GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

Mutations in *NR5A1* and *PIN1* associated with idiopathic hypogonadotropic hypogonadism

S.C. Hu[1,2], J. Ye[1], A.K. Fathi[1], X. Fu[1], S. Huang[1], Q. Ning[1] and X.P. Luo[1]

[1]Department of Pediatrics, Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology, Wuhan, Hubei Province, China
[2]Department of Nephrology and Hematology, Qingdao Women and Children Hospital, Qingdao, Shandong Province, China

Corresponding author: X.P. Luo
E-mail: xpluo@tjh.tjmu.edu.cn

Genet. Mol. Res. 11 (4): 4575-4584 (2012)
Received February 27, 2012
Accepted June 22, 2012
Published October 9, 2012
DOI http://dx.doi.org/10.4238/2012.October.9.6

ABSTRACT. We tested the hypothesis that mutations in *NR5A1* and *PIN1* cause disorders in gonadotropin-gonadal system development and function, throught direct DNA sequencing of the coding sequence and splice-sites of *NR5A1* and *PIN1* in 50 subjects with sporadic idiopathic hypogonadotropic hypogonadism. These patients were recruited from the Pediatrics section of Tongji Hospital, Tongji Medical College, in Wuhan, China. None of the affected subjects had clinical signs of adrenal insufficiency. The *NR5A1* and *PIN1* mutations were found in 7 of the 50 cases. These 7 individuals presented severely low serum concentrations of testosterone or of estradiol and gonadotropin. Adrenal insufficiency was not diagnosed in any of these patients. Consequently, *NR5A1* and *PIN1* mutations should be considered in idiopathic hypogonadotropic hypogonadism patients with normal karyotypes and without adrenal insufficiency.

Key words: Hypogonadotropic hypogonadism; Gene mutations; *NR5A1*; *PIN1*

