Copy number variation in pelvic endometriosis

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Samples consisting of 673 controls and 65 patients with Stage III, IV endometriosis were used. The average age of subjects was 41.2±11.6 in controls, 35.2±8.0 in patients with Stage III endometriosis and 34.5±8.3 in patients with Stage IV. In CNVR of 1q21.3, there was no coding gene and the frequency of copy number loss CNVs was significantly lower in endometriosis patients compared to controls (p<0.028). In CNVR of 1p13.3, GSTM1 gene was positioned and the frequency of copy number gain CNVs was significantly higher in endometriosis patients than controls (p<0.038). It was found through Genomic qPCR that 1q21.3 CNVs had a significant difference in an independent replication set as well. While 20 out of 42 controls had deletion with the intensity ratio of 0.5 or less, only 9 out of 37 patients showed deletion (p=0.032). The average signal intensity ratio of controls was also lower than that of cases (0.6 versus 0.8). When it comes to CNVR 1p13.3, the average signal intensity ratio of patients was higher than that of controls (3.7 versus 3.0) and the frequency of copy number gains (intensity ratio ≥2) was higher in patient group (19 out of 37) than control group (16 out of 42). However, there was no statistical significance (p=0.235). This study identified two CNVRs for endometriosis. 1q21.3 and 1p13.3 could be a potential target for screening and diagnosis of pelvic endometriosis.

Biography

Jae Yen Song has completed his PhD from The Catholic University of Korea. She is the Assistant Professor of the Catholic University of Korea. She is Committee Member of many Korean associations and societies. She has published more than 35 papers in reputed journals and has been serving as an Editorial Board Member of repute. She has received the Best Oral Presentation Award of the Korean Society of Menopause and Best Poster Presentation Award of Korean Association of Obstetrics & Gynecology Shan S. Ratnam-Young Gynecologist Award et al., and research funds.

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