SUMMARY QUESTIONS – ***You should be prepared to discuss these specific questions.***

Introduce your patient to the class!

Who is she? What is her story?
(see the referral form)

What was the preliminary diagnosis and the rationale for it?
(see the referral form & NCBI's MedGen database)

What did the genetic test find and how does this relate to the preliminary diagnosis?
(see the genetic test result form & NCBI's ClinVar database)

What is the implicated/affected gene and what is its normal function?
(NCBI's Gene database should help!)

Where in the gene and gene product is the patient's genetic variant located?
(Where in the gene? In what part of the mRNA? Where in the protein? In what functional part of the protein?)

What is the molecular impact of the genetic variant on the gene product?
(What do you think the variant ended up doing to the protein structurally?)

What *do you think might be* the functional impact of the variant on the gene product and in the patient?
(What impact do you think the variant had on the function of the protein? How might this relate to the patient's symptoms?)

***Now that you're done*…..SELF-ASSESSMENT TIME!**

My initial ideas about this case:

(Why did I think this?
How confident was I?)

What did I miss?

(Why did I miss it?
How could I have thought about it differently?)

What specific content areas do I need to review?