|  |  |  |  |
| --- | --- | --- | --- |
| Disease | Enzyme/*GENE* | Glycogen Levels | Predicted Phenotypes |
| Fanconi-Bickel | GLUT2/*SLC4A2* | Higher | Hepatomegaly (Large liver)  Glucose/Galactose Intolerance  Fasting Hypoglycemia  Renal Nephropathy  Short Stature |
| Von Gierke’s | G6Pase/*G6PC* | Higher | Lactic Acidosis  Severe Hypoglycemia  Hepatomegaly  Short Stature  Hyperlipidemia |
| Tarui’s | Muscle PFK1/*PFKM* | Higher | Muscle weakness  Fatigue  Cardiomyopathy |
| GSD Type 0 | Liver GS/*GYSL* | Lower | Lethargy  Mental confusion, attention  Growth delay  Fasting hypoglycemia  Elevated fasting ketones |
| Cori’s Disease | AGL/*AGL* | Higher (more branched) | Hepatomegaly  Cardiomopathy  Hypoglycemia  Failure to thrive |
| Anderson’s | Branching Enzyme/*GBE* | Higher (less branched) | Muscle wasting  Cardiomyopathy  Hepatomegaly/Liver Failure |
| McArdle’s | Muscle Phosphorylase/*PYGM* | Higher in Muscle | Weakness after starting exercise but tolerance after 8-10 minutes (second wind)  Muscle pain/cramping  Muscle breakdown |
| Hers’ | Liver Phosphorylase/*PYGL* | Higher in Liver | Developmental delay  Hepatomegaly  Hypoglycemia  Hyperketosis  Hyperlipidemia |
| GSD IX | Phosphorylase Kinase/  *PHKA1/2* | Higher | Developmental delay  Delayed motor development  Hepatomegaly  Hypoglycemia  Hyperketosis |

Review your thoughts once I show the complete table

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

# Take Home Portion

Pick one gene/disease and refer to the completed table

* Briefly explain why each phenotype may exist, to the best of your ability.
* Search for this gene on the [ExAC database](http://exac.broadinstitute.org/" \t "_blank).  Filter to show missense and loss of function (Missense + LoF) or just loss of function (LoF) variants.  Pick one that has the highest frequency in the population and note the SNP ID (should look something like rs5400).  Write both the variant name, the molecular consequence, its frequency and how many people identified as homozygous for this variant.  Based on this information calculate how likely a person is to be heterozygous for this gene (*i.e.* 1/1000).
* Click on that variant to get more details.  Comment on the population frequencies of this particular allele.  Is it higher or lower in some populations?
* Go to the University of Michgan's [PheWeb](http://pheweb.sph.umich.edu:5000/" \t "_blank).  This database links electronic health records and various phenotypes in the [UK Biobank](https://www.ukbiobank.ac.uk/) to genetic variants.  Enter the SNP ID and summarize any phenotypes associated with this variant.
* Describe what you think the role genetics might play in understanding individual differences in glycogen metabolism.