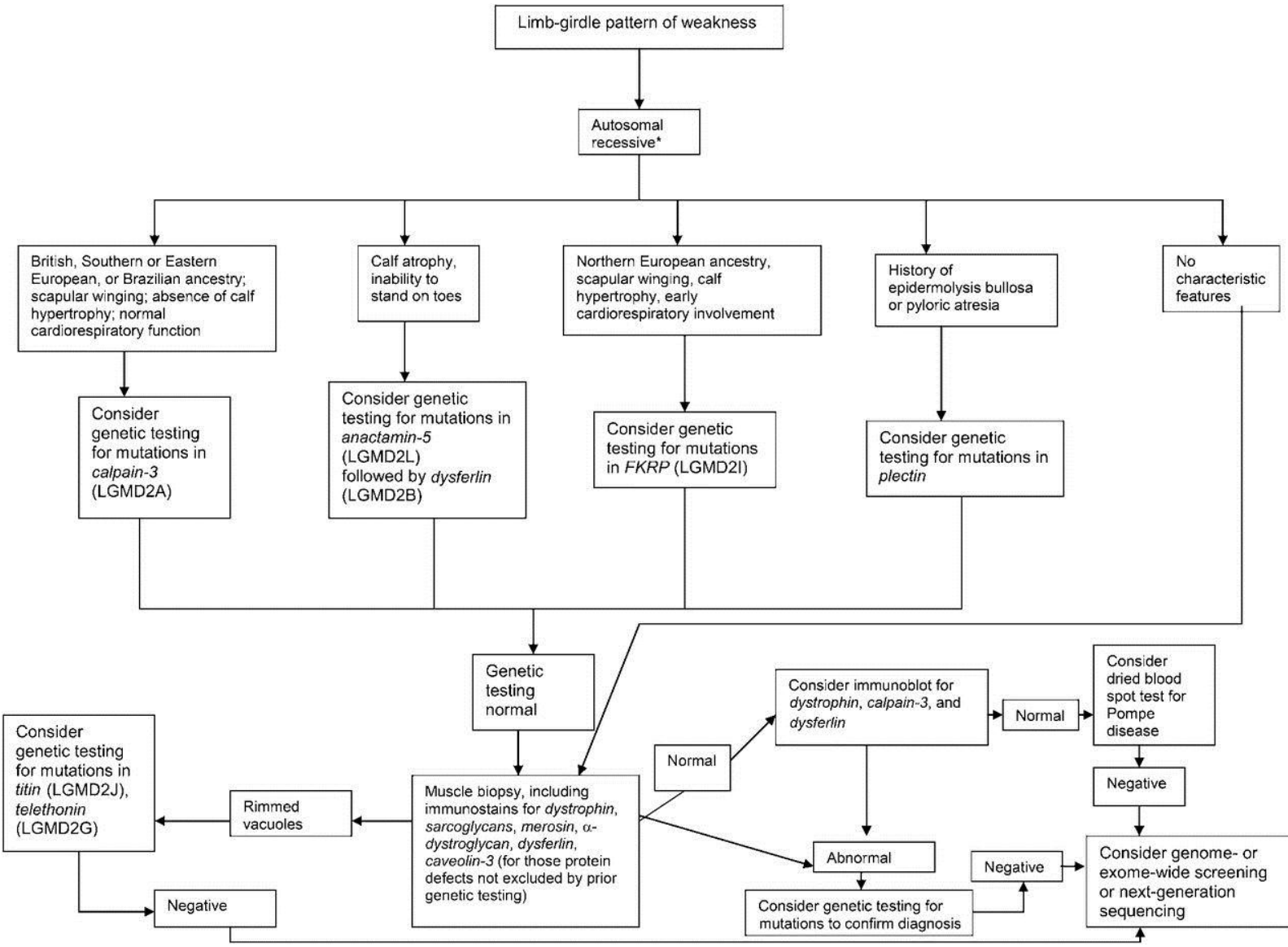
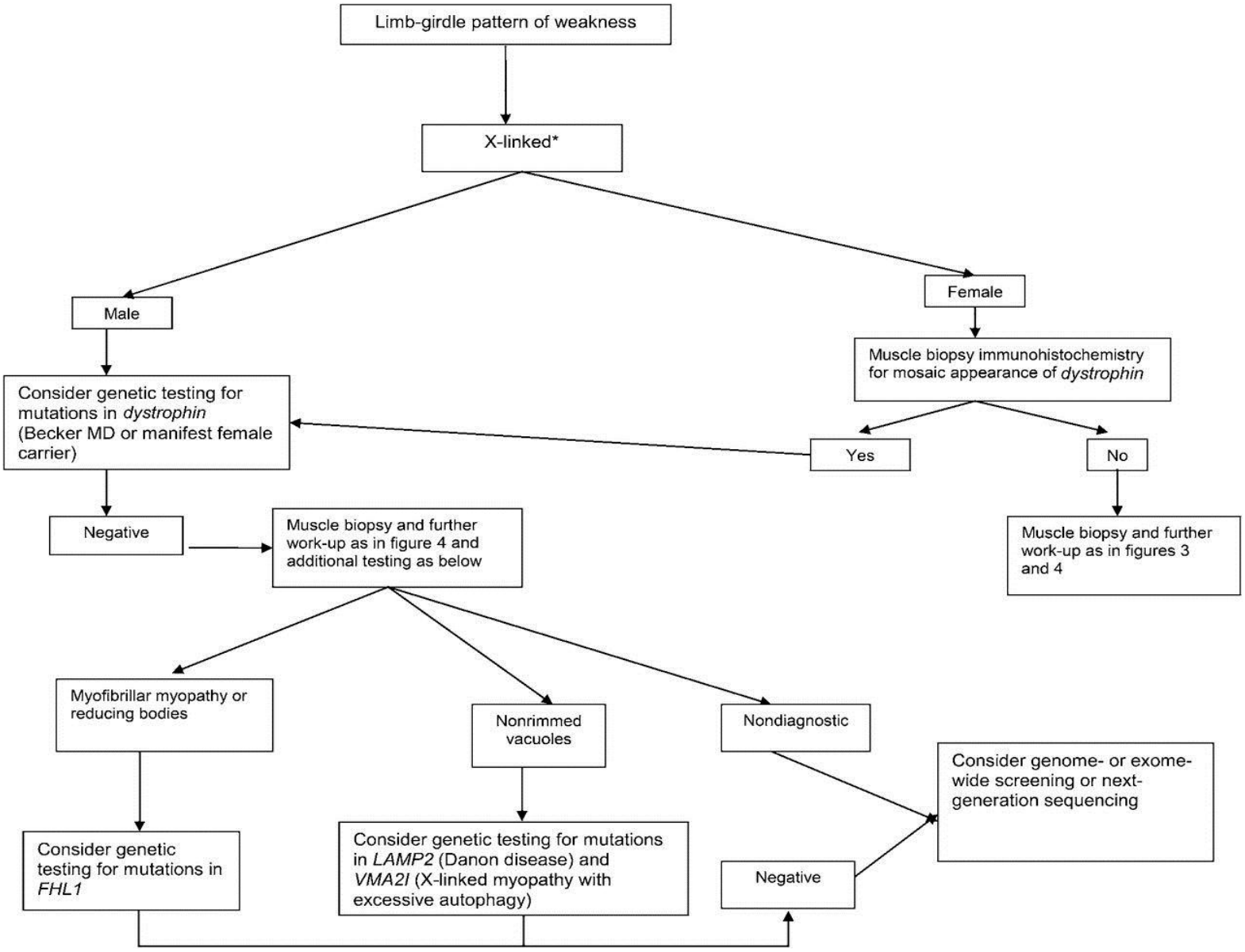
Diagnostic approach to patients with a limb-girdle pattern of weakness and suspected muscular dystrophy with an autosomal recessive inheritance pattern



Pushpa Narayanaswami et al. Neurology 2014;83:1453-1463

Diagnostic approach to patients with a limb-girdle pattern of weakness and suspected muscular dystrophy with an X-linked recessive inheritance pattern



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Diagnostic approach to patients with suspected metabolic myopathy

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