Assessing neuromuscular disease in your general practice

Presenting muscle weakness

Muscle weakness is a common presenting symptom with a wide range of possible causes. As healthcare providers, you are well-positioned to **assess** your patient's symptoms and initiate the diagnostic process.

Take action: assess muscle weakness in your patient

When a patient presents to your practice with muscle weakness, two simple initial steps can help you arrive at a conditional diagnosis or make a detailed referral to the appropriate specialist. It is important to remember that some neuromuscular diseases are multi-system disorders and multiple organ systems may be involved.[1]

Assessment of myopathy should begin with taking a medical history and performing a focused physical examination to assess your patient's muscle strength and function.[2]

Taking a history

The nature of the complaint, its location and pattern of symptoms – in addition to other details described below – can focus the diagnostic process, and help to inform further tests and consultations, or your referral to the appropriate specialist.

Define weakness

Determine what the patient means by weakness – for example, are they describing fatigue, lack of endurance, motor abnormalities? Symptoms of muscle disease are generally categorized as "negative" complaints such as weakness, exercise intolerance and fatigue, or "positive" complaints such as spasms, cramps or muscle stiffness.[2,3]

Identify location

Determining the location and pattern of neuromuscular symptoms is integral to the process of identifying the disease or etiology that triggered your patient's consultation.[3]

Determine anatomic distribution / **pattern of weakness** and focal wasting or hypertrophy of muscle groups:[4]

- Symmetrical versus asymmetrical
- Predominantly proximal versus distal or generalized (a key differentiating factor)
- Upper versus lower limb predominance
- Presence versus absence of bulbar involvement

Associated symptoms

Certain associated symptoms may help locate the area(s) of damage within the nervous system, while other symptoms may help place the condition in the larger context of disease.[4,5] The initial question would be whether the muscle weakness accompanied by any systemic signs or symptoms, such as sensory changes, cramping, aching, stiffness or pain?[1]

Temporal characteristics: onset and course

- Was the symptom onset acute (days to weeks) or chronic (months to years); episodic?
- Are symptoms worsening, stable or improving?
- If strength is deteriorating, attempt to determine the rate of progression (i.e., has weakness increased over days, weeks, months or years?)
- Is there any history of potentially related exposure to medications, toxins or infectious agents?[5]

Contributing factors

Identify factors that aggravate or alleviate the primary symptoms (e.g., exercise versus rest).

Family history

Assessment of the patient's medical family history may reveal a known history of hereditary disorders, including myopathy, neuropathy, channelopathy, malignant hyperthermia, etc. OR non-specific symptoms, such as the "Smith family feet" (high arches and hammer toes suggesting Charcot-Marie-Tooth neuropathy), my mom and grandfather "walked funny" or "were in a wheelchair later in life", etc.[6]

Key questions to ask your patient[7,8]

1. Were you or someone in the family ever diagnosed with an elevated CK level (creatine kinase, a muscle enzyme)?
2. Have your or a family member's liver enzymes ever been elevated without apparent reason?
3. Is it particularly challenging to walk uphill or climb stairs?
4. Have you noticed changes in your ability to perform routine tasks (e.g., lift a bag of groceries)?
5. Do you have difficulties standing up from a seated position or getting up from the floor?
6. Has your performance in sports and other physical activities declined?
7. Do you often stumble when you walk or do your feet feel "sticky"?
8. Do you have breathing difficulties, during mild/moderate exercise, when lying down, or while sleeping?
9. Do you have morning headaches and/or daytime sleepiness (related to breathing problems)?
10. Do you have frequent chest infections with prolonged recovery time?
11. Are you aware of any relevant family / personal medical history?

Practice point

Myopathies are generally proximal and symmetrical, so that with rare exceptions, patients present with weakness affecting the arms and legs, as opposed to the hands and feet.[3,4]

Physical examination

Muscle function and muscle tone can be assessed in your office simply and easily using the approaches below.

Test muscle function

You can perform an efficient practical in-office evaluation of muscle function using the **Medical Research Council (MRC) scale** (0 to 5).[9,10]

- Test shoulder abduction, hip flexion and adduction
- Passive ranges of motion (to check for contractures)
- Ask the patient to walk in the office so you can evaluate gait characteristics: watch for abnormalities such as toe walking, excessive lordosis, Trendelenburg or antalgic gait, etc.
- Gower manoeuvre: observe the patient rising from a seated position and note their need to use their arms to help them stand (using arms to walk up the thighs)

Assess muscle tone

Evaluation of muscle tone also provides important diagnostic clues. Pathologically increased tone may manifest as spasticity – a sudden resistance during rapid passive movement felt as a "catch and release" (associated with upper motor neuron disorders) – or as rigidity – an increase in tone that persists throughout the passive range of motion (associated with basal ganglia disorders).[9]

Increased muscle tone can be observed in patients by rapidly extending the forearm (biceps tone). Test for myotonia by asking the individual to reverse a muscle action quickly (i.e., trying to rapidly open a tightly clenched fist) or to quickly get up from a chair and take a few steps.[5,9]

The lower limbs can be evaluated with the patient seated with the legs dangling. Spasticity is best assessed in the ankles with a passive and rapid dorsi-flexion movement.[9]

Making a referral

If you suspect your patient's symptoms have a neuromuscular cause based on the medical history and clinical assessment, refer the patient to a neurologist or neuromuscular specialist for further assessments. These might include an EMG/nerve conduction study, a muscle biopsy, a dried blood spot (DBS) analysis and/or genetic testing.

Today, improved accessibility of diagnostic tests and in many cases, treatment, make **timely referral** important.[6]

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