Chapter 42: Genetic Disorders of Glycosylation

1. How do you define a “glycosylation” disorder? Describe the methods used today to identify a glycosylation disorder.

2. Serum transferrin has two N-glycosylation sites and each glycan consists of biantennary sugar chains with sialic acid. What kinds of glycan patterns would you expect in patients with congenital disorders of glycosylation (CDGs)?

3. What types of cells might be especially susceptible to loss of heterozygosity or spontaneous mutations that cause glycosylation disorders?

4. Explain how “gain-of-function” mutations can cause a glycosylation disorder.

5. How would you assess the genetic and environmental contributions to a glycosylation disorder?

Chapter 44: Glycosylation Changes in Cancer

1. Explain why many cancer-specific markers detected by monoclonal antibodies turn out to be directed against glycan epitopes.

2. Many cancer cell types exhibit altered branching of N-glycans, excessive expression of mucins, changes in hyaluronan production and turnover, and decreased expression and sulfation of heparan sulfate. Discuss how these changes come about and how they would affect cancer growth and metastasis.

3. Sialyl-Tn expression is a prominent feature of many carcinomas. What explains the high frequency of this expression despite the fact that the enzyme responsible for its synthesis is not always upregulated?

4. Consider the potential roles of selectins and selectin ligands in cancer progression and metastasis.

5. What are the potential ways in which alterations in glycan structure could be used advantageously for diagnosing or treating cancer?