

Normal growth in aromatase excess syndrome by pharmacological inhibition of aromatase activity

Beate Deubzer, Gerhard Binder

Pediatric Endocrinology and Diabetology,

University Children's Hospital, Tübingen, Germany



UNIVERSITÄTS
KLINIKUM
TÜBINGEN

Introduction

In aromatase excess syndrome the turnover from androgens to estrogens is constitutively increased. Males suffer from hyperestrogenism (particularly gynecomastia), hypogonadism and short stature due to early epiphyseal closure.

We compared the statural growth of two affected first degree cousins who received and did not receive treatment with an aromatase inhibitor during puberty. Both cousins have a heterozygote deletion within the regulatory part of the CYP19A1 gene locus of the aromatase gene.

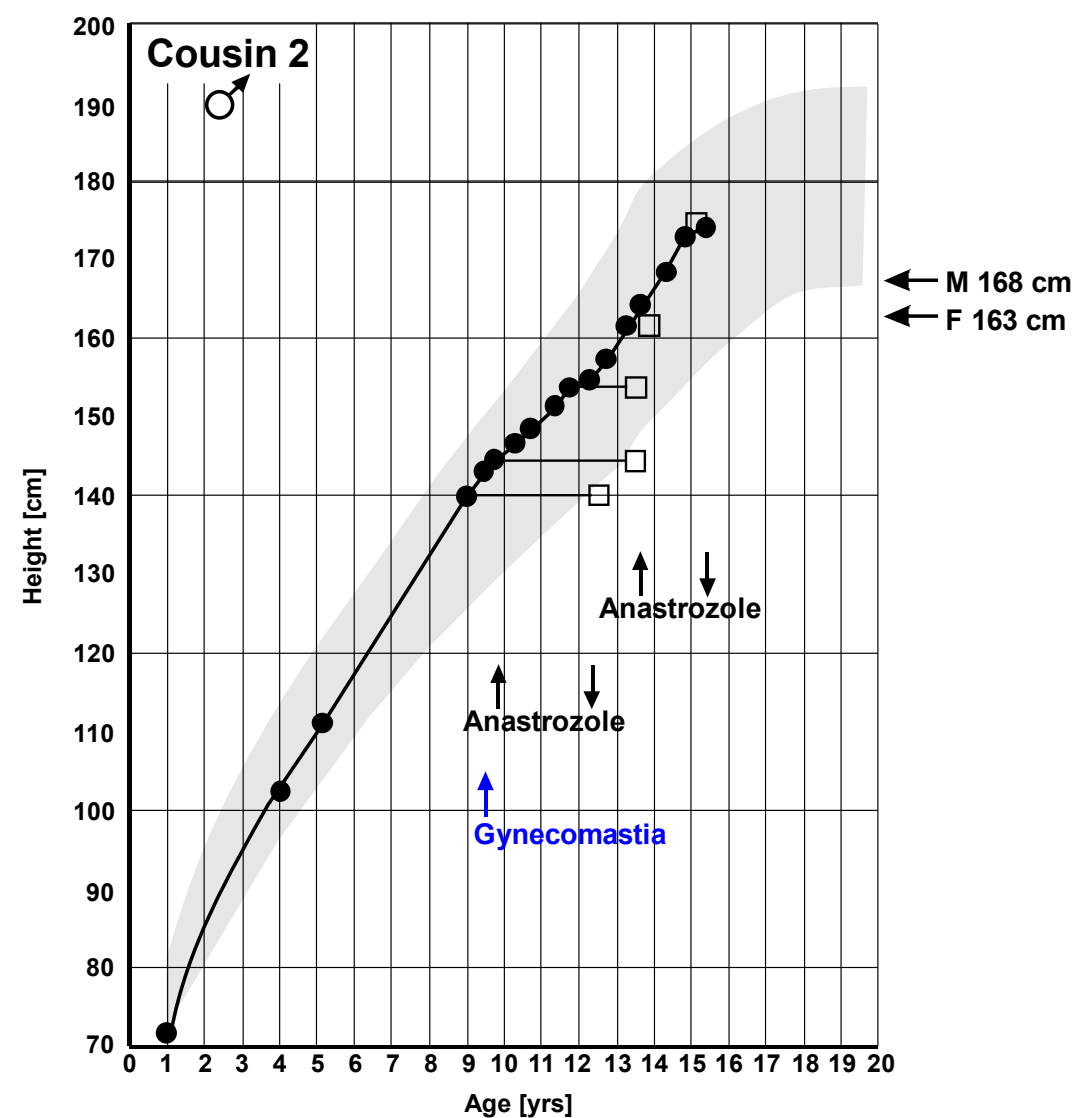
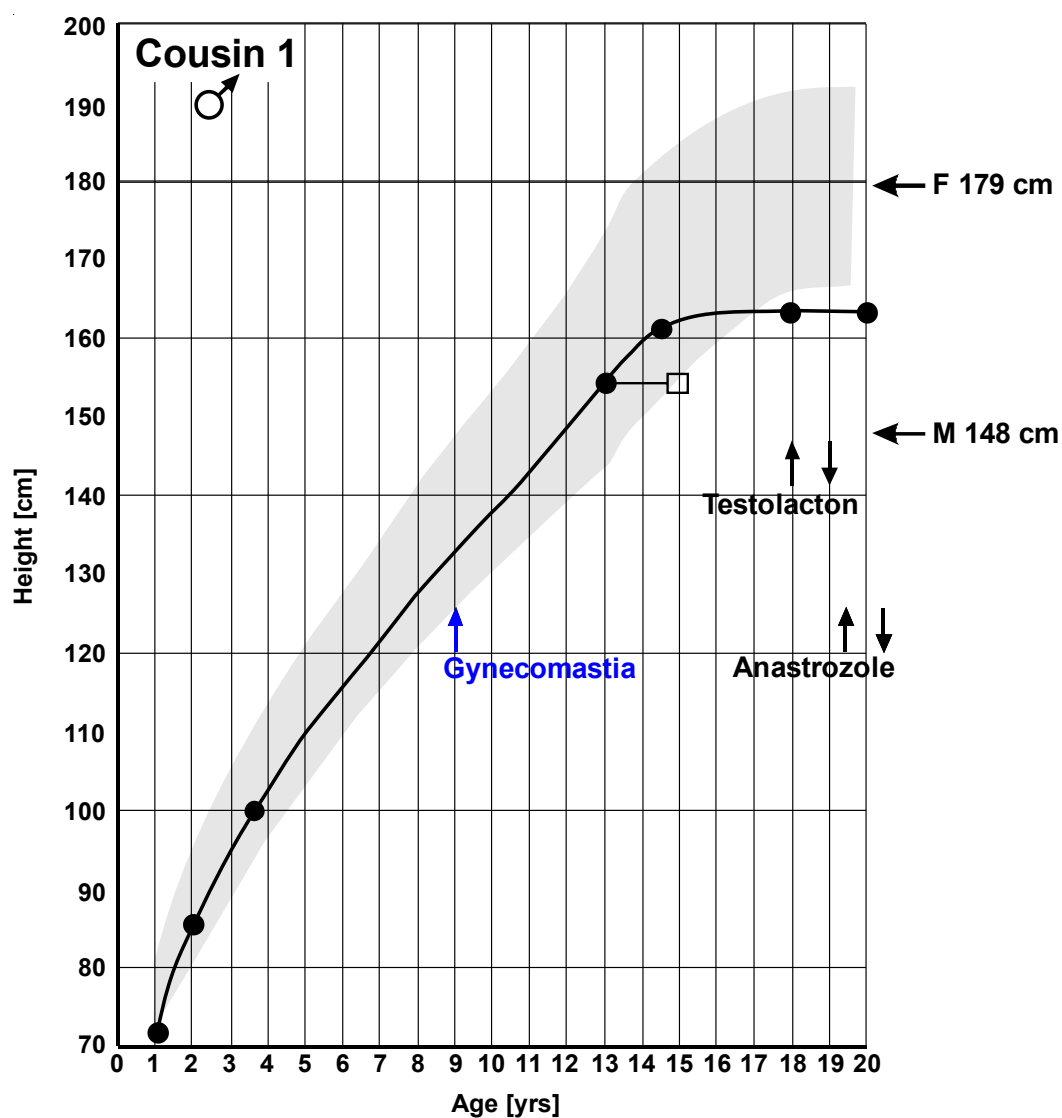
Conclusion

