

# Genetic Screening

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가 , 가 , 가 , 가 , 가 , 가 , 가 , 가

BRCA1(breast cancer susceptibility gene 1)

BRCA2 . BRCA1 BRCA2 17q21

13q12.3 100,000bp 70,000bp

가 . BRCA

가 (integrity)

DNA , , . BRCA1/BRCA2

(penetrance) 가 55 - 75%

가 . TP53, PTEN, STK11/LKB1, ATM, MLH1/MSH2 3

(Table 1).

BRCA1/BRCA2

1.

5 - 10%

**Table 1.** Genetic Predisposition to Breast Cancer

Syndrome	Gene	Inheritance	Cancers
Breast/ovarian cancer syndrome	BRCA1	AD	Breast, ovary
	BRCA2	AD	Breast, ovary, prostate, pancreas
Li-Fraumeni syndrome	TP53	AD	Breast, brain, sarcoma, leukemia, adrenocortical carcinoma
Cowden disease	PTEN	AD	Breast, ovary, thyroid, colon
Peutz-Jegher syndrome	STK11/LKB1	AD	GIT, breast
Ataxia-telangiectasia	ATM	AD	Breast
Site-specific breast cancer	CHEK2	AD	Breast
Muir-Torre syndrome	MLH1/MSH2	AD	Colorectal, breast

2. BRCA1/BRCA2

(2004) 40 60  
 15% BRCA1/BRCA2  
 (founder mutation) BRCA 가  
 가 가  
 BRCA1/BRCA2 BRCA (2004)  
 BRCA 가 , 2.8%  
 가 , 가 , (BRCA1 1.8%, BRCA2 1.0%) BRCA  
 Ashkenazi 2% , 12.7%  
 BRCA1/BRCA2 가, (BRCA1 8.7%, BRCA2 4%)가  
 0.1% BRCA1 가 .  
 first - degree relatives 가 ,  
 BRCA1 0~5.7%, , 40  
 BRCA2가 0.2~8.5% , (<35 )  
 BRCA1 0.7~8.6%, BRCA2가 1.3~6.6%  
 (Table 2).  
 BRCA1 84%, 가 44%  
 가 55% , (Table 3) 가  
 가 75% 가  
 (2002) , BRCA2 BRCA1  
 가 21 가 9가 .  
 (43%) BRCA1/BRCA2 BRCA  
 5 Breast Cancer Information Core (BIC)

**Table 2.** BRCA Mutations Among Cases of Female Breast Cancer Unselected for Family History (Modified Data from Table 1 in Hum Mutat 20: 413-424, 2002)

Population	Groups	Estimated prevalence		
		BRCA1 (%)	BRCA2 (%)	Total (%)
Europe	Unselected	0.4 - 5.7	0.2 - 8.5	1.8 - 5.9
	Selected*	0.7 - 6.8	1.3 - 2.4	2.0 - 9.0
North America	Unselected	0.0 - 2.6	3.1	3.1
	Selected	5.9 - 7.5	3.4 - 6.6	9.4 - 13.1
Australia	Unselected	-	-	-
	Selected	2.3 - 3.8	2.3	4.6
Asia	Unselected	0.8 - 4.4	2.3 - 4.1	5.1 - 6.7
	Selected	3.6 - 8.6	2.4	6.0
Korea	Unselected			
	( )	1.4	1.2	
	( )	1.8	1.0	2.8
	Unselected			
( )	8.7	4.0	12.7	
( )	10	8.7	15	

\*Selected groups were collected by age, family history, and bilaterality etc.

3.

