Precocious puberty defines itself by the appearance of secondary sexual characters before the age of 9 years old for boys. Congenital adrenal hyperplasia (CAH) is an autosomal recessive disease resulting from mutation of genes encoding enzymes for hormone production (corticosteroid, aldosterone, and androgens). It is the most frequent cause of peripheral precocious puberty (PPP).

We bring back the case of a precocious pseudo puberty (PPP) in a sexual having evolved towards a central precocious puberty (CPP) revealed by an advance of secondary bone age because of a delay of diagnosis of CAH by 21 hydroxylase block enzyme.

Observation

It’s about a 7 years old boy, eldest in a family of 4 children, from a full-term pregnancy of a non-consanguineous marriage, accepted for reappraisal of a PPP linked to a bilateral testicular increase of measuring volume between 6-8ml a Tanner stage 2G23. Birth weight 3900g / Size, Aggar unspecified, vaccination well done without notion of loss of salts. His brother aged 4 is followed for CAH revealed by PPP tightly controlled under hormonal treatment, the remaining siblings are healthy.

The diagnosis of PPP was made at the age of 4 months and 8 years with clinical signs of hyper androgenesis appeared since the age of 3 such as acne located on face and bust, a husky voice, a wingspan android: broad shoulders, good muscle development.

Appearance of external genitalia: P3G1 testicular volume of 2.5ml right 3ml left, no axillary hair, no erection neither ejaculation.

Stature advance (+ 3 SDS corresponding to age of 11 years) and weight with BMI about 24kg / m2 as well as bone age of thirteen-year-old in spite of a suppressive treatment by Dexamethasone 0,25mg / j, Acetate of testosterone: 1,3nmol/l (0.1

Results

Hormonal check up at the diagnosis of CAH: ACTH: 269 pg/ml (N 7.2 – 63.3)

Cortisol 8h: 103,5nmol/l (171; 536) 17OHP: 21.09 ng/ml (0.31. 2.111) composit 5: 1,3nmol/l (1-4-5)

Testo: 14.67 nmol/l (0.1- 1.12) LH: 0.1μ UI/l (0.2 - 1.4) FSH: 0.11μUI/l (0.2-3.8)

motivated by the increase of testicules size: 1/sctral ultrasound ; each measuring 30nn homogeneous appearance e (anterior size: right 17X7mm, left18.6X7 mm)

2/gonadal check up: FSH: 2,34μIU/ml (nl<1) LH: 1.94μIU/ml (nl<1) Testo: 1.72nmol/l (0,1-1,12)

Pituitary-hypothalamic MRI was not requested. Gonadotrophines analogues (GnRHa) treatment was initiated six months after the time of diagnosis of PPC. 3.75mg 1 injection/month

Test of GnRHa 100ug after 3months of TRT : LH: 0,51μl/ml FSH: 0,33μl/ml Testo : 20 ng/dl

Discussion

although fetal adrenal steroidogenesis is established in early gestation, a boy with CAH rarely has signs of virilization at birth, despite plasma testosterone concentration that are often within the normal adult male range. Studies have shown that patients with CAH and CPP had significant increase in growth and accelerated bone age during the first two years of life which is explained by the effects of androgens during early infancy growth through both direct and indirect actions of testosterone:

Direct action by stimulation of sulfate of proteoglycans in chondrocytes. Testosterone acts synergistically with growth hormone (GH), the indirect effect is the result of increasing GH secretion.

Gonadotrophine analogues (GnRHa) treatment of CAH complicated by CPP was first described in 1985 by Pescovitz et al.(1)

Others study reports on the effectiveness of GnRHa therapy in arresting puberty and in improving final height in late treated cases of CAH (2)

Our patient required 30 mg / day hydrocortisone lifelong treatment, while GnRHa will be stopped at a chronological age of 13 years.

The predicted final height (PH) was calculated in our patient according to tables of Bofnig & Schwarz (table 1) because: child with CAH has a growth puberal peak lower than the one of a normal child making most application of curves Bayley & Pinneau done on healthy children.According to the latest consensus endocrine society indicates a treatment with GH when CAH if the PH is lower than -2.25 SDS (4).The GH combination with GnRHa was ranked among the treatments improving linear growth. We did not use GH therapy in our case because PH: 169,5cm corresponding to -1SDS and due to the patient’s stature advance whereas in our country the treatment by GH is only recommended for patients with GH deficiency in our country

Conclusion

CPP can be observed at the time of diagnosis in patients belatedly diagnosed with or undertreated for CAH, particularly in countries where a routine neonatal screening programme for this condition is lacking. GnRHa treatment appears to improve linear growth and final height, bringing it closer to the one expected from genetic potentials. 

Knowledgements:


Tableau 1: Bofnig & Schwarz table predicted height

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