Clinical Genetics

AB039. Abnormal thyroid function in Prader-Willi syndrome

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Background: Prader-Willi syndrome (PWS) is a complex genetic disorder with multisystem involvement. Multiple endocrinological abnormalities have been reported in PWS. However, there are limited published data on thyroid function in PWS patients. This study intended to describe abnormality of thyroid function in PWS patients.

Methods: Twenty-five patients with PWS confirmed by *SNRPN* methylation-specific PCR (MS-PCR) analysis and fluorescence *in situ* hybridization (FISH) analysis for 15q11-13 deletion at Srinagarind Hospital, Thailand were included in the study. Thyroid function tests including thyroid-stimulating hormone (TSH), serum total (T) T4, T3 and free (F) T4 were measured and compared to standard reference.

Results: Median age (interquartile range, IQR) of PWS diagnosis was 1.0 (0.1–4.7) years. Twenty-one (84%) patients were found to cause by 15q11-q13 deletion. Thyroid function tests were performed in 24 subjects. Abnormal thyroid function tests were found in 9 (37.5%) patients. Seven (29.2%) patients had subclinical hypothyroidism and the remaining two patients had primary and secondary hypothyroidisms. Mean thyroid hormone levels of the PWS patients with and without subclinical hypothyroidism were as follow: TSH, 6.23 (4.29–9.80) *vs.* 3.15 (0.07–8.12) mIU/L; (P<0.01); TT3, 142.12 (87.90–187.80) *vs.* 158.56 (116.20–206.10) ng/dL; TT4, 7.33 (6.57–8.14) *vs.* 7.64 (1.15–11.73) µg/dL; and FT4, 1.14 (0.95–1.34) *vs.* 1.33 (0.72–2.31) µg/dL. One patient had central hypothyroidism (TSH 0.07 mIU/L, TT3 116.2 ng/dL, FT4 1.53 ng/dL) and the other had primary hypothyroidism (TSH 8.12 mIU/L, TT3 143.5 ng/dL, FT4 0.74 ng/dL). Correlation between the prevalence of hypothyroidism and the causes of PWS was not found in this study (P=0.64).

Conclusions: The prevalence of thyroid hormone abnormalities in PWS patients was significant higher than those (1:3,314 during the year 1996–2001) in Thai general population. Apart from central hypothyroidism, primary and subclinical hypothyroidisms are also evident in PWS patients. Thyroid function tests are highly recommended in all PWS patients.

Keywords: Prader-Willi syndrome (PWS); thyroid hormone; hypothyroidism

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