How do the exam findings help focus your differential diagnosis?

- Open this form to summarize and possibly reprioritize your differential diagnosis. Please include any key exam findings (normal or abnormal) and how they affect your thoughts on the ranking your differential diagnosis.

- After you have finished entering your thoughts, you may proceed with the case.

(Type your responses in the online form)

Differential Diagnosis Revisited

- B cell defects
  - This remains most likely due to the difficult to visualize tonsils and absence of other major findings. Examples of B cell defects include X-linked agammaglobulinemia, X-linked hyper IgM syndrome, Ig heavy chain deficiencies, common variable immunodeficiency. Ataxia telangiectasia is less likely without abnormal exam findings. You are suspicious of an X-linked disorder due to PJ's deceased uncle.
- T cell defects (SCID, or severe combined immunodeficiency)
  - Some forms of SCID (T-/B-) are possible, perhaps a less severe form as PJ is presenting at an older age, with otherwise mostly normal development. Examples of T-/B- SCID include ADA or PNP defects, and VDJ recombination pathway defects. It is less likely he has DiGeorge syndrome without the typical exam findings. As noted above, PJ likely has a B cell defect, making a T-/B+ SCID such as cytokine receptor mutations (common gamma chain gene) less likely.
- Complement Defects
  - Less likely as he is having frequent severe infections, but certainly should be included in the initial differential diagnosis. He has not had a Neisseria infection. Nothing on exam suggests an autoimmune disease, which can be seen in some complement deficiencies.