

# DETECTION OF PHENYLKETONURIA BY THE NEWBORN SCREENING PROGRAM IN THAILAND

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**Abstract.** This study evaluated the newborn screening program for phenylketonuria (PKU) in Thailand from 1996 to 2006. During the study period, 5,243,841 newborns were screened, of which 16 were confirmed to have PKU. The phenylalanine levels ranged from 20.30-30.68 mg/dl (mean 25.82 mg/dl). All the patients who were diagnosed through the newborn screening program had normal growth and development after treatment except for 2 cases who were subsequently found to have a 6-pyruvoyltetrahydropterin synthase deficiency. Four additional cases of PKU diagnosed were siblings of screening detected cases who all presented with mental retardation, microcephaly, hypopigmented hair and skin and seizures in one case. Although these patients were treated with a phenylalanine restricted diet, all of them had moderate to severe psychomotor retardation. The results of this study confirm the benefit of early detection and treatment of PKU through the screening program.

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