Results

Table (6): **Mean values ± SD of woman age and age at onset of menarche among control and breast cancer groups.**

Groups / Parameter	Control		Breast cancer groups		F	*p*
	Without family history N =10	With family history N=10	Without family history N =15	With family history N=15		
	Mean ± SD					
Age	34 ± 6.1	28 ±10.7	40.9 ±13.3	39.7 ± 6.9	**4.14**	**>0.05**
Onset of menarche	12.3 ±0.66	11.7 ±0.95	11.5 ±0.99	11.5 ±0.64	**2.52**	**>0.05**

***p*>0.05 : non significant**

***Fig. 3*: Woman age and age at onset of menarche among control and breast cancer groups.**

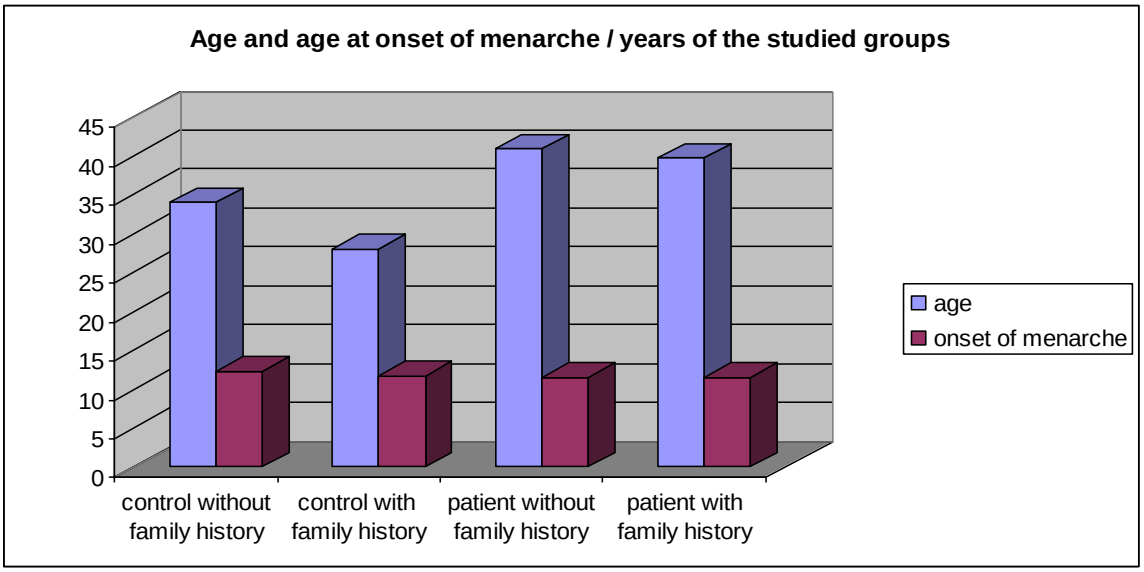


Table (6) & Fig. (3) Show that there is no statistically significant difference in age and age at onset of menarche among the different studied groups.

Table (7): **Subject number and percentage of control and breast cancer groups according to some risk factors**

groups / character	Controls		Breast cancer groups		X^2	*P*
	Without family history N =10	With family history N=10	Without family history N=15	With family history N=15		
	Number (%)					
Marital status						
• **Married**	10/10 (100)	8/10 (80)	14/15 (93.3)	14/15 (93.3)	**2.8**	**>0.05**
• **Single**	0/10 (0)	2/10 (20)	1/15 (6.7)	1/15 (6.7)		
Lactation						
• **+ve**	10/10 (100)	6/10 (60)	12/15 (80)	12/15 (80)	**5**	**>0.05**
• **-ve**	0/10 (0)	4/10 (40)	3/15 (20)	3/15 (20)		
Parity						
• **+ve**	10/10 (100)	6/10 60	13/15 (87.7)	12/15 (80)	**5.74**	**>0.05**
• **-ve**	0/10 (0)	4/10 (40)	2/15 (13.3)	3/15 (20)		
Hormonal Contraception						
• **+ve**	3/l0 (30)	3/10 (30)	3/15 (20)	5/15 (33.3)	**4.93**	**>0.05**
• **-ve**	7/10 (70)	710 (70)	12/15 (80)	10/15 (66.6)		
History of DM						
• **+ve**	0/10 (0)	0/10 (0)	3/15 (20)	2/15 (13.3)	**4.07**	**>0.05**
• **-ve**	10/10 (100)	10/10 (100)	12/15 (80)	13/15 (86.7)		

p>0.05 : non significant

Table (7) shows the number and percentage of each group subjects according to some risk factors and indicates that there is no statistically significant difference in number of females among studied groups according to marital status, parity, lactation, contraception and history of DM (p>0.05).

