

Intermediate Physiology 3 (PHGY 313) Term Paper Assignment

Case Study #6 (GARCIA DOS SANTOS)

Details of the Case Study:

Walter, a 21 year old man, went to the Jewish General Hospital in Montreal after feeling extreme fatigue and weakness. During his consultation with the physician, he mentioned other symptoms including headaches and shortness of breath. The physician requested a complete blood count, which showed the following results: hemoglobin, 4.7 g/dL; red blood cells (RBC), $3.4 \times 10^9/L$; hematocrit, 16%; mean corpuscular volume, 45 fL; mean corpuscular hemoglobin, 14.4 pg; white blood cells, $6.8 \times 10^6/L$; and platelets, $600 \times 10^6/L$. A blood smear revealed hypochromic and microcytic RBC. When the iron status was evaluated, it showed serum iron level of 262 $\mu\text{g}/\text{dL}$ and total iron binding capacity of 278 $\mu\text{g}/\text{dL}$. Additionally, ferritin levels were 702 ng/mL. Bone marrow aspirate stained for iron (Pearls staining) and showed more than 60% of erythroblasts with perinuclear accumulation of iron. The physician initially considered that Walter has had MDS-RARS, however, his young age and severe microcytic anemia suggested congenital sideroblastic anemia instead. Walter had his DNA extracted and ALAS2 gene sequence examined for possible mutations. A substitution of thymine with a guanine in the exon 9 of ALAS2 gene was found, thus confirming the diagnosis of *X-linked sideroblastic anemia*.

In your paper, you should discuss the following:

1. What are the clinical features of the condition/disease?
2. What is/are the underlying causes of the condition/disease? Are there specific mutations or cellular/tissue abnormalities that predispose the person to the disease?
3. How does the pathological condition differ from normal physiology of the tissue/organ?
4. What are the clinical approaches to correct the condition/disease?
5. Describe *in detail* a recent basic research advance into our understanding of the condition/disease. How has new technology allowed us to better understand the condition?

The term paper will be graded on two parts: content (70%) and formatting/referencing (30%). **Your paper should be a MAXIMUM of 5 pages long, 1.5 line spacing, typed in 12-point Times New Roman font, with page margins set at 2 cm.** Your term paper should include a 200 – 300 word abstract summarizing your work, and be structured with logical sub-headings to aid in the flow of discussion. It must be fully referenced according to the style and standards of the *Journal of Immunology*. You should consult the following web site for details on reference formatting:

<http://www.jimmunol.org/site/misc/authorinstructions.xhtml>

Do not limit yourself to summarizing information from review articles. You should read and include information from original research papers. One page of Figures or Tables is permitted, but must include legible and complete Legends to accompany the Figure. The Reference List and Figures do not count as part of the 5 page limit.

NOTE: You are not permitted to use web sites or URLs as part of your reference list. Data from web sites are not peer-reviewed and as such to not meet the burden of scientific proof to support a scientific theory.