

## Newborn Screening, Inborn Errors of Metabolism

## AB009. Biochemical and molecular research on lysosomal storage disorders in Thai patients

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**Background:** Lysosomal storage disorders (LSDs) are a large group of inherited metabolic diseases which mostly involve the defects of acid hydrolases, resulting in impaired substrate degradation in lysosome. The progressive accumulation of undegraded metabolites causes cell and tissue dysfunction, leading to multi-systemic pathology. The LSDs have yet to be well-studied in Thailand, but more work on mutation analysis has been carried out in the last 10 years, including work from our laboratory. In collaboration with pediatricians from Siriraj Hospital, Ramathibodi Hospital and Queen Sirikit National Institute

of Child Health we are focusing on biochemical research and molecular aspects of LSDs.

**Methods:** Blood sample from more than 50 patients who clinically diagnosed to be LSDs, including Mucopolysaccharidosis (MPS), Gaucher disease, Pompe disease, and Fabry disease were characterized in term of the defective enzymes and mutational analysis.

**Results:** All samples were enzymatic assay and mutations resulting in defective enzymes were detected, 40 cases were confirmed having an MPS, 10 cases were Gaucher disease, 3 cases were Pompe disease and 4 cases were Fabry disease. Mutations were found including splicing variants, crossing-over, nonsense and missense mutations, several of them were first described.

**Conclusions:** The reported data provide information for the molecular aspects of mutations causing LSDs in Thai population that could help to plan for genetic counseling as well as useful for therapies available for most of the cases diagnosis or target treatment for specific mutations.

**Keywords:** Lysosomal storage disorders (LSDs); metabolic diseases; acid hydrolase; Thailand

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