Table (8): **Percentage of 1st Degree and 2nd degree relatives and the history of breast and ovarian cancers in the control and Breast cancer groups with family history**

Group` / Item		Control group with +ve family history	Breast cancer group with +ve family history
		No. (%)	No. (%)
Degree of family relative	1st degree	10/10 (100)	14/15 (93.3)
	2nd degree	0 (0)	1/15 (6.7)
Type of cancer	Breast cancer	9 (90)	14 (93.3)
	Ovarian cancer	1 (10)	1 (6.7)

Table (8) shows that the percentage of the first degree relative with breast and/or ovarian cancer is extremely higher than that of the second degree relative in the control and breast cancer groups with family history ***(Fig. 4)*** .It also shows that the percentage of the history of breast cancer is much higher than that of the history of ovarian cancer in the control and breast cancer groups with family history.

Fig. (4): **Percentage of relative's degree among breast cancer patients with family history**

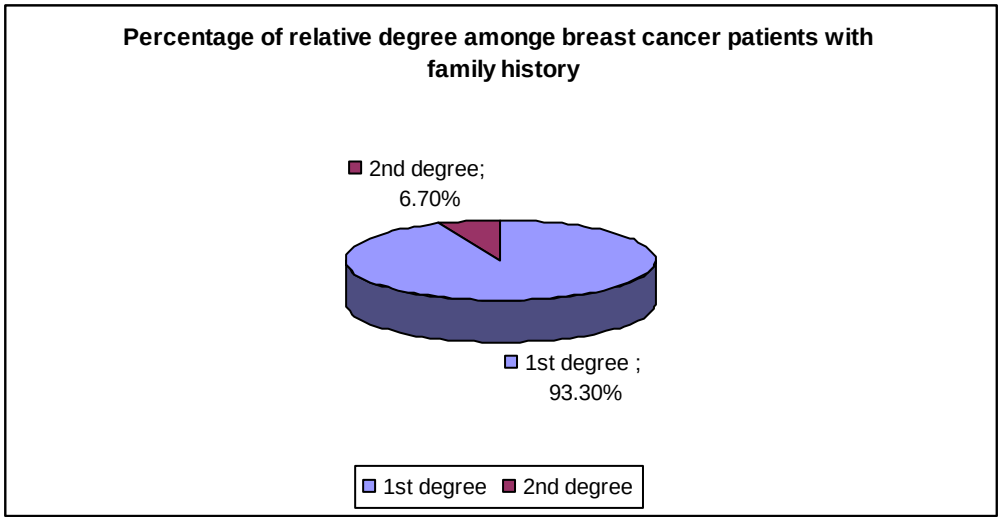


Table (9): **Exons mutation frequencies in BRCA1Gene among control and breast cancer groups**

Groups / Exons mutation frequency	control		Breast cancer groups		X^2	p
	Without family history N =10	With family history N=10	Without family history N =15	With family history N=15		
	Number (%)					
• **Normal gene**	5/10 (50)	4/10 (40)	2/15 (13.3)	2/15 (13.3)	12.3	>0.05
• **One exon mutation**	5/10 (50)	3/10 (30)	7/15 (46.7)	4/15 (26.7)		
• **Two or three exons mutation**	0/10 (0)	3/10 (30)	6/15 (40)	9/15 (60)		
• **Total**	5/10 (50)	6/10 (60)	13/15 (86.7)	13/15 (86.7)	23.3	<0.05

Fig.(5): **Exons mutation frequencies in BRCA1Gene among control and breast cancer groups**

