|  |  |  |  |
| --- | --- | --- | --- |
| Disease | Enzyme/*GENE* | Glycogen Levels | Phenotypes |
| Fanconi-Bickel | GLUT2/*SLC4A2* |  |  |
| Von Gierke’s | G6Pase/*G6PC* |  |  |
| Tarui’s | Muscle PFK1/*PFKM* |  |  |
| GSD Type 0 | Liver GS/*GYSL* |  |  |
| Cori’s Disease | AGL/*AGL* |  |  |
| Anderson’s | Branching Enzyme/*GBE* |  |  |
| McArdle’s | Muscle Phosphorylase/*PYGM* |  |  |
| Her’s | Liver Phosphorylase/*PYGL* |  |  |
| GSD IX | Phosphorylase Kinase/  *PHKA1/2* |  |  |

# Take Home Questions

Review your thoughts

* Are there symptoms/do your predicted phenotypes match what you thought?
* Are there any you are confused by?

Pick one disease/gene from that list

* Research its prevalence in the population and its mode of inheritance (dominant/recessive, sex linked/autosomal)
* Go onto bravo.sph.umich.edu
  + Find that gene
  + Filter for loss of function variants (LoF button). These are variants predicted to **lose** function of the protein
  + Add up the frequency of heterozygous (Het column) variants for a dominant disease or the frequency of homozygosity (HomAlt column) for a recessive disease. Comment on how that relates that to the prevalence of the disease
  + Filter for missense and loss of function variants (LoF+Missense button). These are variants predicted to **change** function of the protein
  + Calculate the frequency of these variants relative to the total number of alleles

Write a brief (4-5) sentence summary of what you learned from this exercise and hand in at the beginning of next lecture.