M4.3 Multi-disciplinary Management of Neurometabolic Disorders — 14:30 Convention Hall A

Laboratory Investigations of Neurometabolic Disorders

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Neurometabolic disorders can be defined as inborn errors of metabolism (IEM) with prominent neurological manifestations such as seizure and mental retardation. Accurate and timely diagnosis of these disorders is important and yet extremely challenging. One factor contributing to the difficulty is that the signs and symptoms of neurometabolic disorders can be very non-specific and can mimic other common conditions. Also, neurometabolic disorder is very rare and is thus extremely difficult for local paediatric units to accumulate experience. Moreover, some special laboratory investigations are only available in overseas centres which create numerous logistical and financial hurdles to frontline clinical colleagues.

Neurometabolic disorders affect the myriad of human metabolic pathways, each of which may require special laboratory investigations. From a local perspective, laboratory investigations for neurometabolic disorders can be divided into three groups. The first group is the basic investigations, for example, ammonia, lactate, plasma amino acids and urine organic acids. These investigations are readily available and should be requested for patients with suspected neurometabolic disorders regardless of the actual clinical presentation. The second group is the more specific metabolic investigations available locally, for example, urine glycosaminoglycans, urine oligosaccharides, serum very-long-chain fatty acids, serum transferrin isoelectric focusing and cerebrospinal fluid neurotransmitters. These are the screening tests for mucopolysaccharidosis, lysosomal storage disorders, peroxisomal disorders, congenital disorders of glycosylation and specific IEM that affects the synthesis of dopamine and serotonin in the central nervous system. Although they are considered screening tests for certain disease groups, their sensitivity may not be high enough to exclude a disease group by a negative analysis result. The third group is the analysis of disease-causing genes by various genetic or genomic techniques. With increasing clinical application of whole exome sequencing, our reliance on genomic analysis to diagnose neurometabolic disorders has increased dramatically in the past few years and this trend is likely to continue.