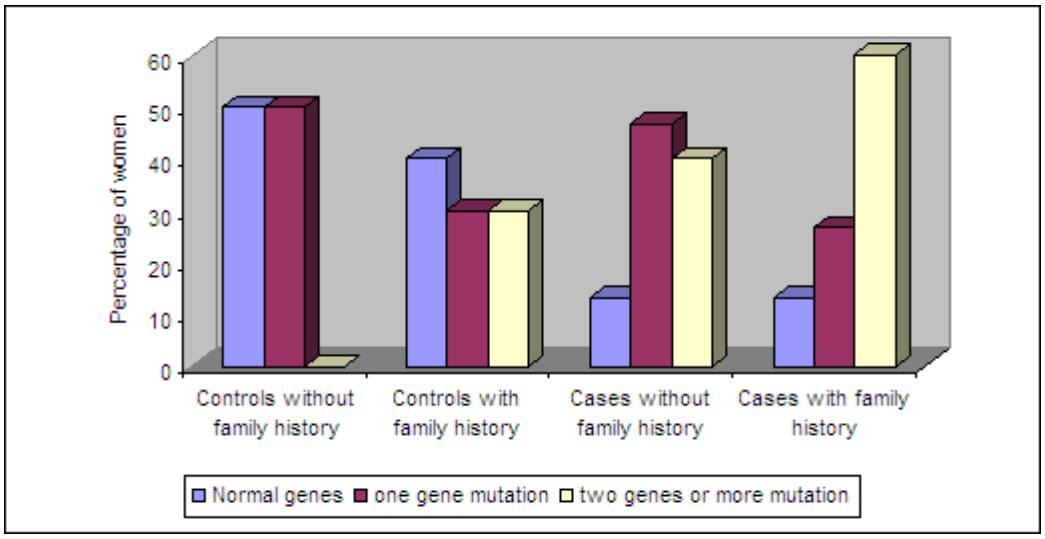


Fig.(6): **Incidence of BRCA1 gene mutations among control groups**

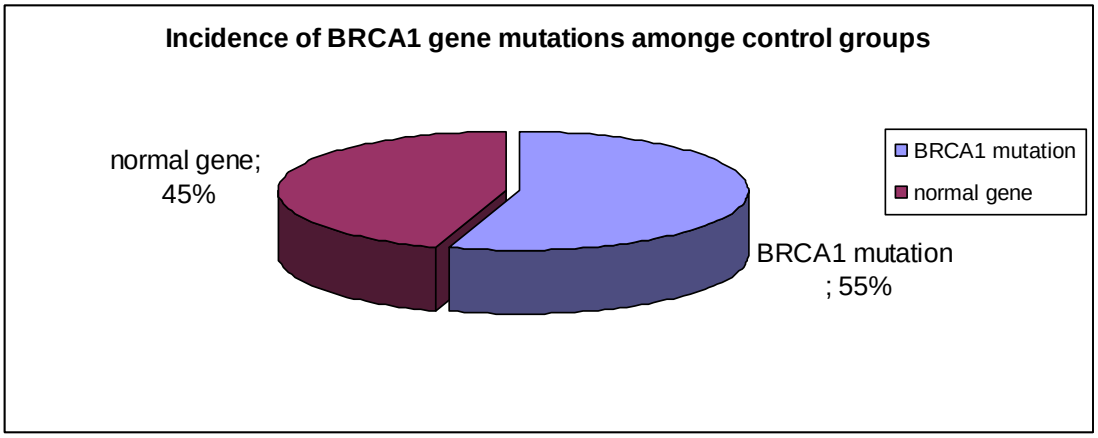
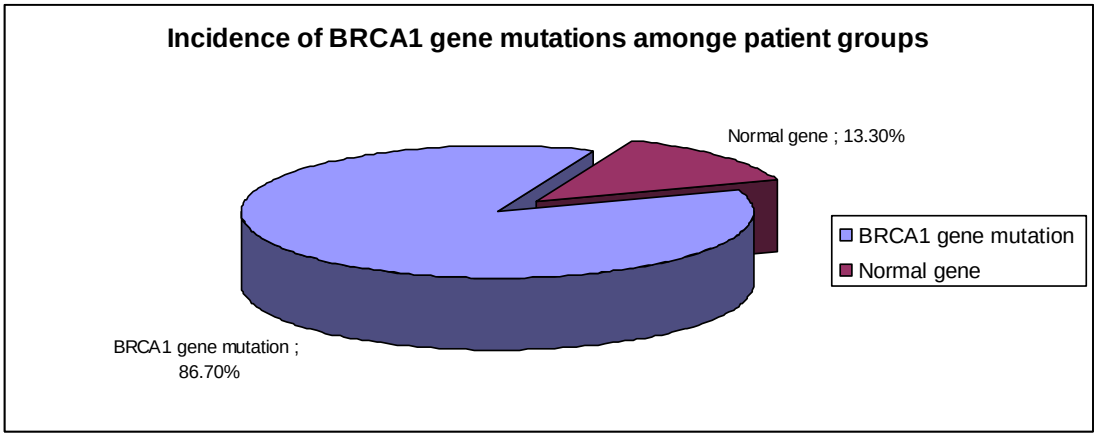


