STUDY QUESTIONS FOR GLYCOSYLATION DISORDERS AND THERAPY
JUNE 2, 2016

1. How do you define a "glycosylation" disorder? What methods are used today to identify a glycosylation disorder?
2. If you were presented with a patient thought to have a glycosylation disorder how would you determine the genetic defect—you'll need to use all your skills plus knowledge of glycans.
3. 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)? Can you identify the genetic lesion based on a glycosylation pattern?
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?