Metabolic brain disorders are a diverse group of hundreds of different diseases with many different etiologies. Although the commonly held incidence of metabolic disorders is thought to be 1 in 5000, studies have contended that the incidence of metabolic disorders may be higher than believed, possibly affecting 1 in 800-2500. These disorders account for roughly 1-7% of developmental delay presentations. Thus, while any individual metabolic disorder is quite rare, overall these disorders are quite common.

Diagnosing any given metabolic disorder is challenging for the neuroradiologist for a number of reasons. Many such disorders exist, and they are classified in many different ways (e.g., by affected biochemical pathway, by affected cellular organelle, etc.). Clinical signs and symptoms overlap significantly, offering little help in separating one disease from another. The imaging appearances of many disorders overlap, and vary with the stage of the disease. And while it is possible to perform every known biochemical and genetic test on patients in order to detect known metabolic disorders, the cost of such testing is prohibitive.

This talk will attempt to organize and present the more common metabolic brain disorders in a case-based format. For each case, the underlying metabolic anomaly will be reviewed and the relevant imaging characteristics described. The role of different imaging and spectroscopic techniques will be discussed. The talk will conclude with a review of the strengths and limitations of our current imaging tools, and recommendations of what to explore next, specifically, new relevant imaging and spectroscopic techniques.