Fig.(7)): **Incidence of BRCA1 gene mutations among patient groups**



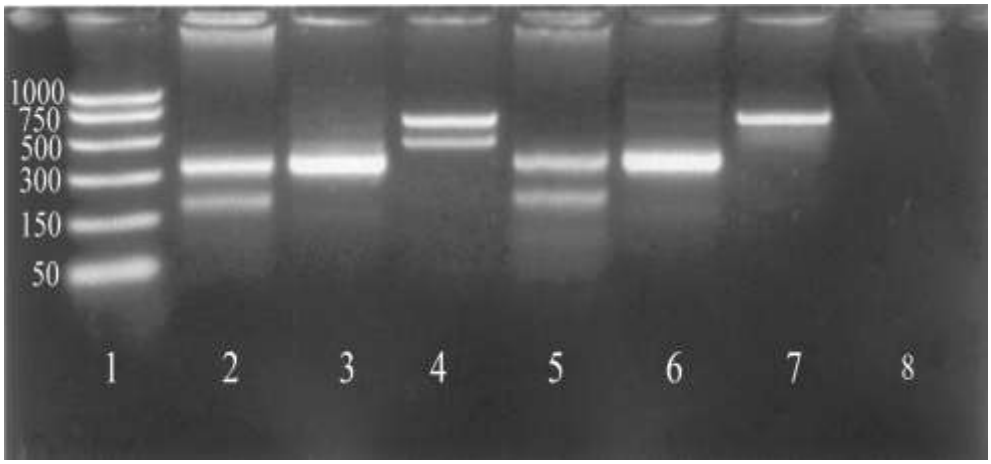


Fig. (8): gel electrophoresis of amplified products of BRCA1 gene of exon 2 and exon 11 , lane 1 indicate DNA ladder 1000bp ,In lane 4 and7 exon 2 two bands indicate one for wild and the second for mutant at 638bp and 533bp, respectively lanes 2,3.5 and 6 indicates exon 11 at 354 bp and at227bp

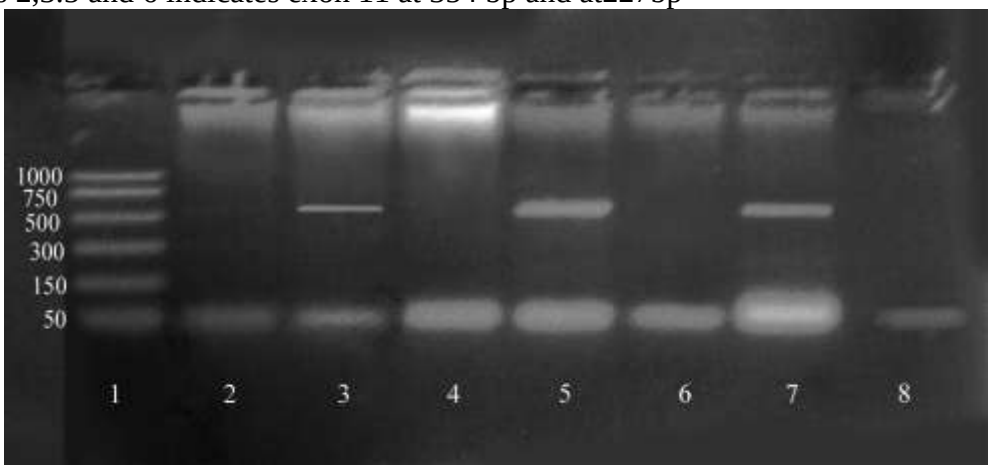


Fig. (9):gel electrophoresis of amplified products of BRCA1 gene of exon 5, lane 1 indicate DNA ladder 1000bp ,before digestion with avaII in lanes 3,5 and 7

