

## Effect of Genetic Counseling on Consanguineous Spouses Attending Maternal and Child Health Centers

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**Abstract:** Consanguineous spouses relation plays an important role in the prevalence of genetic disorders. Genetic counseling is directed towards reducing the number of children born with genetically determined disorders. **Aim:** The study aimed to evaluate the effect of genetic counseling on knowledge of consanguineous spouses' related to genetic disorders and making decision toward reproductive health. **Research design:** This is a quasi-experimental study. **Setting:** This study was carried out at maternal and child health centers affiliated to Ministry of Health in El-Qaliobia governorate. **Sample:** A purposive sample of one hundred spouses with 1st, 2nd or 3rd degree of consanguinity relation who attended the previously mentions MCH centers. **Tools:** Two tools were used for data collection; the first tool was an interviewing questionnaire to collect data about sociodemographic characteristics of spouses; past and present health history of spouse and spouses knowledge related to genetic disorders, genetic counseling and making decision toward reproductive health. The second tool obtained from record review of laboratory investigations regarding to genetic tests. **Results:** This study proved that genetic counseling helps in increasing spouses' knowledge regarding purpose of genetic counseling, genetic disorders, how it will be inherited and the risk of having an affected child. Approximately one third of spouses conducted genetic investigations, 10 Out of 18 Pregnant women received antenatal care (more than half), and very few of them had using family planning methods. The study also showed significant statistical correlations between spouses' performances of genetic investigations and their monthly income. **Conclusion:** The finding of this study showed that genetic counseling increased spouses' knowledge regarding the effects of consanguinity marriage on their offspring's and helps them to make proper reproductive decision through suggestion of reproductive choices. **Recommendations:** The study recommended that, genetic clinics should be accessible in all maternal and child health centers especially in rural areas supplied with necessary facilities and coordinated team work for conducting genetic investigations and genetic counseling program.

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**Keywords:** Consanguinity relation, Genetic disorders, genetic investigations, Genetic counseling.

### 1. Introduction

Consanguineous marriage refers to unions contracted between biologically-related individuals. In clinical genetics, a consanguineous marriage means union between couples who are related as second cousins or closer. In population genetics, consanguinity may refer to union of individuals with at least one common ancestor such as those occurring within small towns and tribes (Serenson and Cheuvront, 2005). Consanguineous marriage is widely favored in large majority of Egyptian population estimates of consanguinity ratios in different parts of Egypt rated from 29% -50%. The highest incidence is that in the rural areas. Research worldwide has indicated that consanguinity could have an effect on some reproductive health parameters such as postnatal mortality and rates of congenital malformation (Mokhtar and Abdel Fattah, 2006). Genetic disorders are diseases that are not acquired or caused from infection or trauma, but rather children inherit the gene from their parents. It could be congenital. It can be seen the disease at birth

immediately but sometimes, if the children have the gene, the disease will develop later on (El-Sobkey, 2007). These disorders include the sickle cell gene, thalassemia, the hemophilia, inborn errors of metabolism and red cell enzymopathies (Vessey and Jackson, 2006). Genetic counseling services have been recommended by the World Health Organization to reduce the prevalence of genetic disorders (WHO, 2008). Genetic counseling is a process of communication between patients and trained professionals intended to provide patients who have genetic disease or risk of such a disease with information about their condition and its effect on their family. This allows patients and their families to make informed reproductive and other medical decisions (Michie et al., 2005).

Nurse as an important member in counseling team, plays a vital role in genetic counseling. She must use genetic knowledge in their clinical practice to know when and how to refer patients and families to experts in genetic health care (Mekussick, 2006).

**Significance of the study:**

Many studies have shown that 30% of admission and about 40-50% of deaths occurring in pediatric hospitals are accounted for children with genetic disorders or congenital malformations (Gomaa, 2007). The genetic disorders are chronic in nature and therefore require continuous support and health care. Consequently, the genetic diseases causes formidable economic and psychosocial burden on the family with negative reflection on the community at large. At the present time, the most effective means of preventing genetic diseases remains the provision of genetic counseling for individuals at risk of having a child with genetic disorders, especially spouses with consanguinity marriage.

