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ELEVEN YEARS OF LETROZOLE TREATMENT IN A CHILD WITH 11-β HYDROXYLASE DEFICIENCY: EFFECT ON BONE AGE AND HEIGHT PROGNOSIS

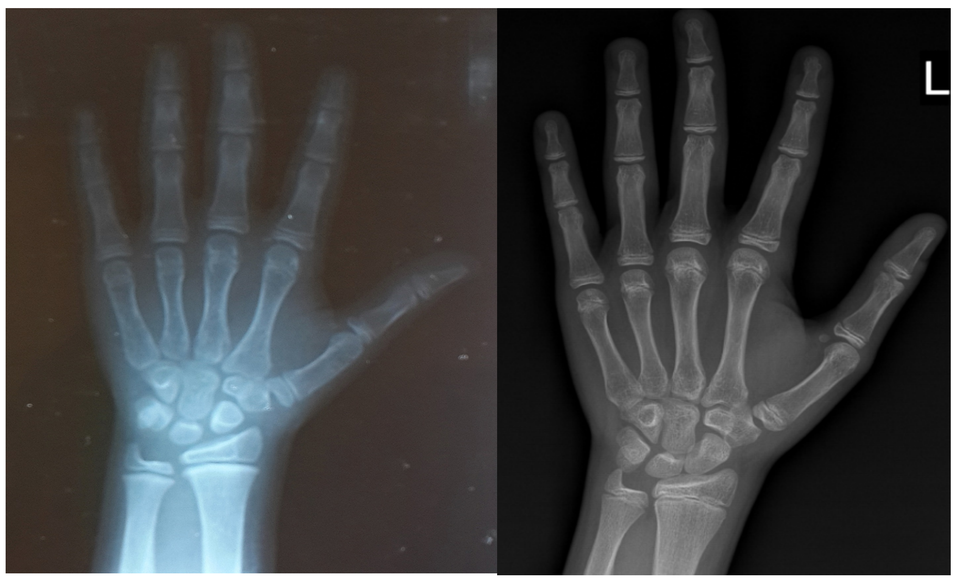
Abdullah Bereket, Zeynep Atay, Tülay Güran, Belma Haliloğlu, Saygın Abalı, Serpil Baş, Serap Turan

Marmara University, Medical Faculty, Pediatric Endocrinology; Istanbul, Turkey

Background

- **Short stature is an important long-term sequela in children with congenital adrenal hyperplasia (CAH).**
- **Bone age advancement due to excess adrenal androgens resulting in early epiphyseal fusion is the main reason for short stature.**
- **Boys with CAH who are not salt-wasters are especially at risk for short stature because of significant delay in diagnosis and treatment.**
- **Aromatase inhibitors (AI) are being used in clinical trials in children related to peripheral precocious puberty, and idiopathic short stature to improve height prognosis.**
- **Here we report a child with 11-β hydoxylase deficiency presenting at the age of $2^{11/12}$ years with bone age of 13 years in whom the use of letrozole, a potent aromatase inhbitor, for 11 years have proved benefit with a gain of 34,9 cm in predicted adult height, at the expense of a decrease in bone mineral density (DEXA) Z score from -0.3 to -0.9 during treatment**

Figure.1. Bone age at presentation (at age 2 $^{11/12}$ year and and at 15 $^{5/12}$ year



Case

- **2 $^{11/12}$ year old at presentation**
- **penile enlargement and pubic hair development.**
- **Well developed muscular boy with oily skin and acnea**
- **Weight: 22 kg (>97p), height: 110.1cm (>97p)**
- **Blood pressure: 140/90 mmHg**
- **Testicular volumes: 6 cc, stretched penile length: 9 cm**
- **Tanner stage II pubic hair.**
- **Bone age: 13 years; Predicted adult height:129.5 cm**
- **Genetic analysis: g.4643_4644insGA mutation in CYP11β1**

