Jane is a normal 12 year old girl in most respects except during strenuous exercise. She can walk to school OK, but can’t run with friends during recess, or sprint in gym class. She tells her gym teacher that she is not “being lazy” in gym, and that she just can’t sprint no matter how hard she tries. When trying to perform strenuous activity she gets fatigued easily and suffers from severe muscle cramps.

[Before going further: Think and discuss about general possibilities of what could be wrong with Jane.]

You see Jane during your family practice rotation. The attending physician doesn’t have any immediate ideas as to what could be wrong. While the attending steps out for a moment you, as a curious, insightful, bold (and foolish?) M1, decide to perform a simple test of anaerobic muscle performance. You place a blood pressure cuff around her forearm, inflate pressure to greater than systolic pressure, and ask her to perform as many muscle contractions as she can in one minute. You then remove the cuff. At 1, 2, 4 and 10 minutes post test, you are surprised to find that blood lactate level has not changed. (For the sake of this discussion, lets for the moment not think about what the attending will do to you when she gets back!).

1. Why was this test performed? Why was a cuff inflated around the arm? Why not just do the test without it? Think about energy substrates for muscle activity.

   A. What is the immediate energy source for muscle contraction?

   B. In the conversion of chemical energy into muscular work what are chemical byproducts of this process and could they affect the work performed?
2. What should normally happen to blood lactate during this type of strenuous activity? Why? Think about it in biochemical terms.

3. When the attending returns you tell her about your findings, and suggest that the patient may have a metabolic enzyme deficiency. The attending asks you what are the possible metabolic enzymes that could be affected here, and you quickly respond....????

4. How come Jane can perform aerobic activity without apparent deficit? That is, with this disorder how come she has no problem walking to school?

**Part II**

**IF** a muscle biopsy was performed before and after the ischemic fatigue test and showed no change in glycogen present in the fibers before and after the test would that help understand what is wrong with Jane?

1. Does this result help confirm your suspicion of a metabolic disorder? Does it help narrow down the possibilities? What are the candidates?

2. If the only defect Jane had was this inability to utilize glycogen, would this also explain why she could perform low intensity aerobic work without much difficulty?

   A. As a starting point think about the various “fuels” the body can use in the ultimate production of ATP for muscle use.

3. If Jane had mostly slow twitch muscle fibers would her problem be worse, better, no change? Why?

4. Any ideas to help Jane?