

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

**Introduce your patient to the class!**



*Who is he? What is his 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?**