Paediatric Neurometabolic Imaging: Over-Simplifying A Complicated Problem

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Previously, neuroimaging in pediatric patients with metabolic disease often revealed non-specific late-stage generalized atrophy in chronic situations. However, with better access to MRI, diagnosis in the early stages may be possible in infants and young children. Clinicians and radiologists are often confronted with long and bewildering lists of possible diagnosis in metabolic disease, complex classifications and pattern recognition. This presentation will attempt to simplify (and over-simplify) a complex problem we face, sometimes in specialized pediatric hospital imaging departments but rarely in most general hospitals. It will focus on typical examples of white and gray matter and mixed patterns of metabolic diseases, but will not cover CT, ultrasound, PET/SPECT imaging, or leukodystrophies (eg Canavan's) or demyelination/inflammation.

The genetic or chemical causes need to be matched with an imaging pattern recognition approach, and detect features of selective anatomical vulnerability: MRI findings may predominantly affect gray or white matter, or both. Preferential involvement of globus pallidus, striatum or thalamus in gray matter, subcortical U-fiber sparing or not, may offer clues in some conditions. The presence of other clinical data such as bony lesions, skin and ophthalmic findings is often helpful.

In addition to inherited or inborn errors of metabolism – acquired metabolic disease may be encountered especially among older children, including hypoxia, near drowning, posterior reversible encephalopathic syndrome (PRES), and drug induced/iatrogenic causes etc, which overlap with adult pattern of metabolic disease.

Newer MRI techniques that may be helpful in research and clinical imaging include diffusion-weighted MR imaging (DWI) and MR spectroscopy (MRS). Nevertheless, radiologists should be aware that these pattern recognition schema are not exclusive, are greatly simplified, with high complexity and overlap of syndromes and causes: clinical diagnosis is challenging.