454 Take Home Exam 2: *Case Study (10 pts).*

Please type out-DUE Tuesday 9:30AM. Name:_____

I have neither given nor received help (except from instructor) on this exam:

Signature _____.

Case study: *Rah, rah, ouch!*

Clinical Synopsis:

In her senior year of high school, Katie was the leader of her dance team. She worked out quite regularly throughout her dance season, but midway through the season she started to complain of feeling weakness in her muscles. Her coach told her she was probably just sore for working out and needed to get used to it. It wasn't until the last month of her dance season when Katie felt dizzy and very cold which caused her to faint during an after school practice. Her mother decided it was time to take her to the doctor to see what was wrong with her.

When the doctor asked her preliminary questions about her health and lifestyle habits, Katie was first reluctant to reveal her more personal secrets. But finally, concerned about her health and future in dance, Katie explained she had an eating disorder and had been limiting her caloric intake to a bare minimum for over half a year now. She also described her muscle weakness and feelings of coldness and dizziness.

After a few tests, the doctor started to diagnose some more symptoms of Katie's health condition. The results showed she had low blood glucose levels (hypoglycemia), low ketone body levels (hypoketosis), and high ammonia levels (hyperammonemia) with some renal insufficiency. Next the doctor did a study to measure her ability to break down fatty acids (Table 1). It is possible to culture almost anyone's fibroblasts from a biopsy and these can be used for metabolic diagnosis. The doctors fed her cultured fibroblast cells three different types of ^{14}C radioactively-labeled fatty acids. After a few hours, the doctors assayed a sample of Katie's cells and measured the amount of radioactively-labeled carbon dioxide. These results were then compared to control group of patients. At that point the doctor knew what metabolic defect Katie had and looked for further treatment accordingly.

	Monitored as the production of		
	$^{14}CO_2$ from [1-^{14}C]palmitate	$^{14}CO_2$ from [1-^{14}C]octanoate	$^{3}H_2O$ from 9,10 *n*[^{3}H]palmitate
	nmol product formed/mg protein per h		
Patient	0.03; 0.03	1.61	0.08
Control	2.11±0.73 (1.24–3.18) (*n* = 12)	1.72±0.78 (1.04–3.33) (*n* = 12)	9.39±1.70 (6.75–13.40) (*n* = 12)

Table 1: Fatty acid oxidation in fibroblasts of the patient and control patients

Questions: Please answer by typing in this template document at the end of each question. Print and sign and hand in Tuesday at the beginning of class.

1. What is the likely enzyme(s) defect here? What other defect might have similar symptoms? What might be the underlying biochemical causes of the hypoglycemia, hypoketosis, and hyperammonemia?

2. Why were middle-chain fatty acids oxidized almost as effectively as the control patients?

3. There are many disorders of this type with varying severity. Considering the case history, what do you think is the specific metabolic defect in the patient? See OMIM (http://www.ncbi.nlm.nih.gov/sites/entrez?db=omim) and give the specific OMIM number for this specific form of the disorder. What chromosome is the defective gene located on?