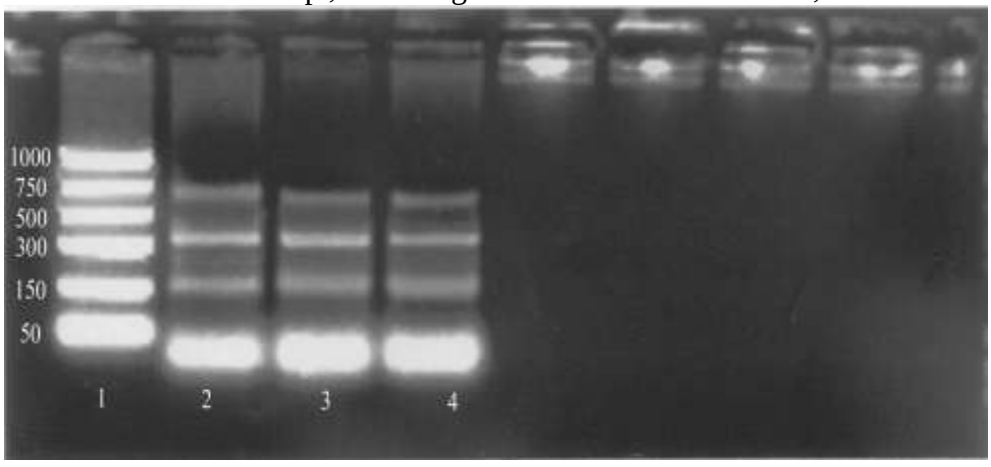


Fig. (10):gel electrophoresis of amplified products of BRCA1 gene of exon 5, lane 1 indicate DNA ladder 1000bp,,after digestion with avaII, three bands indicate herterozgous in lanes 2,3 and 4.

Table(10): **Genotype frequency of exons 2 , 5 and 11 among control and breast cancer groups**

Groups / Genotype frequency	control group		Breast cancer group		Total	X^2	p
	Without family history N =10	With family history N=10	Without family history N =15	With family history N=15			
	Number %						
Exon 2 Mutation	0/10 (0)	2/10 (20)	4/15 (26.7)	5/15 (33)	11/50 (22)	4.16	>0.05
Exon11 Mutation	3/10 (30)	5/10 (50)	11/15 (73.3)	9/15 (60)	28/50 (56)	4.8	>0.05
Exon 5 Mutation	2/10 (20)	4/10 (40)	5/15 (33)	9/15 (60)	20/50 (40)	4.44	>0.05

P>0.05: non significant

Fig.(11): **Genotype frequency of exons 2 , 5 and 11 among control and breast cancer groups**

