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Genetic study and TRH test for re-evaluation of congenital hypothyroidism in Taiwan

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TSHR mutations result in a wide spectrum of clinical manifestations ranging from mild to severe congenital hypothyroidism (CHT) and thyroid hypoplasia. To date, several loss of function mutations in the TSHR gene have been reported in patients with CHT. In this study, we found that the frequency of heterozygous and homozygous p.R450H (c.1349G/A and A/A) TSHR mutations was approximately 7.0% in the Taiwanese CHT patients study. The thyrotropin-releasing hormone (TRH) test is useful for differentiating central and primary hypothyroidism. We reviewed a 228 cases with eutopic thyroid glands and neonatal hyperthyrotropinemia under levothyroxine replacement for 3 years. Analyzed the clinical use of the TRH test in the re-evaluation of CHT. At the age of 3 years, 31.6% of the patients still had hypothyroidism. There was no significant difference between basal TSH level and TRH test in the diagnosis of hypothyroidism ($p = 0.23$). The negative predictive value of the basal TSH level was 100%, however, the positive predictive value was only 43.6%. When the TSH level was near the upper limit of the normal range (4.5–8.5 mIU/L), the TRH test result had a better correlation with hypothyroidism than the basal TSH level ($p = 0.03$). The threshold of the TRH test set at 60 mIU/L had the greatest area under the curve, with a negative predictive value of 95.2% and a positive predictive value of 80.2%.

Biography

Mei-Chyn Chao graduated from Kaohsiung Medical University and is an Associate Professor of Kaohsiung Medical University. She was the Chief Director of Pediatric Department and a Director, Division of Pediatric Genetics, Endocrine and Metabolism of Kaohsiung Medical University Hospital. Currently, she is a Director, Division of Pediatric Genetics, Changhua Christian Children's Hospital Taiwan.

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