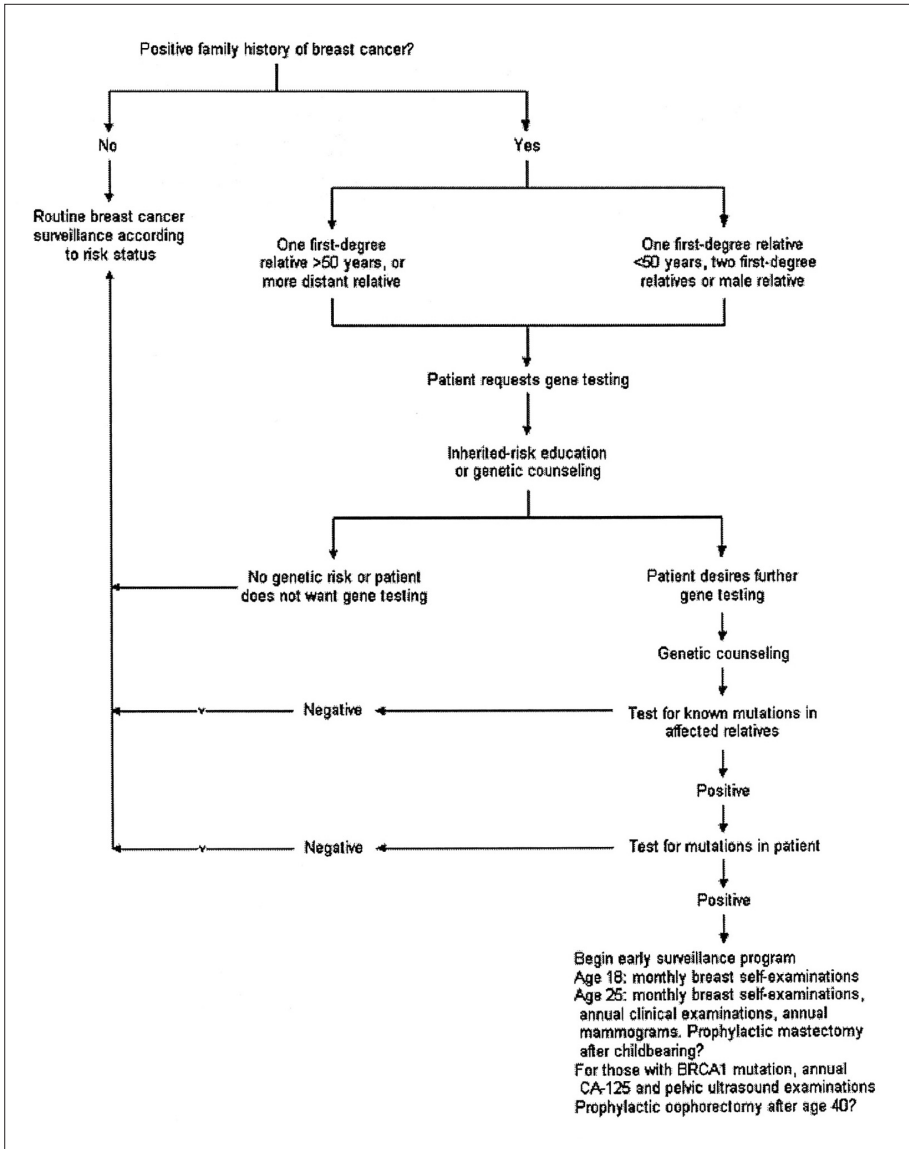
National Cancer Institute(NCI)  
 BRCA1/BRCA2 가 ( )  
 Johns Hopkins , , 가  
 , 가 , , , 가  
 가  
 ,  
 , Ashkenazi  
 ,  
 , BRCA1 BRCA2 가  
 .  
 가  
 , 35 .  
 , ,  
 .  
 가  
 , F - CSGE (fluorescent conformation - sensitive gel electrophoresis)  
 DHPLC(denaturing high performance liquid chromatography) 가  
 가 . BRCA1/BRCA2 가  
 100/100 가  
 297,000 (\$2700)

가 가 .  
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 4. 가  
 가  
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 ,  
 BRCA1/BRCA2 가  
 가  
 가  
 가  
 가  
 100%  
 BRCA1/BRCA2 penetrance 가  
 , BRCA1 84%,  
 가  
 44%  
 18 가  
 , 25 가  
 . BRCA1  
 CA - 125

**Table 3.** Cumulative Risk by Age of Breast Cancer in Women from Families with BRCA1 and BRCA2 Mutations (Easton et al, 1995, 1997)

Age (yrs)	Cumulative risk (%)	
	BRCA1	BRCA2
30	3.2	4.6
40	19.1	12.0
50	50.8	46.0
60	54.2	61.0
70	85.0	86.0

- 
- Personal Characteristics**  
 Breast cancer diagnosed at an early age  
 Bilateral breast cancer  
 A history of both breast and ovarian cancer  
 The presence of breast cancer in 1 or more male family members
- Family Characteristics1**  
 Multiple cases of breast cancer in the family  
 Both breast and ovarian cancer in the family  
 One or more family members with 2 primary cancers  
 Ashkenazi Jewish background
-



**Fig. 1.** Suggested algorithm for genetic screening in women with a family history of breast cancer (based on recommendations of NIH and National Human Genome Research Institute)

ifene , tamoxifen, ralox -

BRCA1/

BRCA2 , founder mutation

penetrance .

가 . , 가

5. , 가

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1. : . 10  
, 2004. Breast and ovarian surveillance service (Johns Hopkins):  
<http://www.hopkinsmedicine.org/breastcenter/care/boss/>
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