Treatment with aromatase inhibitors can effectively prevent short stature in aromatase excess syndrome.

Case Report

Cousin 1 was diagnosed at the age of 18 years (index patient of the family). He suffered from incomplete virilisation and had had gynecomastia and advanced bone age during puberty. His testosterone to estrogen level was pathologically low and his LH and FSH levels were low normal. By treatment with an aromatase inhibitor for promotion of virilisation his adult height of 162 cm (-2.3 SDS) could not be improved (testolacton 5 mg/kg/d for 1 year, then anastrozole 1 mg/d for 1 year). His mother [M] was of short stature (148 cm) due to untreated aromatase excess syndrome.

Cousin 2 presented at an age of 9 years with beginning signs of hyperestrogenism (breast development) and was immediately diagnosed due to his relationship with *cousin 1*. Treatment with anastrozole 1 mg orally was given and he already reached a normal adult height of 175 cm at the age of 15 years (0.75 SDS). His father [F] was of short stature (162.7 cm) due to untreated aromatase excess syndrome.



	Height at 4 years	Age at start of treatment	Height at start of treatment	Bone age at start of treatment	Medication	Age at end of treatment	Height at end of treatment
Cousin 1	101 cm (-0.9 SDS)	18 years	162 cm (-2.3 SDS)	19 years	testolacton 5 mg/d; anastrozole 1 mg/d	20 years	162 cm (-2.3 SDS)
Cousin 2	102 cm (-0.7 SDS)	9 years	140 cm (+0.8 SDS)	12 years	anastrozole 1 mg/d	15.5 years	175 cm (+0.4 SDS)