Pediatric Neuroimaging in Metabolic Disorders

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Neuroimaging in pediatric metabolic disease may reveal findings with non-specific generalized enlarged ventricles and sulci in chronic situations. However, with better access to MRI, initial diagnosis in the early stages may be possible in infants and young children. Clinical radiologists are often confronted with long and bewildering lists of possible diagnosis in metabolic disease, and grapple with challenges of classification and pattern recognition. In broad terms, classification systems of genetic or chemical causes need to be matched with an imaging pattern approach, and different metabolic diseases may have different selective anatomical vulnerability to brain damage. Hence, MRI patterns may be divided into those predominantly affecting gray or white matter, or both. Within these subdivisions, preferential involvement of globus pallidus, striatum or thalamus in gray matter, subcortical U-fiber sparing or not (and whether the head size is increased) in white matter patterns, may offer clues in characteristic appearances. The presence of cysts, calcification, and cerebellar abnormalities may also be helpful for detection and differential diagnosis; aggregating other clinical data such as bony lesions, skin and ophthalmic findings is often invaluable.

For example, typical MRI findings of diffuse or bilaterally symmetrical involvement of cortical and deep gray matter include disorders of iron metabolism (eg PKAN), mitochondrial disorders (Leigh's disease, MELAS), and Wilson's disease. White matter involvement with inflammatory swelling, cystic degeneration and vacuolation may be seen in leukodystrophies (see other speaker in the same session), but predominantly gray matter diseases may also have concomitant white matter abnormalities.

Not all bilateral symmetrical abnormalities on MRI are due to inherited or inborn errors of metabolism – acquired 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.

Recent advances in new MRI techniques that may be helpful in research and clinical imaging include diffusion-weighted MR imaging (DWI) and MR spectroscopy (MRS). The extent and severity of brain damage on MRI is also being researched for prognostication. Nevertheless, even with a pattern recognition guide, radiologists should be aware that these schema are not exclusive, are greatly simplified, with high complexity and overlap of syndromes and causes. This has a tendency to make clinical diagnosis challenging and sometimes, unrewarding. This presentation will focus on typical examples of white and gray matter and mixed patterns of metabolic diseases, but not the leukodystrophies (eg Canavan's) or demyelination/inflammation. Clinical examples, mimics and pitfalls will be highlighted.