Chapter-7

Summary
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1. CCL18/PARC a chemokine is as reliable as chitotriosidase as a biomarker in patients with Gaucher disease and patients with Niemann Pick Disease.

2. Utility for monitoring patients with a wide spectrum of severe to attenuated phenotypes requires participation of a large cohort, either from different parts of the country or by enrolling in an international consortium.

3. Heparin Co- Factor II Thrombin is a robust marker for patients with MPS 1 and II where the primary storage material is dermatan sulfate. It has an advantage over the existing biomarker glycosaminoglycans in not being influenced by renal status.

4. Its role in monitoring natural disease course requires a uniform set of phenotypic expression of the disease and a longer duration of follow up in the attenuated phenotype group.

5. Probably as it can be inferred from the disease process itself, one biomarker may not answer all questions and delineate all aspects of this complex widely varying disorder.

6. We need to comprehend that rather than focusing on ‘one stop- shop biomarker’ we need to utilize a panel of biomarkers to correlate with prediction, typifaction, disease progression and response to therapy.