



# Central precocious puberty in a boy with Prader-Willi syndrome during growth hormone replacement therapy

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## BACKGROUND

Prader-Willi syndrome is a genetic disorder characterized by obesity, short stature, hypotonia and hypogonadism. Delayed or incomplete puberty are usually found in PWS, whereas central precocious puberty is very rare.

## OBJECTIVES

This study aimed to report the case of a boy with PWS who was diagnosed with precocious puberty during growth hormone replacement therapy.

## METHODS

We retrospectively analyzed the genetics, clinical characteristics and laboratory findings of the boy.

## RESULTS

By the age of 4, the boy had mental retardation, epilepsy, characteristic face features, short stature with feeding difficulty in Neonate, and many clinical criteria of PWS diagnosis, which was confirmed by DNA methylation test (MS-MLPA). Therapy with recombinant human growth hormone (rhGH) replacement (0.1 IU/kg/day) was started. 2 years later, he performed increased testicular volume and growth velocity, high testosterone levels and advanced bone age. An ACTH test yielded a normal response and A GnRH test showed premature activation of the hypothalamic-pituitary-gonadal axis with pubertal gonadotropin and testosterone levels (gonadotropin-releasing hormone stimulated LH peak 20.51 IU/L, testosterone 3.32 nmol/L). Magnetic resonance imaging (MRI) of hypothalamic-pituitary region was normal.

## CONCLUSIONS

In PWS, puberty is usually delayed and secondary sexual characteristics are almost always incomplete. True precocious puberty is very rare in PWS and only a few cases have been reported. Our patient fulfilled all diagnostic criteria for CPP. The rare manifestations of CPP in patients with PWS has been attributed to brain lesions. We hypothesize that our patient's precocious puberty resulted from abnormal brain discharge caused by epilepsy. Next step, we will treat the patient with gonadotropin-releasing hormone analog (GnRHa) and follow up his pubertal development.

## REFERENCE

Ludwig NG, et al. Arch Endocrinol Metab 2016;60(6):596  
Lee HS, Hwang JS. J Pediatr Endocr Met 2013;26(11-12):1201

## NOTHING to DISCLOSE

WL, ZZ, JN, XL, LX, FL

