Clinical Reasoning: A teenager with left arm weakness

**SECTION 1:** This is your patient presentation. For the history, start at the beginning of the patient’s symptoms and proceed chronologically.

**QUESTIONS:** Clinical Reasoning articles are designed to teach thought process. What should the reader think when they see this type of patient?

**PRO TIP:** For many cases, the first step is to assess localization and then make a differential diagnosis.

**SECTION 2:** Make sure you answer the questions you just asked!

**DISCUSSION:** The last section is a discussion of the final diagnosis.

**DISCUSSION:** Start by defining the disease process, it helps to cite a recent review article in case the reader wants to learn more.

**DISCUSSION:** Connect the general disease process to your patient’s presentation.

**TITLE:** This is your take-home point. Include clinical features that helped clinch the diagnosis.

**CASE REPORT hint:** Use concise, specific language to convey timing information. Use time points and words like acutely or progressively.

**DIFFERENTIAL DIAGNOSIS:** When giving a differential, clarify which features support or go against that etiology.

**REFERENCES:** Each statement or group of statements that is not general knowledge should have a reference. Make sure the information you are citing comes from the paper you list as the reference. Make sure references are in the order they appear.

**PRO TIP:** For many cases, the first step is to assess localization and then make a differential diagnosis.

**GO TO SECTION 2**

**Section 1**
A 16-year-old right-handed boy presented with left arm weakness. He had broken his right humerus in November 2015 and was in cast until January 2016. During that time, he noticed left arm weakness while carrying a heavy backpack. The weakness improved and he was able to play lacrosse during spring 2016. In summer 2016, he noticed substantial weakness of his left arm after rowing 7 km, and sought medical attention. He denied any pain or sensory symptoms, lower limb weakness, and bladder or bowel symptoms. There was no preceding illness or trauma. On neurologic examination in October 2016, there was atrophy of the deltoid, biceps, and periscapular muscles, as well as mild scapular winging. There was weakness of the following muscles: right/left deltoid (Medical Research Council [MRC] grades): deltoids (5/5), biceps (5/4), supraspinatus (5/4/5), infraspinatus (5/4/5). First dorsal interosseous, abductor digit minimi, abductor pollicis brevis, finger extensors and flexors, wrist extension/lexion, brachioradialis, triceps, and rhombooids were 5/5. Tendon reflexes were 2+ throughout and sensory examination was normal.

**Questions for consideration:**
1. What is the localization for his presentation?
2. What are the differential diagnoses?

**Section 2**
The patient in this vignette presented with painless recurrent left arm weakness in the distribution of CS-C6 myotomes. Weakness and atrophy suggested lower motor neuron involvement. This could result from lesions of the cervical spinal cord, anterior horn cells, roots, brachial plexus, peripheral nerves, or muscles. Intramedullary lesions (e.g., syringomyelia) typically involve spinothalamic tracts initially leading to dissociated sensory loss (not seen in our case) before extension to the anterior horn cells. Anterior horn cell syndrome can be due to various disorders: poliomyelitis or acute flaccid myelitis (recurrent episodes make them unlikely); genetic motor neuronopathies; spinal muscular atrophy; Kennedy disease (unlikely as these disorders cause diffuse symmetric involvement with hypo/areflexia); distal hereditary motor neuronopathies (unlikely as there was no distal weakness); juvenile variant of amyotrophic lateral sclerosis (no upper motor neuron signs); and Hirayama disease (unlike as it presents with distal upper extremity weakness and atrophy). Lesions of the cervical roots (CS-6) could present in a similar way but lack of neck pain, sensory symptoms, and preserved biceps and brachioradialis reflexes made it less likely. Upper trunk brachial plexopathy could have similar presentation. Among these lesions, neuralgic amyotrophy usually presents with acute onset of pain (not seen in our case) followed by muscle weakness and atrophy. Multifocal motor neuropathy with conduction block can present with weakness and wasting with pre-dilection for upper limbs. Genetic myopathies, particularly facioscapulohumeral muscular dystrophy, could be considered in the differential due to the distribution of weakness. However, the patient did not have facial weakness.

**Question for consideration:**
1. What investigations can help to narrow the differential diagnosis?

**Discussion**
HNPP is characterized by recurrent entrapment neuropathy beginning in adolescence or young adulthood. HNPP is an autosomal dominant disorder caused by microdeletion of the PMP22 gene and is allelic with CMT1A, which is caused by duplication of PMP22.

HNPP typically presents with painless focal mononeuropathies. About two-thirds of patients have preceding minor trauma/compression or physical exercise, which can account for the neuropathy. Our patient’s episodes of weakness were preceded by carrying a heavy backpack and rowing.

The most common sites of mononeuropathy in HNPP are peroneal nerve at the fibular head, ulnar nerve at the elbow, and median nerve at the wrist. Other nerves include brachial plexus, radial nerve, and rarely hypoglossal or facial nerves. Other rare presentations of HNPP include recurrent positional sensory symptoms, progressive mononeuropathy, chronic polyneuropathy, and chronic inflammatory demyelinating polyneuropathy–like disorder.

**References**