Neuromuscular disease in practice: polyneuropathies and myopathies

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Disclusure information: none

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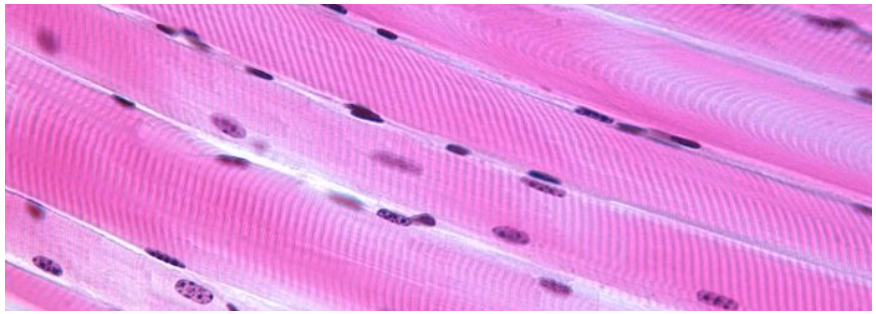
Learning objective

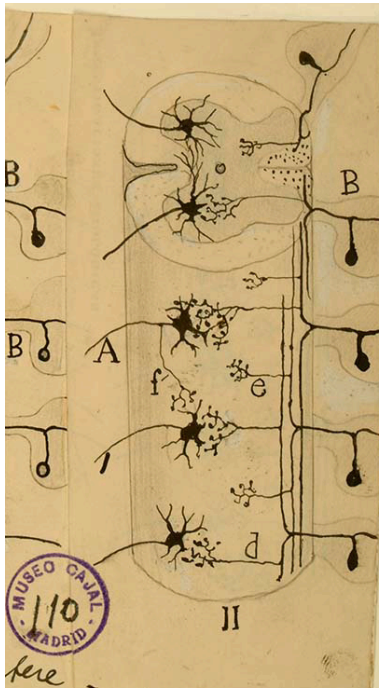
To review a systematic method to diagnose neuropathies and myopathies

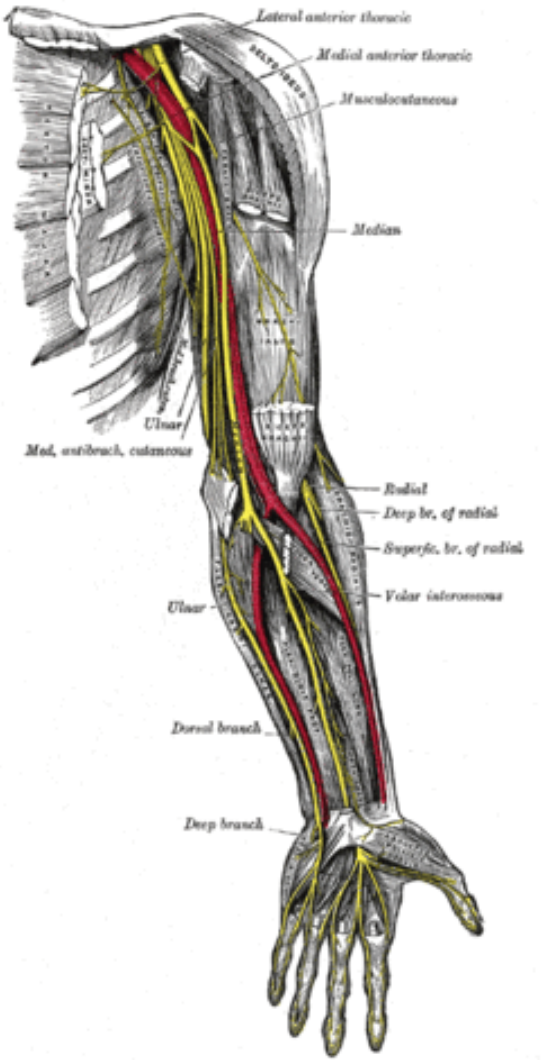
Key message

Knowledge of pathology and an orderly procedure are fundamental

Methods to approach a patient with either a myopathy or a polyneuropathy

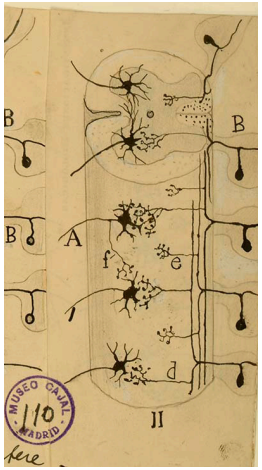






The prevalence of peripheral neuropathy is estimated to be between 2% and 8%.