#### **Aim of the study:**

The aim of this study is to evaluate the effect of genetic counseling on knowledge of consanguineous spouses' related to genetic disorders and making decision toward reproductive health.

#### **Hypotheses:**

Genetic counseling will improve consanguineous spouses' knowledge related to genetic disorders and help spouses make proper reproductive decision

## **2. Subjects and methods:**

#### **Research design:**

This is a quasi-experimental study was conducted to explore the effect of genetic counseling on consanguineous spouses.

#### **Setting**

This study was carried out at 5% of maternal and child health (MCH) centers affiliated to Ministry of Health in El-Qaliobia governorate which represent 7 centers in rural areas from the total number of 140 MCH centers were chosen randomly for application of genetic counseling program where a large proportion of marriages are contracted between blood relatives.

#### **Sample:**

A purposive sample of one hundred (100) spouses who attended the previously mentioned MCH centers for antenatal follow up or for other MCH services were chosen according to the following inclusion criteria; spouses with 1st, 2nd or 3rd degree of consanguinity relation, at reproductive age from 18 to 49 years and didn't previously attend genetic counseling program.

#### **Tools of data collection:**

**First tool:** An interviewing questionnaire was developed to assess the sociodemographic characteristics and spouses knowledge related to genetic disorders and genetic counseling. It consisted of three parts:

**A)** Sociodemographic characteristics of spouses which include age, level of education, job, consanguinity degree and income.

**B)** Past and present health history of spouses as regards family history, obstetric history, consanguinity degree (were documented in the form of pedigree (Load, et al., 2005), first degree: first cousin; second degree: second cousin; third degree: step or distant relative.

**C)** Spouses knowledge related to genetic disorders and genetic counseling, it was used before and after the counseling program.

Answer of individual knowledge questions were categorized on two-point scale as satisfactory and unsatisfactory. For the total evaluation of knowledge the correct answers was added for every participant and normalized to a value on a 100 point scale. A satisfactory overall knowledge level is considered if the total degree is more than or equal 50; otherwise it was regarded as unsatisfactory knowledge level.

**Second tool:** Data collection from medical record, it contained results of genetic investigations (karyotype test, ultrasound and biological tests), ante natal care and reproductive decision.

Content validity revised by five expertise group from faculty members of Community Health Nursing and specialized physicians in genetic.

#### **Pilot study:**

A pilot study was carried out on seven spouses at El Qualiobia Governorate ;one from each maternal and child health center before embarking on data collection, in order to test the applicability of tools, as well its feasibility. The modification of tools was done to reach to the finalized form. Subjects in the pilot were not included in the main study sample to avoid bias.

#### **Field work:**

Official letters was issued to the directors of selected maternal and child health centers for permission of data collection and conducting the study. Oral consent was secured from each spouse after explaining the aim of the study and ensuring that all information will be used only for research purposes.

Data were collected over nine months (September 2010-June 2011)

**The Genetic counseling construction** has passed through different phases as follow:

#### **The preparatory phase and assessment phase:**

In this phase the researchers' revised current local and international related literature which helped in designing the tools.

The researchers attended the maternal and child health centers from 9.00 am to 1.00 pm for two days/week to collect data till the sample size reached the previously determined number. Based on actual educational needs 'assessment of spouses and guided by relevant literature, counseling sessions were

developed. **The general objective of genetic counseling** was to acquire the consanguineous spouses' knowledge related to genetic disorders and help them to make informed decisions toward reproductive health and genetic screening. At the end of counseling sessions, the spouses will:

- Explain the facts contributing to genetic disorders that may affect offspring.
- Comprehend how heredity affecting offspring.
- Take a decision toward their reproductive health.
- Propose the available management.

