Molecular Genetics, Genomics, Mechanisms of Diseases

AB129. Prevalence of *ATP7B* mutation hotspots in Thai population

Boonyawish Kunakorn[1], Sermsiri Chitphuk[2], Manisa Boosabaratana[3], Atchara Tunteeratum[3], Thanyachai Sura[3], Donniphat Dejsuphong[4]

[1]Medical Program, [2]Research Center, [3]Department of Internal Medicine, [4]Section for Translational Medicine, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Salaya, Thailand

Background: Wilson disease (WD), an autosomal recessive disorder, is a disease which effects copper excretion from liver, leading to copper deposit in many organs like liver, central nervous system and skin. This disease is one of a common genetic disorder which carrier prevalence approximate one in a hundred people worldwide. However, there are very few studies in Thai people.

Methods: We identified the prevalence among Thai people by looking at *ATP7B* mutation hotspots in Asian people from more than 500 DNA samples obtained from staff of Electricity Generating Authority of Thailand (EGAT). We used PCR-restriction enzyme and gel electrophoresis technique to identify each mutation.

Results: According to our study, the prevalence of WD carrier is similar to many Asian countries.

Conclusions: The result will benefit WD carriers for screening and prevention of new cases and decrease the burden of the disease in Thai population.

Keywords: *ATP7B*; Wilson disease (WD); prevalence; Thailand

doi: 10.21037/atm.2017.s129

Cite this abstract as: Kunakorn B, Chitphuk S, Boosabaratana M, Tunteeratum A, Sura T, Dejsuphong D. Prevalence of *ATP7B* mutation hotspots in Thai population. Ann Transl Med 2017;5(Suppl 2):AB129. doi: 10.21037/atm.2017.s129