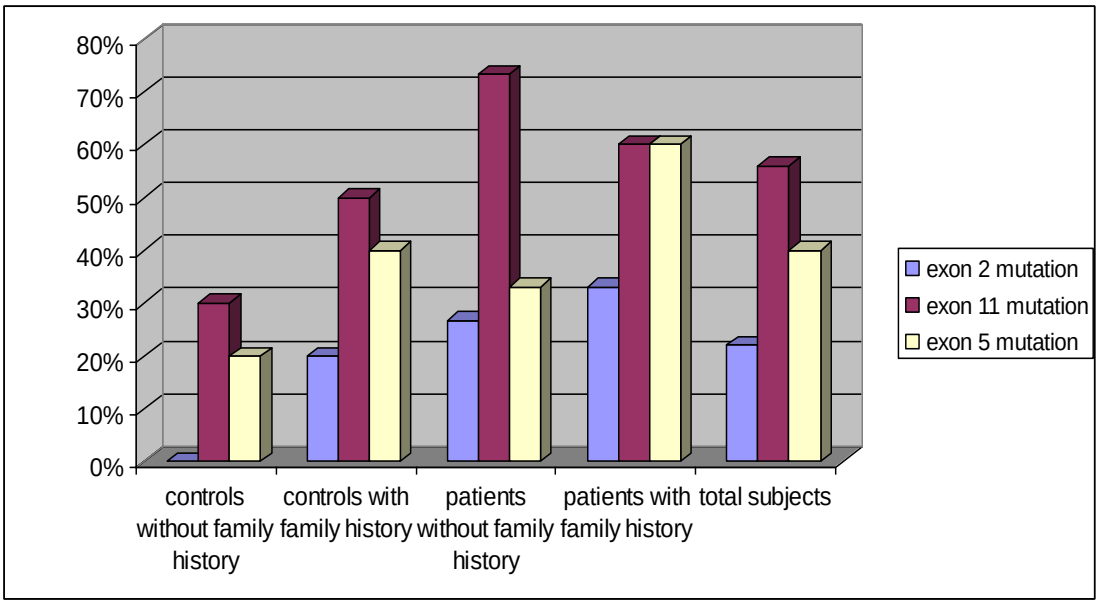


Table (10) & Fig (11): show that, there was a high percentage of mutations in BRCA 1 gene exons among our patients as compared to that of controls , but the most frequent mutation was in exon 11 followed

by exon5 then exon 2. Also we observed the absence of exon 2 mutations in control women who had no family history of breast cancer.

Table (11) : **Allelic frequency of exons 2 , 5 and 11 among control and breast cancer groups**

Groups / Allelic frequency	control		Breast cancer groups		Total	X^2	*p*
	Without family history N =10	With family history N=10	Without family history N =15	With family history N=15			
	Number (%)						
Exon 2:							
• **Wild**	10/10 (100)	8/10 (80)	11/15 (73)	10/15 (66.7)	39/50	8.06	>0.05
• **Heterozygous**	0/10 0	0/10 0	0/15 (0)	2/15 (13)	2/50		
• **Homozygous**	0/10 0	2/10 (20)	4/15 (26.7)	3/15 (20)	9/50		
Exon11							
• **Wild**	7/10 (70)	5/10 (50)	4/15 (26.7)	6/15 (40)	22/50	11.52	>0.05
• **Heterozygous**	3/10 (30)	0/10 (0)	2/15 (13)	2/15 (13)	7/50		
• **Homozygous**	0/10 0	5/10 (50)	9/15 (60)	7/15 (46.7)	21/50		
Exon 5							
• **Wild**	8/10 80	6/10 60	10/15 66.7	6/15 40	30/50	4.44	>0.05
• **Heterozygous**	2/10 20	4/10 40	5/15 33	9/15 60	20/50		

