

PEDIATRIC METABOLIC DISORDERS: NEUROIMAGING REVIEW
2017 CNSF Resident Review Course
Tuesday, 20 June 2017

Metabolic disorders encompass a wide spectrum of conditions that can affect the pediatric brain.

These inborn errors of metabolism cause toxic effects on the brain and body as result of their genetic mutations leading to either a lack of production of a normal biochemical substance, or the accumulation of an abnormal biochemical substrate.

The diagnosis of these disorders can be challenging as patients often present with non-specific clinical symptoms and signs.

Neuroimaging now plays an important role in the identification, localization and characterization of abnormalities in affected patients. Although MRI is sensitive to detect metabolic-related abnormalities, the imaging features can be non-specific and overlapping although their details often help narrow the clinical differential diagnosis.

There are different proposed classification systems of metabolic disorders. One of the more common systems is to use an organelle-based and non-organelle-based approach. The leukodystrophies, or dysmyelinating diseases, most often result from either lysosomal, peroxisomal, or mitochondrial dysfunction. Non-organelle-based metabolic disorders are caused by aminoaciduria, organic academia, nuclear DNA repair defect, defects in genes encoding myelin proteins, and other miscellaneous conditions.

There are many excellent published and on-line educational resources that already discuss and provide detailed imaging descriptions of pediatric metabolic disease. This lecture will discuss a basic radiologic approach to interpreting neuroimaging exams of children with metabolic disorders, focusing on several more common and important conditions with recognizable imaging features.

SUGGESTED RESOURCES:

Barkovitch, A.J., 2007. Diagnostic Imaging: Pediatric Neuroradiology. 1st ed. Salt Lake City, Utah: Amirsys.

Cheon, J-E. et al. Leukodystrophy in Children: A Pictorial Review of MR Imaging Features. Radiographics (2002); 22(3): 461-476.

Kanekar, S. and Gustas, C. Metabolic Disorders of the Brain: Part I. Seminars in Ultrasound CT and MRI (2011); 32(6): 590-614.

Kanekar, S. and Gustas, C. Metabolic Disorders of the Brain: Part II. Seminars in Ultrasound CT and MRI (2011); 32(6): 615-636.

Yoon, H.J. et al. Devastating Metabolic Brain Disorders of Newborns and Young Infants. Radiographics (2014); 34: 1257-1272.

Reichert, R. et al. Neuroimaging Findings in Patients with Mucopolysaccharidosis: What You Really Need to Know. *Radiographics* (2016); 36: 1448-1462.

U.S. National Library of Medicine National Institutes of Health - Genetics Home Reference (<https://ghr.nlm.nih.gov>).