Genotype and Phenotype Characteristics of Thai Duchenne Muscular Dystrophy (DMD) Children

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Background: Since genetic based treatment option for Duchenne muscular dystrophy (DMD) is promising and available worldwide, the need of knowing genotype and clinical characteristics is crucial. Diagnosis of DMD in Thailand is made by muscle biopsy or multiplex ligation-dependent probe amplification (MLPA). Chance of getting treatment is less in developing country as next generation sequencing is expensive and limited.

Objectives: Therefore we aim to identify clinical and molecular characteristics of DMD children.

Methods: Patients with DMD aged 0-22 years in pediatric neuromuscular clinic at Siriraj Hospital Mahidol University during 2 years were recruited. Retrospective chart review of clinical and laboratory data was done. Patients with negative MLPA were informed consent for dystrophin gene sequencing to identify the mutation.

Results: Ninety-four patients were enrolled. Mean age of onset and diagnosis were 4 and 7 years. Approximately 70% of patients have lost ambulation at an age of 9.6 ± 1.8 years. 80% were treated with glucocorticoids. Genetic testing was done in 70 patients. Molecular analysis disclosed 50% deletions, 20% nonsense, 14.3% frameshift, 5.7% duplications, 4.3% substitution, 2% splice site.

Conclusion: Early diagnosis and genetic testing is essential for upcoming genetic based therapy in DMD.