PP-47

Delayed Diagnoses of Prader Willi Syndrome in a 19-Year-Old

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INTRODUCTION

Prader-Willi syndrome (PWS) is a complex genetic disorder caused by lack of expression of genes on the paternally inherited chromosome 15q11.2-q13. Hypothalamic dysfunction has been implicated in many manifestations of this syndrome, including multiple endocrine abnormalities. These include growth hormone deficiency, central adrenal insufficiency, hypogonadism, hypothyroidism, and complications of obesity such as type 2 diabetes mellitus

CASE

We report a 19-year-old lady who was initially referred to the Endocrine Unit for primary amenorrhoea by a gynaecology clinic. She has one younger brother with normal pubertal development. Further history revealed that she had developmental delay and learning difficulty. She was also noted to have polyphagia and gradual weight gain since 3 years of age. Findings on physical examination were body mass index of 44 kg/m^2 and height of 150 cm (below third centile), compared to midparental height of 158 cm. Pubic hair and breast development were Tanner stage II. She had no features of Cushing's syndrome.

Basal hormonal evaluation revealed follicle stimulating hormone (FSH) of 11 IU/L, luteinising hormone (LH) of 2.6 IU/L and low estrogen level of 19.4 pmol/L, indicative of secondary hypogonadism. Other hormone results were as follows: cortisol 158.6 nmol/L, insulin-like growth factor -1 (IGF-1) 99 µg/L (normal value 284 to 713), prolactin 10.34 ng/L, T4 14.6 pmol/L and TSH 1.40 mIU/L. Her 17-OH progesterone level was not elevated. Short Synacthen test showed adequate adrenal response. Magnetic resonance imaging of the brain reported a normal pituitary gland. Pelvic ultrasonography showed an anteverted uterus 5.6 cm x 2.5 cm with normal ovaries. Genetic study by DNA methylation testing confirmed the clinical diagnosis of PWS. She is currently managed by a multidisciplinary team consisting of a gynaecologist for cyclic oral contraceptive pills, occupational sports medicine for her weight loss program and dietary plans, and endocrinology for regular screening of endocrine manifestations of PWS.

CONCLUSION

PWS is a genetic syndrome in which early diagnosis and careful attention to detail regarding all the potential endocrine and behavioural manifestations can lead to a significant improvement in health and developmental outcomes. The importance of the roles of the providers caring for patients with PWS cannot be overstated.

PP-48

Spectrum of Thyroid Disorder in Amiodarone-Induced Thyroid Dysfunction – A Case Report

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INTRODUCTION

Amiodarone is the most commonly used anti-arrhythmic drug worldwide. It can lead to both hypothyroidism (amiodarone-induced hypothyroidism, AIH) and less commonly, hyperthyroidism (amiodarone-induced thyrotoxicosis, AIT). While AIH is more common in iodine-sufficient populations, AIT is seen more frequently in iodine-deficient areas.

CASE

A 53-year-old man had hyperthyroidism and subsequently had hypothyroidism following amiodarone treatment. He had underlying non-ischaemic cardiomyopathy with recurrent ventricular tachycardia requiring ICD insertion in 2008. He was dependent on amiodarone for a few years in view of recurrent ventricular tachycardia, despite having two cardiac ablations. Initially, he developed hyperthyroidism following amiodarone treatment and was started on Carbimazole. Subsequently, he became hypothyroid. Clinically, he was euthyroid with no palpable goiter. Anti-thyroid stimulating hormone (TSH) receptor antibody was undetectable. Ultrasonography of the thyroid revealed normal thyroid gland with reduced vascularity. Thyroid uptake scan showed a hypofunctioning thyroid gland. Based on his series of thyroid function tests, he had AIT type 2 and subsequently developed overt AIH. We noticed, however, that the patient had elevated free thyroxine (FT4) with high TSH and was asymptomatic for 3 months duration while still on amiodarone. Upon review one year later, he currently has subclinical hypothyroidism.

CONCLUSION

This case illustrates the spectrum of thyroid function abnormalities in patients on amiodarone. In the absence of hypothyroid symptoms or thyroid antibodies, patients with moderately elevated serum TSH (<20 mIU/L) but high-normal or raised serum FT4 may reflect amiodarone-induced alteration in thyroid function parameters or subclinical hypothyroidism. Therefore, thyroid function test results while on amiodarone should be scrutinized before definitive treatment is instituted. A baseline evaluation will help identify patients who may be predisposed to developing thyroid dysfunction while on amiodarone.