Introduction

DAX-1 is Dosage-sensitive sex reversal - Adrenal hypoplasia congenita critical region on the X chromosome 1, caused by mutation of NR0B1.

It commonly presents X-Linked adrenal hypoplasia congenital, hypogonadotropic hypogonadism and infertility.

However, we observe two patients whose testosterone elevated in their early infancy, which tended to be misdiagnosed as congenital adrenal hyperplasia.

Methods

We observe two cases of DAX-1 deficiency with elevated testosterone during their early infancy and summarize DAX-1’s distinct features between congenital adrenal hyperplasia.

Case 1

For case 2, jaundice is his only symptom. His maximal ACTH is up to 2000pg/ml, minimal cortisol is 0.71nmol/L, maximal testosterone is 181ng/ml, serum sodium is 121mmol/L, serum potassium is 9.1 mmol/L, and 17-hydroxyprogesterone is 3.01nmol/L (Table 1). Adrenal ultrasound shows hypoechoic nodule in adrenal region which is similar to adrenal gland structure, the size is smaller than normal. Gene test finds NR0B1 (NM_000475.4) Intron1: c.1169-1G>T.

Results

After the replacement of hydrocortisone and fludrocortisone, their levels of testosterone come down to normal range in 4 months, 6 months respectively.

Bone age of Case 1 is 2.5 years when his chronological age is 3 years and 4 months.

Both of them get smaller testes which is consistent with his disease.

Conclusions

Although DAX-1 commonly occurs hypogonadotropic hypogonadism at puberty or in early adult, gonad and sex hormones could be normal or even temporal elevated in early time.

With this report we can summarize DAX-1’s distinct features as follows: 1. DAX-1 is absent from elevated 17-Hydroxyprogesterone in general; 2. DAX-1 occurs delayed bone age which is opposite to congenital adrenal hyperplasia; 3. Adrenal ultrasound has certain value for recognizing DAX-1 and congenital adrenal hyperplasia; 4. Genetic test is an optimal way to distinguish DAX-1 deficiency from other diseases.

References


Table 1: Hypogonadotropic hypogonadism.

<table>
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<tr>
<th>Case</th>
<th>ACTH</th>
<th>Cortisol</th>
<th>Testosterone</th>
<th>Sodium</th>
<th>Potassium</th>
<th>17-Hydroxyprogesterone</th>
<th>Bone Age</th>
<th>Chronological Age</th>
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</table>

For the Table, ACTH: adrenocorticotropic hormone, Cortisol: cortisol, Testosterone: testosterone, Sodium: serum sodium, Potassium: serum potassium, 17-Hydroxyprogesterone: 17-hydroxyprogesterone. Bone age and chronological age are in years.