Central Precocious Puberty Presented due to Late Started Treatment for Familial Testotoxicosis

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Background: Peripheral precocious puberty (GnRH independed): precocious development of secondary sexual characteristics may also be caused by mechanisms that do not involve activation of pulsatile GnRH secretion. Familial male-limited precocious puberty, also known testotoxicosis is a rare dominant form of gonadotropin independent precocity caused by constitutively activating mutations of the human LH choriogonadotropin receptor (LHCGR). If do not treat with appropriate drugs such as aromatase inhibitors, anti androgens, ketoconazole spironolacton etc. High testosterone levels can cause to central precocious puberty. We presented here a boy have testotoxicosis but late started treatment and therefore induced central precocity.

Case: A boy 8.5 year old admitted for pubic and axillary hair started after 4 year. He have two healthy sisters and no consanguinity between mother and father's. In physical examination height 141 cm (O97per, C2.5 SD), weight 35 kg (O97p) BMI 17.6 kg/m2 (50–75per), pubic and axillary hair tanner stage IV, penil length 12 cm(O97per, OC2.5 SD) testes volume bilaterally 10 ml was found. Laboratory analysis LH! 0.1 mIU/ml, FSH 0.2 mIU/ml, testosterone 650 ng/dl, alpha-fetoprotein 0.9 IU/ml, HCG 0.1 IU/ml, DHEA-SO4 75 mg/dl, ACTH 30 U/l, bone age 13 year, bone age/chronological age: 1.73. Geneticalley evaluation showed LHCGR gene p.T577I (c.1730COT) heterozygot mutation. After a month of treatment initiation with bicalutamide 50 mg/day and letrozole 2.5 mg/day LH 2.2 FSH 6.8 testosterone 580 ng/dl was found. Triptorelin 7.5 mg/monthly added to treatment. After 6 month of treatment initiation LH: 0.48 mIU/ml, FSH: 0.4 mIU/ml testosterone 115 ng/dl. Bone age13.5 year and advancement stopped.

Conclusion: Testotoxicosis is a rare dominant form of gonadotropin independent precocity. Bicalutamide and letrozole can be used safety and effectively. It should be noted if treatment for peripheral precocious puberty do not started early high testosterone levels can lead to central precocity.

ESPE 2015 1160--P3

Puberty

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DOI: 10.3252/pso.eu.54espe.2015

Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology