

Thesis Title	Molecular Analysis of Human Growth Hormone (GH1) Gene in Thai Patients with Growth Hormone Deficiency.
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Date of graduation	15 September B.E. 2538 (1995)

Abstract

One defect of hereditary growth hormone deficiency (GHD) which results in short stature can be caused by growth hormone (GH1) gene deletion. 90 Thai patients (43 females and 47 males) with short stature of pretreatment height lied on or below the 3rd percentile evaluated at the Department of Pediatrics, Faculty of Medicine Siriraj Hospital were screened for GH1 gene deletions using PCR analysis. Their ages at the time of examination were range from 1 to 15 years old. The other diagnostic criteria included 1) decreased growth rate with increment growth per year less than 5 cm. , 2) delayed bone age and 3) had normal thyroid function. 14 patients (15.5%) also had family history of short stature. PCR amplification were carried out using total DNA extracted from white blood cells. Two GH1 gene flanking regions which highly homologous were amplified by using one pair of primers, followed by digestion with restriction enzyme *Sma* I , *Bgl* I and *Hae* II. The various of GH1 gene deletion will be detected after agarose gel electrophoresis. None of the deletion was found in all patients. However, 30 patients (34.4%) who had isolated growth hormone deficiency (IGHD) were selected to confirm by Southern blot hybridization using ³²P-labelled GH800 cDNA as a probe and received the same results compatible with PCR analysis. 2 affected sibs with family history

were also selected in the gene cloning for further study of other possible defects such as point mutation.