P>0.05: non significant

***Fig. (12)*: Allelic frequency of exon2**

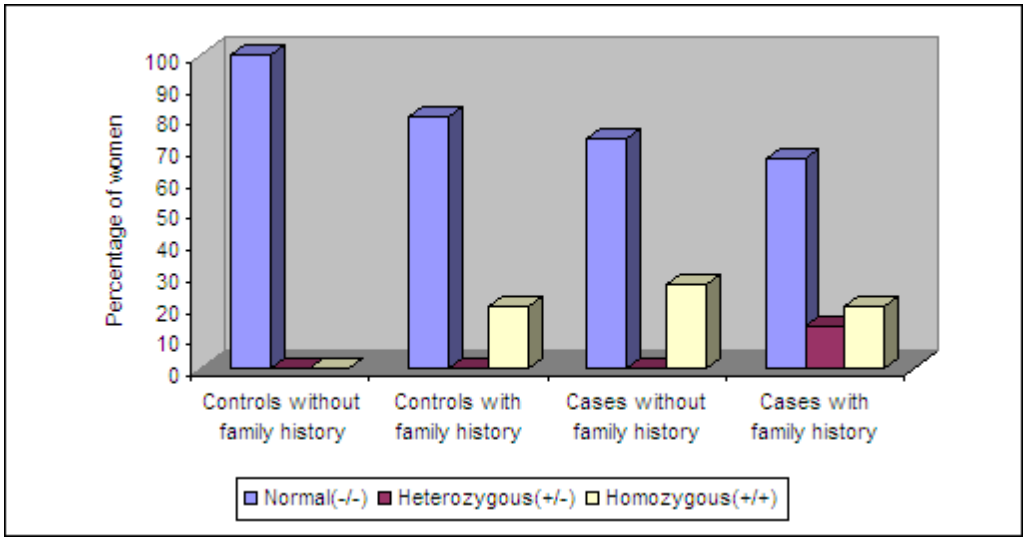


***Fig. (13):* Allelic frequency of exon 5**

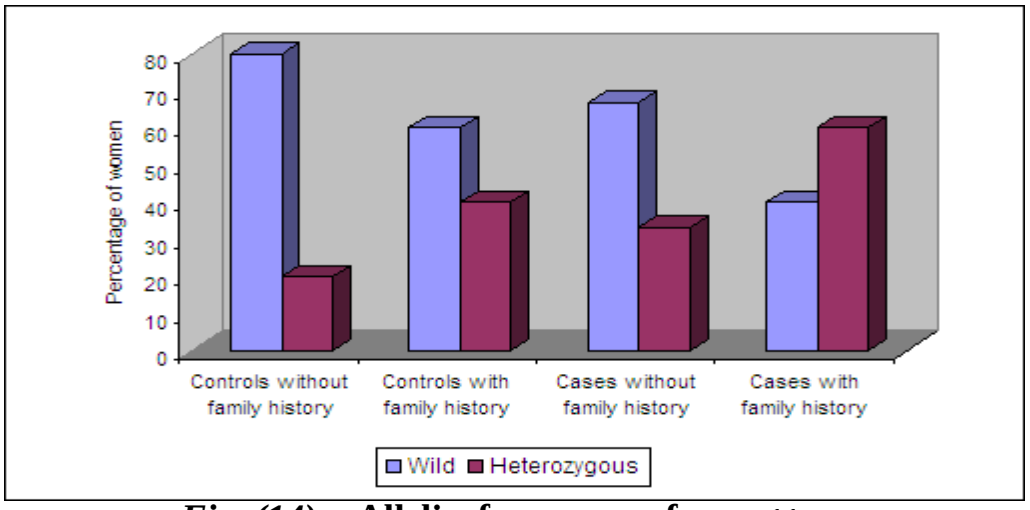


***Fig. (14) :* Allelic frequency of exon 11**

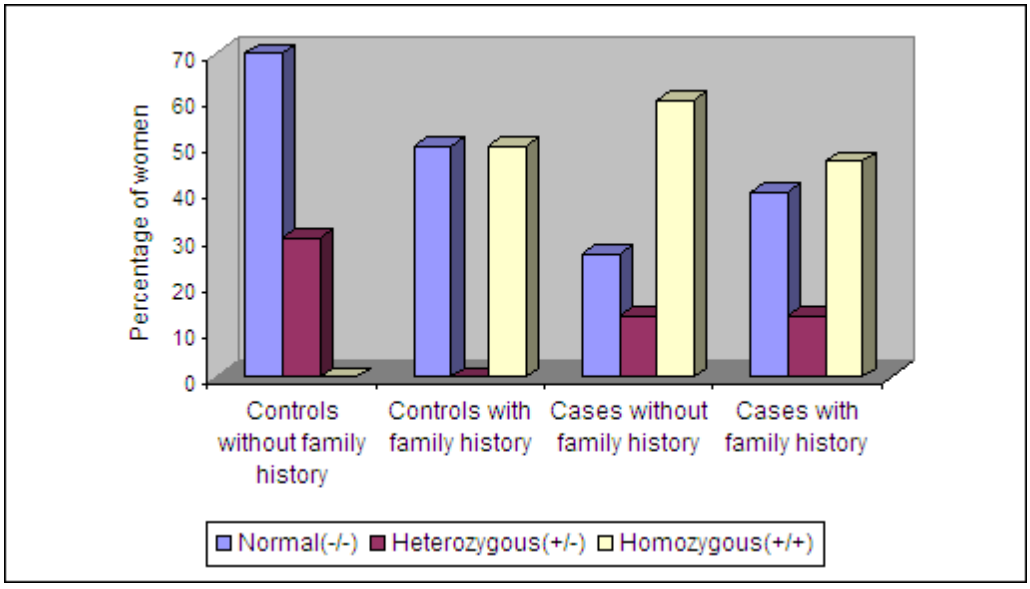
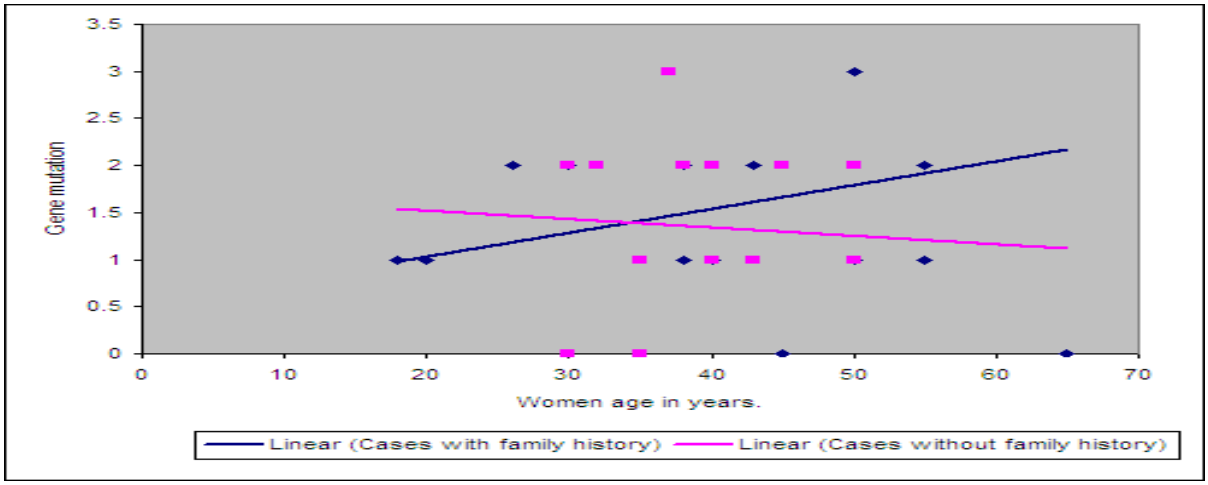


