

Genetic Markers For The Diagnosis Of Genital Prolapsed

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Abstract: Prolapse is assumed to be caused by genetically specified alterations in the genital connective tissue. As a result, genetic variables that cause connective tissue pathology take center stage in the quest for genetic predisposition. Collagen proteins are part of the ligaments that ordinarily maintain the pelvic organs and strengthen the muscles involved in pelvic organ prolapse, hence studying their polymorphism in PG is significant.

Keywords: estrogen receptors, allele, genotype, Genital prolapse (GP)

Introduction

The main etiological and pathogenetic factors in the development of pelvic prolapse are:

- violations of the anatomical and neuromuscular interactions of the structures of the pelvic cavity and the ligamentous apparatus of the genital organs;
- a change in the structure of receptor interactions (in particular, the components of the extracellular matrix - and the composition of muscle receptors) and a violation of their interaction;
- receptors form disturbances in the system of intracellular and extracellular mediators of signal transmission.

According to the data obtained, 33.3% of relatives have a genetic predisposition to the development of HP [1, 2]. However, HP is often accompanied by other clinical manifestations of connective tissue dysplasia (CTD): hernias, flat feet, scoliosis, joint hypermobility, varicose veins, etc. [3, 6].

Girls with GP mothers had a 2-3-fold increased risk of acquiring the disease, while familial ties have a 5-fold increased risk [5-4]. Stress urine incontinence and the prevalence of GP in major diversified forms of GP have been documented by a number of writers [7]. Identification of genetic variants that define propensity to GP development and are responsible for the formation of specific pathogenesis pathways plays a significant role in this regard.

Approximately 32 genetic determinants have been identified to date, with polymorphisms causing functional abnormalities of the pelvic organs. [8] Our research looked at COL1A1 (rs1107946) and COL4A1 (rs605143) genetic polymorphisms in the sacral uterine ligament of patients with genital prolapse based on tissue damage, study results and support for GP-related receptor support in the pelvic cavity. studied by detection. In order to determine the importance of polymorphisms of the studied genes in the genesis of PG, we statistically analyzed the frequency of occurrence of alleles and polymorphisms of type IV collagen genes COL4A1 (rs605143) and - type I collagen genes COL1A1 (rs1107946). Statistically significant differences were found when comparing the

frequencies of alleles and genotypes of the gene rs1107946 and rs605143 polymorphisms between the GP and the general group of patients with the population sample. Frequency of occurrence of G and A alleles: 97 and 25 (79.51% and 20.49%) in rs1107946 - in the main group of patients and 107 and 19 (84.92 and 15.08%) - in the control group. It can be seen that an increase in allele frequency (20.49%) was observed in the patient group compared to the control group (15.08%). In control groups 1 and 2 of patients, a negative value of the relative deviation of D (-0.07, -0.07, and -0.07) was found to the left of indicator D 0. This indicates a relatively high frequency of detected true heterozygotes, and is not a theoretical calculation, especially in patients with PTO. However, heterozygous prolapse of 1 group was 3.57 times and 1.28 times in 2 group that is, heterozygous prolapse was more specific to genital processes.

In the group of healthy donors, the H_{exp} value is longer than 0.5, indicating that the heterozygosity of this locus in the population is very low. However, the relative deviation of the expected heterozygosity observed in the control group has a negative value ($D = -0.07$). These data prove the lack of heterozygous COL1A1 and COL4A1 genes rs1107946 and rs605143 in the Uzbek population. Low heterozygosity ($H_{exp} = 0.14$) and absence of homozygous A / A genotype were observed more frequently (especially in the patient group).

In conclusion, some methods and mechanisms of development of many genital prolapses have now been identified at the systemic and molecular genetic and biochemical levels. Moreover, there is a lot of contradicting information about the pathophysiology of PG in the past and present. Furthermore, because the found genetic variations are not particular to this condition, the search for effective techniques to diagnose and treat GP development in advance should be improved.

References

1. Смольнова Т.Ю., Адамян Л.В., Ляшко Е.С. Ассоциированная патология органов малого таза и тазового дна у больных с дисплазией соединительной ткани: Материалы Первой Все российской научно-практической конференции. Омский научный вестник. 2005;32(5):83.
2. Смольнова Т.Ю. Адамян Л.В., Ляшко Е.С. Синдром дисплазии соединительной ткани в акушерстве и гинекологии. В сб.: Кулаков В.И., Адамян Л.В. Современные технологии в диагностике и лечении гинекологических заболеваний. М. 2005;257.
3. Смольнова Т.Ю., Савельев С.В., Гришин В.Л., Титченко Л.И., Яковлева Н.И. Проплапс гениталий — следствие травматичных родов или генерализованной дисплазии соединительной ткани. Акушерство и гинекология. 2001;4:50-53.
4. Andrada Hamer M, Persson J. Familial predisposition to pelvic floor dysfunction: prolapse and incontinence surgery among family members and its relationship with age or parity in a Swedish population. Eur J Obstet Gynecol Reprod Biol. 2013;170(2):559-562.doi: 10.1016/j.ejogrb.2013.07.025
5. Hansell NK, Dietz HP, Treloar SA, Clarke B, Martin NG. Genetic covariation of

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- pelvic organ and elbow mobility in twins and their sisters. *Twin Res.* 2004;7(3):254-260. doi: 10.1375/twin/7/3/254
6. Norton PA, Baker JE, Sharp HC, Warenski J.C. Genitourinary prolapse and joint hypermobility in women. *Obstet Gynecol.* 1995;85(2):225-228. doi: 10.1016/0029-7844(94)00386-R
 7. Carley M, Schaffer J. Urinary incontinence and pelvic organ prolapse in women with Marfan or Ehlers Danlos syndrome. *Am J Obstet Gynecol.* 2000;182(5):1021-1023. doi: 10.1067/mob.2000.105410
 8. Cartwright R, Kirby AC, Tikkinen KA, Mangera A, Thiagamoorthy G, Rajan P, et al. Systematic review and metaanalysis of genetic association studies of urinary symptoms and prolapse in women. *Am J Obstet Gynecol.* 2015;212(2):199.e1-e24. doi: 10.1061/j.ajog.2014.08.005
 9. Chen B, Yeh J. Alterations in connective tissue metabolism in stress incontinence and prolapse. *J Urol.* 2011;186(5):1768-1772. doi: 10.1016/j.juro.2011.06.054 doi: 10.12816/0000402