Commentary on: Identification of pelvic organ prolapse risk susceptibility gene SNP locus in Xinjiang women

Laura (L) Spinnewijn¹

Received: 3 August 2019 / Accepted: 19 August 2019 / Published online: 29 August 2019
© The International Urogynecological Association 2019

In this prospective study 196 women from the Xinjiang region in the northwest of China, with or without pelvic organ prolapse (POP), were genetically screened for POP susceptibility genes. Previous studies have provided evidence that genetics play a role in the complex and multivariate etiology of POP, yet study results on which genes contribute significantly are conflicting between populations. The Xianjing area is populated with people from different ethnic backgrounds that usually do not intermarry with other ethnicities and therefore provides a specific and stable ethnic/genetic isolation group. Identification of susceptibility genes and SNP loci regional specificity within this population could provide a theoretical basis for early POP diagnosis, treatment and prevention.

Eighty-eight women with POP stage III or IV and 108 women without POP (e.g., no more than stage I), with either group consisting of both ethnic minority and non-minority populations, were genotyped by testing their DNA for 16 known POP candidate susceptibility gene SNP loci. Researchers performed the χ^2 test to compare categorical data between cases and controls, calculating odds ratios for POP and using the Hardy-Weinberg equilibrium to enable comparisons of genetic structures with the assumption that they do not evolve over time. Results showed that some POP susceptibility genes indeed increase or decrease the risk of POP, with differences in genotypes between minority and non-minority populations, respectively. In women belonging to the ethnic minorities the following genotypes

lead to significant changes in POP risk: estrogen receptor α gene (ESR1) rs17847075 genotype AG increased POP risk by 2.738 times; ESR1 rs2234693 genotype TC increased POP risk by 2.99 times; zinc finger and AT hook domain gene (ZFAT) rs1036819 genotype CC or allele C increased POP risk by 10.286 or 2.212 times, respectively; fibulin 5-gene (FBLN5) rs12589592 genotype AA or allele A decreased POP risk by 0.111 or 0.482 times, respectively. In non-minority patients ESR1 rs17847075 genotype GG or allele G decreased POP risk by 0.247 and 0.483 times, respectively, and ZFAT rs 1036819 genotype CC decreased the risk by 0.182 times.

This study adds to the limited information on POP susceptibility genes. However, study results seem only valid within this small population, as it consists of several even smaller ethnic groups, with inconsistent presence of candidate POP susceptibility genes between those groups. Furthermore, the clinical significance of these genes in general is yet to be established, as other factors contributing to POP occurrence have to be taken into account as well, and no evidence is available on what interventions or protective measures could treat or even prevent the occurrence of POP in patients with specific genotypes.

Publisher's note Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.

✉ Laura (L) Spinnewijn
lauraspinnnewijn@gmail.com

¹ Department of Obstetrics and Gynaecology, Radboudumc, Geert Grooteplein Zuid 10, 6525GA Nijmegen, The Netherlands