**The sessions** included theoretical background about genetic disorders. Every spouse was interviewed for about 45 minutes to fill the tools (pre test). Then the spouses in the study sample were provided by four constructive teaching sessions. Each session took about 45 minutes integrated with teaching points and the researchers before going on to a new topic used questions to check the spouse recall and understanding of the material already covered. Sessions were conducted by the researchers according to **GATHER** approach:

**Greet:** The investigators established relationship with spouses emphasizing the purposes and benefits of genetic counseling according to their tradition and religion. The investigator reassured spouses that what will be said is confidential.

**Ask:** The investigators asked spouses about their sociodemographic circumstances, past and present health status, and reproductive options. Assessing spouse's genetic knowledge and their levels of anxiety separately using questions similar to first tool constructed by the investigators.

**Tell:** The investigators told spouses about nature of genetic disorders and its consequences, why screening, causes of inherited characteristics, risk of recurrence, indications of genetic counseling, genetic investigations, antenatal care, family planning, life style, and places of genetic services in Qualiobeaa Governorate.

**Help:** The investigators helped spouses choose alternatives management as regards antenatal care, family planning and different methods that could be used.

**Evaluate:** The investigators explained different lines of alternatives of genetic harmful factors in their life style that can be modified to reduce the risk of defects of offspring and suggest appropriate referrals.

**Return to follow up visits:** The investigators returned for follow up evaluation through home phone and records in maternal and child health centers.

Different methods of counseling as face to face and interactive counseling with laptop. Using effective media as posters, real objects and a model for reproductive system, taking into consideration

using simple and clear Arabic language to suit the level of all spouses. A booklet was constructed for spouses as an educational reference after counseling implementation. This booklet contains items about purpose of genetic counseling, risk of consanguinity marriage, early detection of risk factors, antenatal care, healthful factors in lifestyle, transmission of genetic traits from parent to children, types of genetic screening tests and community services of genetic counseling.

#### **Evaluation:**

A post test was conducted immediately at the end of counseling sessions using the aforementioned tools to evaluate the effect of counseling on spouses' knowledge while evaluation of spouses decision making regarding to reproductive health, genetic investigations were conducted three months later post counseling through reviewing spouses records and meeting in maternal and child health centers.

#### **Ethical considerations:**

The researchers took into consideration spouses' rights based on their needs, giving complete information, assuring them that confidentiality will be obtained and no harmful effect on them. They had also the right to withdraw from the study at any time.

#### **Statistical Design:**

Data entry and analysis were performed using SPSS version 11.5

The quantitative data were presented using the arithmetic mean, standard deviation, and analyzed using t-test and analysis of variance ANOVA. Qualitative data are presented by the number and percentage and analyzed by chi-square test to assess the interrelationship among variables. Statistical significant was set at  $p < 0.05$ .

#### **3. Results:**

**Table (1)** reveals that the mean age of wives in this study was  $24.7 \pm 5.13$ , 65 % of them in the age category from 20 to less than 30 years and 20% less than 20 years. The result shows also that the husbands' mean age was  $29.5 \pm 6.08$ , about 63% of them in the age category from 20 to less than 30 years and 27% in the age category from 30 to less than 40 years. As regard to educational level, the secondary level had the highest percent (42%, 47%) for wives and husbands respectively. This result shows also that 73% of wives were housewives and all of husbands were working. The result of this study demonstrates that 26% of spouses monthly income was sufficient with a mean  $\pm$ SD  $358.3 \pm 110.06$ . As regards consanguinity degree between spouses 59% were first degree, 29% were second degree and 12% were third degree.

**Table (2)** demonstrates The wives whom their age during first pregnancy less than 20 years were

33.7%,38.2% were aged between 20 to less than 25 years, while 28.1%were aged 25 or more with a mean age 23.0±3.86. The result also demonstrates that 25%of wives had history of spontaneous abortion less than 3times, and 2%of them had history of spontaneous abortion 3 times or more. The result indicates that 57% of wives delivered one to two times and 14% delivered three to four times. In relation to 18 pregnant wives in this study 33.3% were at first trimester, 50% of them at second trimester and 16.7% at third trimester.

