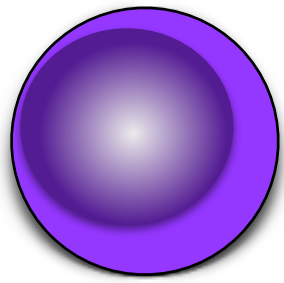


# Differential Diagnosis Revisited

- 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.



# Why is PJ sick?

- Before we proceed with ordering labs to further diagnose PJ's immunodeficiency, watch this 5 minute video to help review the function of the immune system, then proceed with the case:

[How are B cells activated?](#) (Video 3 of 5)