

Author(s): Aken Desai, Michael Mathis, 2008

License: Unless otherwise noted, this material is made available under the terms of the **Creative Commons Attribution – Share Alike 3.0**

License: <http://creativecommons.org/licenses/by-sa/3.0/>

We have reviewed this material in accordance with U.S. Copyright Law **and have tried to maximize your ability to use, share, and adapt it.**

Copyright holders of content included in this material should contact open.michigan@umich.edu with any questions, corrections, or clarification regarding the use of content.

For more information about **how to cite** these materials visit <http://open.umich.edu/education/about/terms-of-use>.

Student works are presented **as is** and may be an interpretation of faculty members' lectures or assignments. These student works are **not a product of faculty members**. Faculty do not guarantee the accuracy of student work nor endorse them in any way.

Any **medical information** in this material is intended to inform and educate and is **not a tool for self-diagnosis** or a replacement for medical evaluation, advice, diagnosis or treatment by a healthcare professional. Please speak to your physician if you have questions about your medical condition.

Viewer discretion is advised: Some medical content is graphic and may not be suitable for all viewers.

Glycogen Storage Diseases

Monday, January 14, 2008

9:00 AM

33. What are the major glycogen storage diseases? How do the deficiencies of specific enzymes result in defects in glycogen?
 - a. Glucose-6-Phosphatase Deficiency
 - i. Increase in G-6-P leads to inhibition of phosphorylase, activation of synthase --> accumulation of glycogen stores
 - ii. Inability to respond to glucagon --> hypoglycemia
 - b. Branching Enzyme Deficiency
 - i. Glycogen concentration is normal
 - ii. Structure is abnormal (long chains, only one non-reducing end)
 - iii. Slower breakdown of glycogen
 - c. Muscle Phosphorylase Deficiency
 - i. Muscle can't use glycogen for energy
 - d. Liver Phosphorylase
 - i. Increase glycogen stores
 - ii. Decreased response to glucagon
34. What is the function of UDP-glucuronate and how is it synthesized?
 - a. Carries products of metabolism out to urine
 - i. Bilirubin
 - ii. Steroids
 - iii. Drugs or toxins
 - b. Synthesized from UDP-Glucose
 - i. Synthesized using 2 NAD --> 2 NADH
35. What are the various functions of UDP-glucose?
 - a. Glycolipids --> neurotransmitters
 - b. Glycogen
 - c. Galactose metabolism
 - d. Oligosaccharides and glycoproteins