Aim

We report the clinical phenotype and hormonal studies of a 46,XY aromatase deficient boy.

Results

Discussion

Normal pubertal development has been assumed in adult men with aromatase deficiency. Estrogen restrain on gonadotropin secretion has been demonstrated in animal and human models of estrogen deficiency acting from the early phases of puberty in males. Interestingly, our patient presented with early and accelerated puberty and apparently normal pituitary gonadal function. This human model of nature suggests that aromatase activity at the pituitary gonadal axis and spermatogenesis.

Most 46XY affected patients presented with normal external genitalia and the condition often remains undiagnosed until late puberty. In adult affected males, slight increment of basal serum FSH (and in some cases also basal serum LH) despite normal or even high serum testosterone and normal inhibit B levels were reported. These findings support the important role of estrogens either from peripheral origin or locally synthesized on the regulation of gonadotropin secretion.

There is scarce information about clinical and biochemical findings in affected boys during prepubertal and pubertal years, since only 2 patients, younger than 4 yr of age, without a long-term follow-up, have been reported.

Clinical Material

The patient is the older brother of a 46,XX affected sister (2). Maternal virilization was present during both pregnancies. Coding exons of the CYP19 gene and the flanking intronic regions were PCR amplified from patient, parents and control cDNA.

Schematic representation of the genomic structure of the CYP19A1 gene showing the C-to-T transition in exon 5

Molecular studies

Sequence alignment: mutated arginine at position 192 is highly conserved in homologues of P450arom from many species analyzed.

References