

■ РЕФЕРАТИ СТАТЕЙ, ОПУБЛІКОВАНИХ  
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**CYSTATHIONINE  $\beta$  SYNTHASE (CBS 844INS68) AND CYTOSOLIC SERINE HYDROXY-METHYLTRANSFERASE (SHMT1) (C1420T) GENE POLYMORPHISMS AND THE RISK OF BREAST CANCER IN EGYPTIAN POPULATION**

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*Breast Cancer BC is the extreme most frequent cancer among women. Death from BC is around 15% of all cancer deaths among women. BC risk factors are classified into non-modifiable risk factors and modifiable risk factors. Excessive studies on folate pathway have been done as a possible mechanism for rising cancer. The present work aimed to study genetic polymorphisms in cystathionine beta-synthase CBS (CBS 844ins68) and the cytosolic serine hydroxy-methyltransferase-1 SHMT1 (C1420T) genes and their expression in patients with breast cancer. Methodology the study included 100 diagnosed breast cancer patient and 100 healthy as a control. Conclusion of this study, results obtained indicated that; there is a significant association*

*between CBS 844ins68 polymorphism and increased risk of breast carcinoma as There was a significant difference in the frequency of the mutant insertion allele between breast cancer patients and control subjects, Also for SHMT1 C1420T SNP, there was a significant difference in the frequency of the mutant T allele between breast cancer patients and control subjects that reflected that SHMT1 C1420T may has a protective effect against breast cancer. On The level of gene expression of CBS and SHMT1 genes, there was insignificant difference between malignant and adjacent normal tissue samples as a control.*

**Key words:** Breast cancer, CBS (CBS 844ins68), SHMT1 (C1420T), gene expression.

**ГЕНЕТИЧНИЙ ПОЛІМОРФІЗМ ЦИСТАТІОНІН В-СИНТАЗИ (CBS 844INS68) І ЦИТОЗОЛЬНОЇ СЕРИН-ГІДРОКСИМЕТИЛТРАНСФЕРАЗИ (SHMT1) (C1420T) ТА РИЗИК ВИНИКНЕННЯ РАКУ МОЛОЧНОЇ ЗАЛОЗИ СЕРЕД НАСЕЛЕННЯ ЄГИПТУ**

Рак молочної залози (РМЗ) — це надзвичайно поширене онкологічне захворювання серед жінок. Летальні випадки, спричинені РМЗ, становлять близько 15 % всіх смертей жінок від раку. Фактори ризику РМЗ поділяють на незмінні та змінні. Було проведено велику кількість досліджень щодо фолатного шляху як можливого механізму виникнення раку. Мета цієї роботи полягала у вивченні генетичних поліморфізмів у генах цистатіонін бета-синтази CBS (CBS 844ins68) та цитозольної серин-гідроксиметилтрансферази-1 SHMT1 (C1420T) та їхнього вираження у пацієнтах із раком молочної залози. Методологія дослідження передбачала участь 100 пацієнток із діагнозом раку молочної залози та 100 здорових жінок в якості контрольної групи. Результати дослідження показали значний зв'язок між поліморфізмом CBS 844ins68 і вищим ризиком карциноми молочної залози. Також існують суттєві відмінності між пацієнтками з раком молочної залози та здоровою контрольною групою щодо частоти вставленого мутантного алеля. Що стосується SHMT1 C1420T SNP, було виявлено суттєву різницю між пацієнтками та контрольною групою щодо частоти мутантного T алеля. Це означає, що SHMT1 C1420T може мати захисний вплив проти виникнення раку молочної залози. На рівні експресії генів CBS та SHMT1 було виявлено незначну

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різницю між зразками злоякісних та прилеглих нормальних тканин, використаних в якості контролю.

**Ключові слова:** рак молочної залози, CBS (CBS 844ins68), SHMT1 (C1420T), експресія генів.

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