Determining the etiology of a polyneuropathy can be challenging.



Ten steps in characterizing and diagnosing patients with peripheral neuropathy

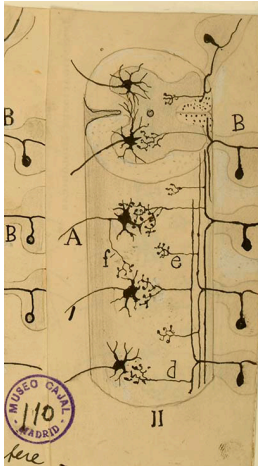
Step 1. Characterize the anatomic-pathologic pattern of involvement

Step 2. Confirm the inferred anatomic-pathologic pattern by use of characterizing tests

Step 3. Infer the pathologic site and mechanism of nerve fiber alterations.

Step 4. Consider the onset and course of neuropathy.

Step 5. Decide whether the disorder is likely to be inherited or acquired.



Dyck PJ el at. Neurology 1996; 47:10-17

Ten steps in characterizing and diagnosing patients with peripheral neuropathy

Step 6. Check for associations with present or past diseases.

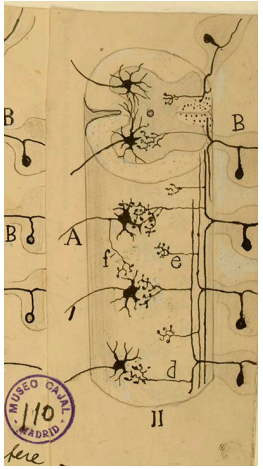
Step 7. Perform hematologic, biochemical, serologic, imaging, and other tests.

Step 8. Evaluate kin.

Step 9. Perform a cutaneous nerve biopsy.

Step 10. Perform a therapeutic trial.

Dyck PJ el at. Neurology 1996; 47:10-17



Ten steps in characterizing and diagnosing patients with peripheral neuropathy

Step 1. Characterize the anatomic-pathologic pattern of involvement

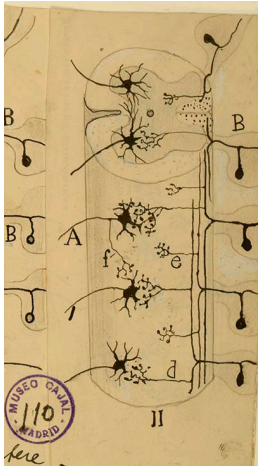
Localice the disorder to a

part of the peripheral nervous system (roots, ganglia, plexuses, nerves) ,

functional or size class of neurons (fibers) or

part of the neuron (soma or distal axon)

Reduces the list of possible causes



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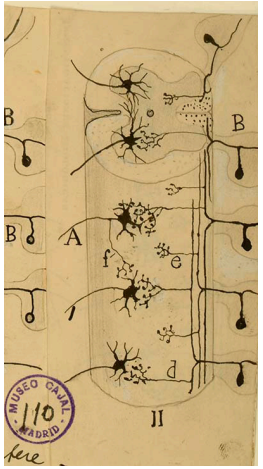
Ten steps in characterizing and diagnosing patients with peripheral neuropathy

Step 4. Consider the onset and course of neuropathy.

The temporal course of the onset and evolution give clues to diagnosis

Dyck PJ el at. Neurology 1996; 47:10-17



Ten steps in characterizing and diagnosing patients with peripheral neuropathy

Step 5. Decide whether the disorder is likely to be inherited or acquired.

