Indian Journal of Basic and Applied Medical Research; June 2014: Vol.-3, Issue- 3, P. 358-367

**Original article:**

**Presence of the ASN680SER Polymorphism in women with Primary Amenorrhea from South India**
**Merin Thomas, Sweta Srivastava, Rema Devi**

Division of Human Genetics, Department of Anatomy, College building first floor, St. John’s Medical College, Bangalore -34. [080- 49466104; 9741123462] , India
C**orresponding author**: Merin Thomas

Date of submission: 15 May 2014 ; Date of Publication: 25 June 2014

**ABSTRACT:**

**Introduction:** The hormones that play a role in the menstrual cycle are FSH, LH, estrogen and progesterone. For a hormone to function normally, the structural and functional integrity of its receptor is a pre-requisite. The Asn680Ser polymorphism in the FSHR gene has been the more studied polymorphism as it seems to play a role in the receptor function. The prevalence of the FSHR gene polymorphism in patients with primary amenorrhea and its possible significance has not yet been explored in the South Indian population

**Methods:** 139 subjects were taken for this study of which 92 of them had primary amenorrhea and 47 [controls] of them had regular menstrual cycles. Cytogenetic analysis was done by routine karyotyping methods to rule out the presence of a chromosomal abnormality. This was followed by DNA extraction using organic solvents phenol and chloroform. The extracted DNA was quantified and then amplified using primers designed for this study. The amplified PCR product was then purified and sequenced.

**Observation & Results:** 26.1% of the patients were found to have a homozygous mutant status while 12.8% of the controls were also found to have a homozygous mutant status.

**Conclusion:** This is the first study to report the prevalence of the FSHR gene polymorphism in women with primary amenorrhea from the South Indian population. 26.1% of the patient group and 12.8% of the control group were found to have the polymorphism. While the FSHR gene polymorphism cannot be attributed as the sole cause of primary amenorrhea; other genes of the menstrual cycle need to be screened to get an answer for the same.

**Keywords:** Asn680Ser, FSHR gene, Primary Amenorrhea