Steven and Sam met and below are the 5 scenarios/queries that we would run in the ILDB:

1. Has this ever been seen as de-novo in any case?   
   --- PM6, PS2

Does GI have a field to store de-novo status of variant?

How do we count de-novo status of cases which do not have associated parental types? How is PM6 assigned?

Sam: Currently tracked through the family number – feature request has been submitted to GI. Can infer a variant was de-novo right now. Not supported in a query-able way beyond family-numbers.

1. Whether the variant is in trans/cis with another pathogenic variant? (autosomal recessive)  
   --- BP2, PM3   
   There’s a few compound heterozygote entries in ClinVar, so if they have variants in trans and they have associated that with a disease, only 46 compounds in ClinVar (as of Sept 15th).
2. Whether there is a pathogenic variant in the same or another gene that could explain the patient’s disease? (autosomal dominant)   
   --- BP5  
   How many patients in the database have different variant in the same gene causing their disease? If large, then unlikely that this variant is responsible.
3. What is the phenotype of all cases that have the variant of interest?  
   --- PP4  
   Could you get that from ClinVar; the conditions that a variant was associated with and the assertions on them. Not very good on the phenotype side. So the quality of the answer is superior with ILDB. Will we be getting patient phenotype – do labs have this data or is it EMR? Does GI support these fields?
4. What were the phenotypes/genotypes of all the proband’s family members who were tested?  
   --- BS4, PP1, PM6, PS2  
   Phenotypes of family members unknown. GI does not collect or maintain family information at all --- not a single field in list.