"lack of prickling suggests inherited neuropathy" (but pure motor involvement or chronic adquired cases…)

Insidiuos progression over years

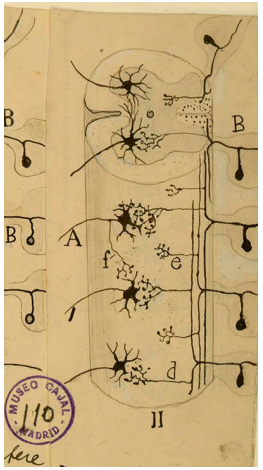
Cutaneous or bony abnormalities

Family history

Typical phenotype

Suggest inherited neuropathies

Dyck PJ el at. Neurology 1996; 47:10-17

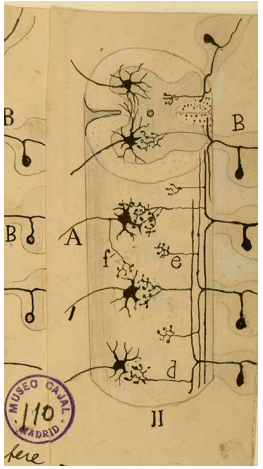


Ten steps in characterizing and diagnosing patients with peripheral neuropathy

Step 7. Perform hematologic, biochemical, serologic, imaging, and other tests.

The degree to which test should be done is not a simple matter

tests that provide the highest yield of abnormality are blood glucose, serum B12 with metabolites (methylmalonic acid with or without homocysteine), and serum protein immunofixation electrophoresis (Level C). If there is no definite evidence of diabetes mellitus by routine testing of blood glucose, testing for impaired glucose tolerance may be considered in distal symmetric sensory polyneuropathy (Level C)



Practice Parameter: Evaluation of distal symmetric polyneuropathy: Role of laboratory and genetic testing (an evidence-based review)
Neurology 2009;72:185 Current guideline. Reaffirmed on July 13, 2013.

Ten steps in characterizing and diagnosing patients with peripheral neuropathy

Step 9. Perform a cutaneous nerve biopsy.

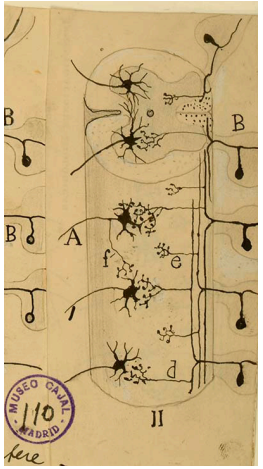
Useful recognizing

Inflammation (necrotizing vasculiits, inflamamtory demielination, granuloma)

Infiltration (amylioidosis, lymphoma)

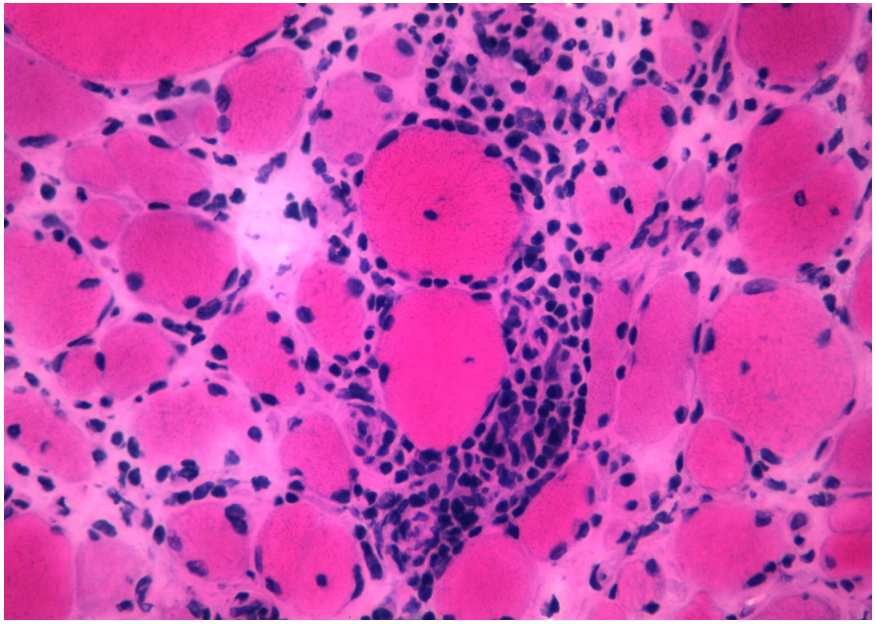
Unique tissue reaction (tomaculae, excesive glycogen deposits)

