Abstract

Title: Role of Mediator Complex Subunit 12 (MED12) Gene Exon 2 Variations in the Etiology of Uterine Fibroids

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Background:

Uterine leiomyomas also known as fibroids are one of the most frequent solid pelvic tumors in female genital tract. It is estimated that one in four women during reproductive period will develop this kind of benign neoplasia. Mediator complex Subunit 12 (MED12) gene is localized on Xq13 spans 45 exons encodes for MED12 a component of the mediator complex that has a regulatory role in RNA polymerase II activity. It participates in various molecular pathways, for example, p53 and Wnt/b-catenin pathways. Most of the mutations in the MED12 gene are identified in exon 2 region which leads to the development or growth of fibroids.

Aim and Objectives:

The aim of the present study is to determine the role of MED12 (exon 2) variations with uterine fibroids in South Indian population from Telangana. The objective of the study is to identify the variations associated with the disease phenotype.

Materials and Methods:

A case-control study was conducted with a total of 104 clinically, ultrasonographically confirmed uterine fibroid patients and 104 healthy women without the history of any gynec problems were included in the present study. Detection of MED12 gene (exon 2) variations were carried out by PCR-amplification followed by SSCP (single stranded conformation polymorphism) analysis.

Results:

The results obtained in the study showed three different band patterns (A, B, C). The frequency of band patterns A, B and C were 60.57%, 23.07% and 16.34% in controls and 65.38%, 6.73% and 27.88% in patients with uterine leiomyoma respectively. The study revealed an increased frequency of band pattern C and low frequency of B in UL cases than in controls.

Conclusion:

The study revealed a possible association of MED12 gene in the etiopathogenesis of uterine leiomyoma. However, large number of samples has to be analyzed to confirm the results obtained.