**Table (3)** reveals that 50% of spouses had genetic family history, 39% had family history of diabetes mellitus, and 79.4%of them were relatives of second degree and 27.1% of first degree. Family history of Down syndrome was found among 10%, which 17.2%of them were relatives of second degree and 8.2%of the first degree. Family history of renal diseases was found among 9%, which 11.9%of them were relatives of first degree and 6.9% of the second degree. Family history of cystic fibrosis was 7% prevalence, 13.8% of them relatives of second degree. Blindness was common among 7%, 20.7% of them relatives of second degree. Hearing loss and muscle atrophy were found among 5% of family history followed by Epilepsy 4%.Thalassemia, sickle cell and hemophilia were found among 3% of family history.

**Table (4)** shows that 24% of children have genetic disorders, 5%of them have Down syndrome and 16.7%of these were third degree between spouses, 3%with cerebral palsy, 3.4%of these from first and second degree between spouses. This table also reveals that 2% an equal percentage of spouses have children suffering from sickle cell anemia, Juvenile D.M, G-6-PD deficiency, color blindness and hydrocephalus. The least equal percentages representing 1% of children were suffered from Thalassemia, PKU, meningocel, imperforated anus, hemophilia and congenital bilateral hip dislocation.

**Table (5)** indicates that there was improvement in the acquisition of knowledge related to genetic disorders, genetic counseling and investigations, for both wives and husbands post counseling.

**Table (6)** demonstrates the difference in the mean scores and standard deviation of knowledge pre genetic counseling between husbands and their wives was not significant (P > 0.05).However, in relation to post counseling it was statistically significant (P < 0.05). The same table also revealed that the mean score and standard deviation of knowledge between wives and husbands pre-post counseling was statistically significant (P < 0.05).

**Table (7)** shows that 30% of spouses performed genetic investigations. As regards karyotype 16.7%of wives and 10.0% of husbands had abnormal structural

aberrations, 6.7% of husbands had abnormal numerical aberrations and 13.3% of wives had sex chromosome abnormality. As for three dimension ultrasound it was performed for 60.0% of pregnant wives, 6.7% of them had intrauterine growth retardation.3 (10.0%) Of pregnant wives performed amniocentesis which revealed one abnormality representing 3.3%

**Table (1): Percentage distribution of spouses according to their sociodemographic characteristics (n=100).**

Items	Wives		Husbands	
	No	%	No	%
<b>*Age (Years):</b>				
• <20	20	20	2	2
• 20-	65	65	63	63
• 30-	15	15	27	27
• 40-50	0	0	8	8
Mean ±SD	24.7±5.13		29.5±6.08	
<b>*Educational level:</b>				
• Illiterate	26	26	13	13
• Read and write	19	19	18	18
• Secondary education	42	42	47	47
• University	13	13	22	22
<b>*Job:</b>				
• Not working	73	73	0	0
• Working	27	27	100	100
	No		%	
<b>*Consanguinity degree:</b>				
• First degree	59		59	
• Second degree	29		29	
• Third degree	12		12	
<b>*Family monthly income:</b>				
• 250-	74		74	
• 500+	26		26	
Mean ±SD	358.3±110.06			

**Table (2): Percentage distribution of wives according to their reproductive health history (n=100).**

Items	No	%
<b>*Age at first pregnancy (Years): n=89</b>		
• < 20	30	33.7
• 20 -	34	38.2
• 25 +	25	28.1
Range	17-32	
Mean ±SD	23.0±3.86	
<b>*Number of spontaneous abortions :</b>		
• 0	73	73
• < 3	25	25
• ≥ 3	2	2
<b>* Number of deliveries:</b>		
• 0	29	29
• 1-2	57	57
• 3-4	14	14
<b>*Current pregnancy gestational age (trimesters) (n= 18 )</b>		
• First trimester	6	33.3
• Second trimester	9	50
• Third trimester	3	16.7