Nerve biopsy is generally accepted as useful in the evaluation of certain neuropathies as in patients with suspected amyloid neuropathy, mononeuropathy multiplex due to vasculitis, or with atypical forms of chronic inflammatory demyelinating polyneuropathy (CIDP). However, the literature is insufficient to provide a recommendation regarding when a nerve biopsy may be useful in the evaluation of DSP (Level U)



Practice Parameter: Evaluation of distal symmetric polyneuropathy: Role of autonomic testing, nerve biopsy, and skin biopsy (an evidence-based review)
Neurology 2009; 72:177 Current guideline. Reaffirmed on July 13, 2013.

A pattern recognition approach to patients with a suspected myopathy



Barohn RJ et al. Neurol Clin 2014; 32: 569–593

Which negative and/or positive symptoms do patients demonstrate

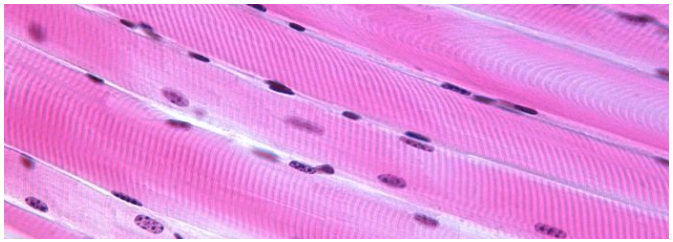
What is the temporal evolution

Is there a family history of a myopathy

Are there precipitating factors that trigger episodic weakness

Are there associated systemic symptoms or signs

What is the distribution of weakness



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Are there precipit

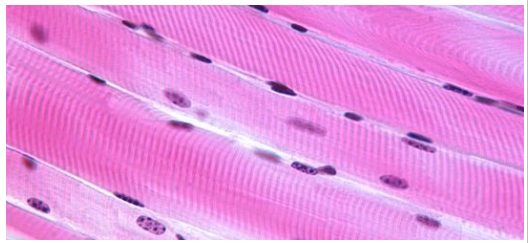
Illegal drugs or pre

Excercise followed

Excercise plus carl

Fever

Cold exposure



Box 9
Drugs that can cause toxic myopathies

Inflammatory
- Cimetidine
- D-penicillamine
- Procainamide
- L-tryptophan
- L-dopa

Noninflammatory necrotizing or vacuolar
- Alcohol
- Cholesterol-lowering agents
- Chloroquine
- Colchicine
- Cyclosporine and tacrolimus
- Emetine
- ε-aminocaproic acid
- Isoretinoic acid (vitamin A analogue)
- Labetalol
- Vincristine

Rhabdomyolysis and myoglobinuria
- Alcohol
- Amphetamine
- Cholesterol-lowering drugs
- Cocaine
- Heroin
- Toluene
- ε-aminocaproic acid

Myosin loss
- Nondepolarizing neuromuscular blocking agents
- Steroids

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Are there associated systemic symptoms or signs

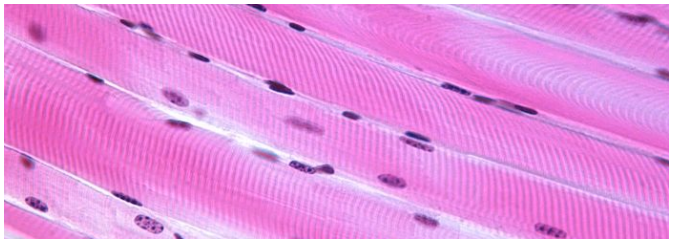
Cardiac disease

Respiratory failure (may be initial symptom of acide maltase def. myotonic dystrophy, centronuclear and nemaline myopathies)