Figure.2. Vertebral X-ray and MRI of the patient

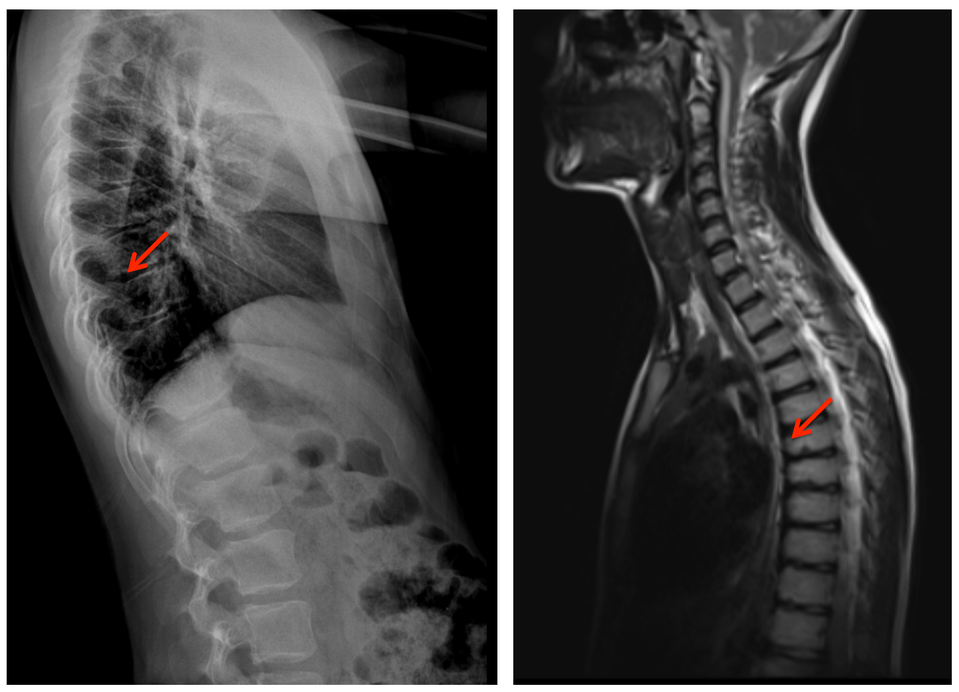
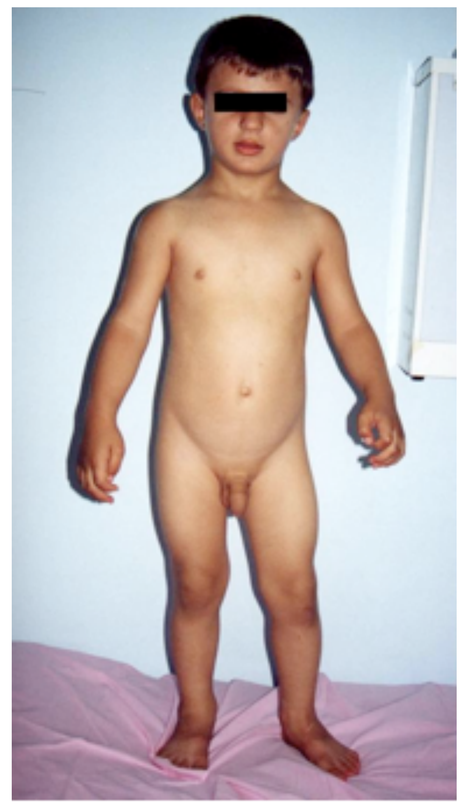


Table 1. Hormonal profile of the patient at presentation

	Patient's values	Reference values
Na	141 mEq/L	138-147 mEq/L
K	3,7 mEq/L	3,5-5,3 mEq/L
LH	< 0,1 mIU/ml	0,02-0,3 mIU/ml
FSH	0,135 mIU/ml	0,26-3,0 mIU/ml
Cortisol	1,4 µg/dl	5-25 µg/dl
T Testosterone	2,89 ng/ml	0,03-0,32 ng/ml
ACTH	1250 pg/ml	0-46 pg/ml
17-OH Progesterone	19,02 ng/ml	0,03-0,9 ng/ml
Plasma Renin Activity	0,1ng/ml/h	0,20-3,40 ng/ml/h
11-Deoxycortisol	140 ng/ml	0-8 ng/ml

Picture of the patient at presentation (Age 3 years)



Conclusions

- **After 11 years of letrozole therapy, at the age of 15 y $^{5/12}$ mo his near-final height is157,7cm, bone age is 14 y $^{9/12}$ mo and predicted adult height is 164,4cm. The expense to this dramatic increase in height prognosis is a decrease in bone mineral density (DEXA) Z score from -0.3 to -0.9 during treatment and the development of Scmorl nodules in thoracic vertabrae.**
- **As far as we know, this is the longest duration of letrozole use to improve height potential and is highly encouraging regarding the use of aromatase inhibitors early in the disease course in selected patients with advanced bone-age and severely impaired height prognosis.**

Figure.3. Growth chart of the patient