

Differential Diagnosis

- B cell defects
 - This is most likely, due to the frequent severe infections at multiple sites. Age of presentation can vary. Examples include X-linked agammaglobulinemia, X-linked hyper IgM syndrome, Ig heavy chain deficiencies (including ataxia telangiectasia), common variable immunodeficiency.
- T cell defects (SCID, or severe combined immunodeficiency)
 - There are numerous types, but by definition these includes (at minimum) T cell defects. Some forms of SCID also affect B cells. This is less likely as he's presenting at an older age. Examples of T-/B- SCID include ADA or PNP defects, VDJ recombination pathway defects. Examples of T-/B+ SCID include cytokine receptor mutations (common gamma chain gene) and DiGeorge syndrome.
- 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, nor evidence of an autoimmune disease, but cannot be ruled out by history alone.

Why is PJ sick?

- Before we proceed with an exam to help narrow our differential diagnosis, watch this 5 minute video to help better contextualize the exam, then proceed with the case:

[How do B cells develop?](#) (Video 2 of 5)