Masterclasses

M4.2

Multi-disciplinary Management of Neurometabolic Disorders 14:30 Convention Hall A

Imaging Neurometabolic Disease – Piecing Together the Puzzle Kan E

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Diagnosing neurometabolic disease is often challenging. The increasing availability of sophisticated diagnostic methods including advanced imaging techniques in magnetic resonance imaging, histopathological and molecular genetic investigational tools and laboratory tests facilitating early and accurate diagnosis have helped to elucidate underlying pathological processes.

Developing an approach using helpful clues in clinical presentation, appreciation of abnormal myelination, pattern recognition (e.g. subcortical vs deep white matter, or frontal vs parieto-occipital), and identification of key imaging features (e.g. contrast enhancement, calcification, cysts and cortical dysplasia) are important clues for diagnosis of these often under-recognised or misdiagnosed diseases.