The lack of *MKRN3* gene mutations in patients with idiopathic sporadic GnRH-dependent precocious puberty

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Introduction

Central precocious puberty (CPP) results from activation of the hypothalamic-pituitary-gonadal axis before the age of 8 years in girls and 9 years in boys. The molecular basis of the maturation of this axis is still poorly understood. The *MRKN3* gene located in the Prader-Willi syndrome critical region (chromosome 15q11-q13), inhibit factors stimulating pulsative. GnRH secretion. In 2013 inactivating mutations in the *MRKN3* gene were discovered to cause some of the cases of familial precocious puberty. Subsequently, there have been few reports of apparently *de novo* mutations causing sporadic central precocious puberty.

Study group

Blood samples were collected from 25 unrelated patients (24 girls and 1 boy), from two university medical centers.
All patients were clinically diagnosed with precocious puberty of central origin.
Clinical signs:
Girls
- enlargements of mammal glands
- estrogenisation of external genitalia

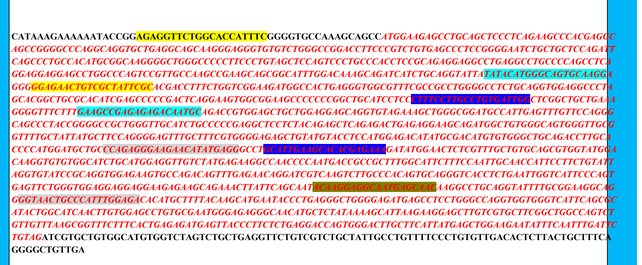
Boys:
- enlargements of testicles

Both:
- acceleration of height velocity
- acceleration of bone maturation
- pubertal result of GnRHa testing

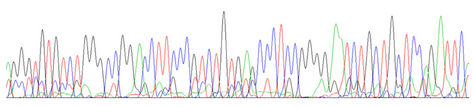
The objective

of the study was to investigate mutations in *MKRN3* gene in patients with apparently sporadic idiopathic CPP.

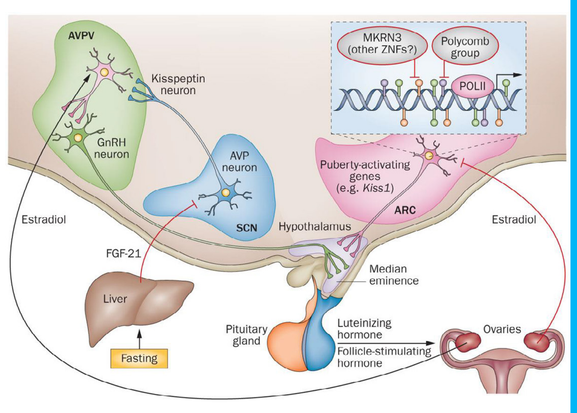
Measurements



DNA was isolated from lymphocytes using standard procedures. The whole coding region of the *MRKN3* gene was divided between five sets of primers. Each fragment was amplified with PCR, routinely cleaned and then sequenced with the classical Sanger's method.



The repressive control of puberty



Ojeda, S. R. & Lomniczi, A. (2013)
Unravelling the mystery of puberty
Nat. Rev. Endocrinol. doi:10.1038/nrendo.2013.233

Results

No pathogenic variants of the *MRKN3* gene were found among the studied group.

Conclusion

Although deficiency of MKRN3 causes central precocious puberty in humans, mutations in *MRKN3* gene are a very rare genetic cause of isolated central precocious puberty.

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