

Methylenetetrahydrofolate Reductase

1, 2, 3
 1, 1, 2, 2, 2, 3
 2, 2, 2

The Analysis of Methylenetetrahydrofolate Reductase Mutation in Recurrent Spontaneous Abortion

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Objective: To analyze the methylenetetrahydrofolate reductase (MTHFR) mutation in patients with recurrent spontaneous abortion.

Material and Method: The blood samples of patients with recurrent spontaneous abortion were tested by PCR-RFLP method.

Results: Of 51 cases of study group, 14 (27.5%) were normal, 25 (49.0%) were heterozygosity, and 12 (23.5%) were homozygosity. Of 58 cases of control group, 20 (34.5%) were normal, 30 (51.7%) were heterozygosity, and 8 (13.8%) were homozygosity. But the difference between two groups was not significant (p=0.190).

Conclusion: Hyperhomocysteinemia due to MTHFR mutation is a cause of recurrent spontaneous abortion. Therefore, the study for MTHFR mutation should be included in the workup of recurrent spontaneous abortion.

Key Words: Recurrent spontaneous abortion, MTHFR mutation

가 , 2,3 가 .
 Homocysteine
 (homocystinuria) homocy-
 8 steine (remethylation)
 (trans-sulfuration) 가
 가
 1
 가 steine homocy-

Table 1. Frequency of MTHFR gene mutation in patient with recurrent spontaneous abortion and control group

Group	Number	Genotypes of MTHFR (%)		
		677CC	677CT	677TT
RSA	51	14 (27.5)	25 (49.0)	12 (23.5)
Control	58	20 (34.5)	30 (51.7)	8 (13.8)

Table 2. Prevalence of 677TT MTHFR mutation in different countries

Country	Homozygous mutant (677TT) of MTHFR		
	RSA	Control	OR (95% CI)
Netherlands	16.0 (29/185)	5.0 (6/113)	3.3 (1.3~8.3)
Italy	18.1 (17/ 94)	18.7 (28/150)	1.0 (0.5~1.9)
France	20.0 (20/100)	14.0 (14/100)	1.5 (0.7~3.2)
Israel	9.7 (4/ 41)	22.2 (4/ 18)	0.4 (0.1~1.7)
U.K.	8.1 (11/129)	8.9 (6/ 67)	0.9 (0.3~2.7)
Korea	23.8 (12/ 51)	13.8 (8/ 58)	1.9 (0.7~5.1)

OR (95% CI): odds ratio and 95% confidence interval calculated for the TT genotype versus the other two genotypes in cases versus control

(51.7%), 가 8 (13.8%) (Table 1), 가 homocysteine homocysteine (p=0.190). CBS homocysteine (homo-cystinuria) homocysteine homocysteine Homocysteine thiol (thromboembolism) 가 methionine (deme- 30 50% thylated derivative) methionine 1 homocysteine homocysteine, cy- steine-homocysteine disulphide homocysteine 가 steine homocysteine Homocys- homocysteine 2가 가 (homozygosity) 가 methionine (remethylation) homocysteine methionine cystathionine cysteine HFR) methylenetetrahydrofolate reductase (MT- (trans-sulfuration) homocysteine methionine synthase, B₁₂가 (heterozygosity) ine â-synthase (CBS) B₆가 50% 23 methylenetetrahydrofolate-homo-

cysteine methyltransferase

homocysteine

MTHFR

homocysteine

가

4 MTHFR

homocysteine

50

MTHFR

(fibroblast)

15%

5

가

homocysteine

V

(activated protein C resistance: APCR)

6

37

46

MTHFR

가

7

5%

17%가

677

alanine

valine

35%

8

40~45%가

37

40~

50%

46

35%

Alanine

valine

MTHFR

homocysteine

가

9

가

10

homocysteine

가

homocysteine

B

가

homocysteine

Homocysteine

homocysteine thiolactone,

homocysteine,

가

homocysteine

70~80%

11

5~15 ì mol/L

12 Homocysteine

15~30 ì mol/L, 30~100 ì mol/L, 100 ì mol/L

37가

homocysteine

가

homocysteine

methionine

14 Methionine

homocysteine

methionine

MTHFR

homocysteine

methionine

가

2

가

methionine

homocysteine

methionine

가

15

8~12

23

(extra-embryonic coelomic fluid)

methionine

가

methionine

ho-

methionine

Methionine

S-adenosyl-

methionine

S-adenosyl-methionine

methionine

가

S-adenosyl-methionine/S-adenosyl-

homocysteine

S-adenosyl-methionine

가

ho-

methionine

가

¹⁶ 가 methionine 가
 homocysteine
 50%
 CBS
 25~33% homocysteine methionine
 ionine
^{17,18}
 MTHFR
 가 3 ¹⁹
 MTHFR
 homocysteine methionine
 76 106
 MTHFR
 가 가 ²⁰
³⁰
 homocysteine 가 가 7 ²¹
 homocysteine
 2~3 Homocysteine
 17 2
 MTHFR 가 2~3
²² homocysteine
 가 ²³
 18% methionine
²⁴ 26%,
 11%, 38%
 homocysteine ²⁵
²⁶
 Homocysteine
 pyridoxine
 1~5 mg homocysteine 가
²⁷
 B₁₂ B₆ B₆ B₁₂
 homocysteine
 4~6 homocysteine 가

2
 1960 B₆
²⁸
 homocysteine
 Homocysteine 가 20%
 가2 가 homo-
 cysteine
 가 homocysteine
 가 Methionine
 homocysteine B₆
 가
 가
 가
 가
 MTHFR
 (thrombophilia)
 가

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