Case Studies – Disorders of Muscle

Case 1
A previously healthy 30-year-old woman presents to you with difficulty walking for one year time. She reports feeling tired and having problems climbing the stairs as well as combing her hair. She works for the housekeeping department and has been accused of being lazy. You see the patient for the first time and note that she has weakness in her proximal muscles of the arms and legs.
A laboratory test of interest might include which of the following?
   a. Hemoglobin A1C
   b. CK
   c. VDRL
   d. BUN

The blood test is positive and you proceed to an EMG. There are fibrillations, small units, and early recruitment. You now suspect polymyositis.
To make the diagnosis you obtain which of the following?
   a. repetitive nerve stimulation
   b. DNA studies
   c. Muscle biopsy
   d. Genetics consult

Case 2
You are asked to see a 3-year-old boy because of a peculiar gait. On visual inspection, he appears to be a normal child with the exception of a malar rash. It is difficult to do manual muscle testing but on attempting to stand, the child crawls up his body to maintain an upright posture.
What is this referred to?
   a. Hoover’s sign
   b. Beaver’s sign
   c. Gower’s maneuver
   d. Adson’s maneuver

When the child is finally upright, you find that he has a waddling toe gait. Laboratory data include an elevated lactic dehydrogenase and a CK which is 50 times normal.
Your next step might include which of the following?
   a. EMG
   b. Nerve biopsy
   c. EKG
   d. Liver biopsy
Before you take that next step, you notice this child’s older 4-year old brother has hypertrophied calves and also toe walks. Both of these children have enlarged tongues.

You now obtain which of the following?
   a. EMG
   b. Genetics studies
   c. Echocardiogram
   d. Muscle biopsy

The more severe form of this disorder is due to an absence of which of these proteins?
   a. frataxin
   b. dystrophin
   c. galactin
   d. exon

Which of the following best describes this disorder?
   a. autosomal dominant
   b. x-linked recessive
   c. x-linked dominant
   d. autosomal recessive