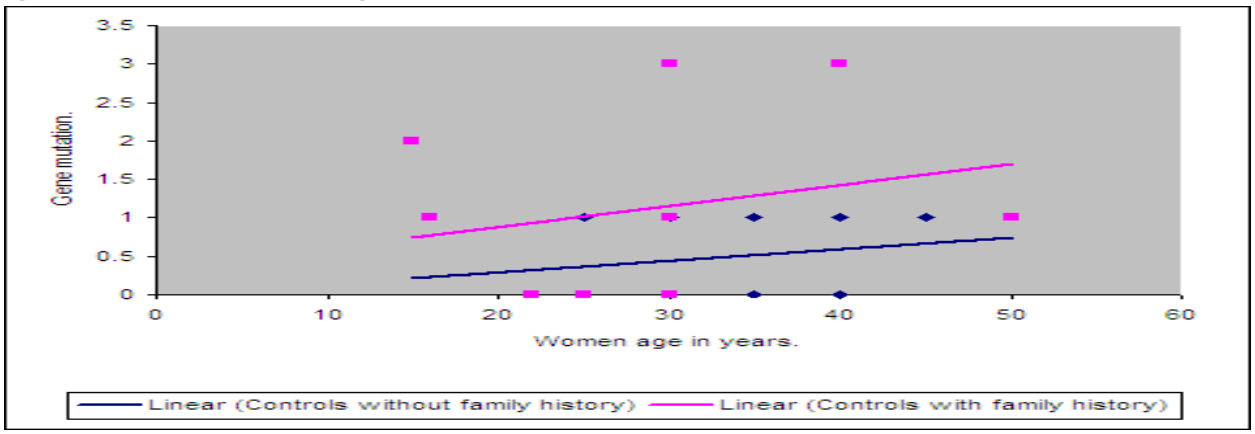
Fig. (15) : **Correlation coefficient (r) between maternal age and frequency of gene mutations in the breast cancer groups**



r= 0.21 p>0.05 r= -0.15 p>0.05

Fig.(15) shows that there were positive correlation between gene mutations and maternal age in the breast cancer group with family history r=0.21 and negative correlation in breast cancer group without family history r=-0.15 but these correlation not reach to significant values
p>0.05

Fig. (16) : **Correlation coefficient (r) between maternal age and frequency of gene mutations among controls.**



r= 0.24 p>0.05 r= 0.17 p>0.05

Figure (16) : shows that there was positive correlation between gene mutations and maternal age in both control groups with and without family history r= 0.24 and r= 0.17 respectively but this correlation not reach to a significant value.