Hepatomegaly

Mental retardation, cataracts

Rash



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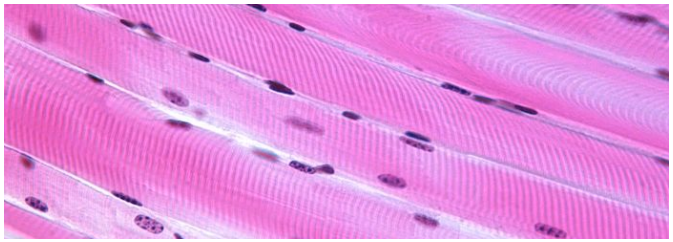
Pattern 1 limb-girdle weakness

Pattern 2 distal weakness

Pattern 3 scapulo-peroneal (proximal arm/distal leg weakness)

Pattern 4 distal arm/proximal leg weakness

Pattern 5 ptosis with or without ophtalmoparesis



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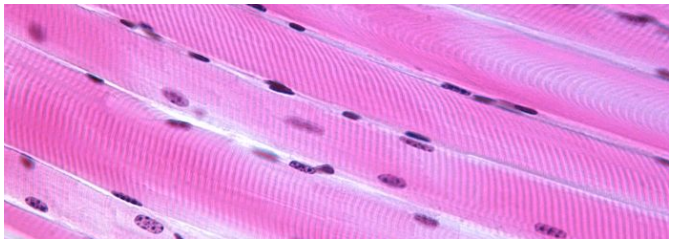
Pattern 6 prominente neck extensor weakness (drop head)

Pattern 7 bulbar weakness

Pattern 8 episodic pain, weakness and myoglobinuria

Pattern 9 episodic weakness delayed or unrelated to excercise

Pattern 10 stiffness and decrease ability to relax



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Pattern 2 distal weakness

Distal myopathies (Welander, Markesbery, Nonaka, Miyoshi, Laing)

Myotonic dystrophy

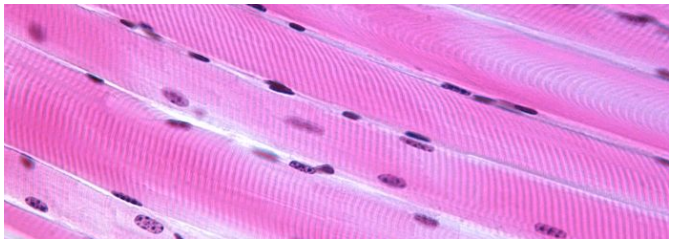
Inclusion body myositis

Hereditary inclusion body myopathy

Centronuclear myopathy

Myofibrillary myopathy

Debrancher deficiency



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Pattern 5 ptosis with or without ophtalmoparesis

<u>Ptosis without ophthalmoparesis</u>

Congenital myopathies (Nemaline and Central core myopathies)

Desmin (myofibrillar) myopathy

Myotonic dystrophy

<u>Ptosis with ophthalmoparesis</u>

Centronuclear myopathy

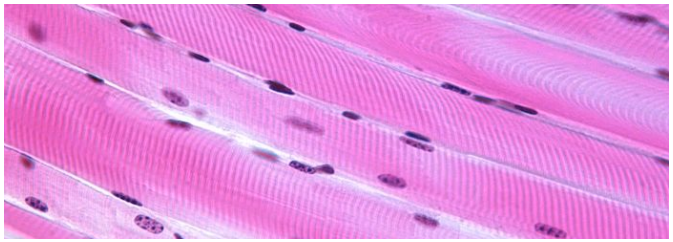
Mitochondrial myopathy

Multicore disease

Oculopharyngeal muscular dystrophy

Oculopharyngodistal myopathy

Neuromuscular junction disease (myasthenia gravis, Lambert-Eaton, botulism)



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