Abstract Sheet

Lecture Title	Rare Diseases Inborn Metabolic Disorders in Children: Thai Experiences
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Abstract 【English】

Rare disease is any disease that affects a small percentage (less than 1 in 2,000) of the population. UN has estimated that 300 million people around the world live with one of rare diseases. There are over 7,000 rare diseases; therefore, collectively rare diseases are common. These disorders can affect infants and adults and involve multiple organs, resulting in complexity in establishing a diagnosis. Inborn errors of metabolism (IEMs) are a group of disorders due to abnormality of function of particular proteins: enzymes**,** cofactors, or transporter. Confirmatory diagnosis of IEMs often requires highly specialized biochemical and molecular genetic tests. Treatments of IEMs involve clinical genetic/metabolic specialists and other supporting specialties. This talk aims to provide general concepts of IEMs and examples of disorders of small molecules: maple syrup urine disease, tyrosinemia, and citrin deficiency and disorders of large molecules: lysosomal storage diseases including Gaucher disease, mucopolysccharidosis, and Sandhoff disease.