

BCH 471
Homework (2)

-Case I-

A 28-year-old Lebanese man presents to the doctor's office with complaints of fatigue and difficulty catching his breath with his normal exercise routine since recently visiting his wife's family. He states he is otherwise healthy. He denies fever, chills, nausea, vomiting or diarrhea. He reports no sick contacts or travel other than his in-law's house where he ate their famous fava bean soup. Upon further questioning, he reports that his urine has become much more concentrated and darker despite adequate fluid intake. On examination, his sclera appeared yellowish. His hematocrit is 21% (normal:40-49%).

- What is the most likely diagnosis?
- What is the inheritance pattern and population distribution of this disorder?
- Explain how dietary challenges caused the symptoms described to appear?
- What is the appropriate treatment in this case?

-Case II-

A 38-year-old vegan woman presents to her primary care doctor with fatigue and tingling in her extremities. The symptoms have been gradually getting worse over the last year. Upon further questioning she reports frequent episodes of diarrhea and weight loss. A neurological examination revealed numbness in all extremities with decreased vibration senses. Also, a complete blood count was ordered. The results showed a markedly increased MCV, low RBC count and hemoglobin concentration.

- What is the most likely diagnosis?
- What is the most likely underlying problem for this patient?
- What is the molecular basis for the large erythrocytes?