**Table (3): Percentage distribution of spouses with family history of genetic disorders according to their consanguinity degree (n=100).**

Items	Consanguinity degree						Total 100	
	First (n= 59 )		Second (n= 29 )		Third (n=12 )			
	No	%	No	%	No	%	No	%
Family with genetic disorders							50	50
• Diabetes mellitus.	16	27.1	23	79.3	0	0	39	39
• Down syndrome.	5	8.5	5	17.2	0	0	10	10
• Renal disease.	7	11.9	2	6.9	0	0	9	9
• Cystic fibrosis.	3	5.1	4	13.8	0	0	7	7
• Hearing loss.	2	3.4	3	10.3	0	0	5	5
• Blindness.	1	1.7	6	20.7	0	0	7	7
• Muscle atrophy.	1	1.7	4	13.8	0	0	5	5
• Epilepsy.	0	0.0	4	13.8	0	0	4	4
• Thalassemia.	0	0.0	0	0.0	3	25	3	3
• Sickle cell anemia.	0	0.0	2	6.9	1	8.3	3	3
• Hemophilia.	1	1.7	2	6.9	0	0	3	3

Items are not mutually exclusive.

**Table (4): Percentage distribution of spouses according to their children with genetic disorders (n=100).**

Items	Consanguinity degree						Total 100	
	First (n= 59 )		Second (n= 29 )		Third (n=12 )			
	No	%	No	%	No	%	No	%
Children with genetic disorders and congenital anomalies							24	24
• Down syndrome	2	3.4	1	3.4	2	16.7	5	5
• Cerebral palsy	2	3.4	1	3.4	0	0	3	3
• Sickle cell anemia.	1	1.7	0	0	1	8.3	2	2
• Juvenile D.M	1	1.7	0	0	1	8.3	2	2
• Thalassemia	0	0	1	3.4	0	0	1	1
• G-6-PD deficiency	1	1.7	0	0	1	8.3	2	2
• Color blindness	0	0	2	6.9	0	0	2	2
• PKU	0	0	1	3.4	0	0	1	1
• Meningocel	0	0	0	0	1	8.3	1	1
• Hydrocephalus	1	1.7	1	3.4	0	0	2	2
• Imperforated anus	0	0	1	3.4	0	0	1	1
• Hemophilia	1	1.7	0	0	0	0	1	1
• Congenital bilateral hip dislocation	0	0	1	3.4	0	0	1	1

**Table (5): Percentage distribution of spouses according to their satisfactory knowledge related to genetic disorders, genetic counseling and investigations pre and post counseling (n=100).**

Items	Wives				Husbands			
	Pre		Post		Pre		Post	
	No	%	No	%	No	%	No	%
<b>* Genetic disorders:</b>								
• Risk of consanguinity marriage	17	17	98	98	20	20	98	98
• Genetic problems in consanguineous marriage.	19	19	97	97	9	9	99	99
• Warning from consanguinity marriage.	10	10	98	98	12	12	99	99
• Causes of increasing genetic disorders.	16	16	98	98	13	13	99	99
• Early detection of genetic disorders.	14	14	100	100	10	10	100	100
<b>* Genetic counseling &amp; investigations:</b>	12	12	97	97	7	7	100	100
• Meaning of genetic counseling.	13	13	99	99	5	5	99	99
• Community services of genetic counseling.	14	14	94	94	2	2	99	99
• Indication of genetic counseling.	14	14	95	95	5	5	99	99
• Genetic investigation before pregnancy.	10	10	99	99	14	14	93	93
• Genetic investigation during pregnancy.	22	22	93	93	12	12	98	98
• Management of genetic disorders.								

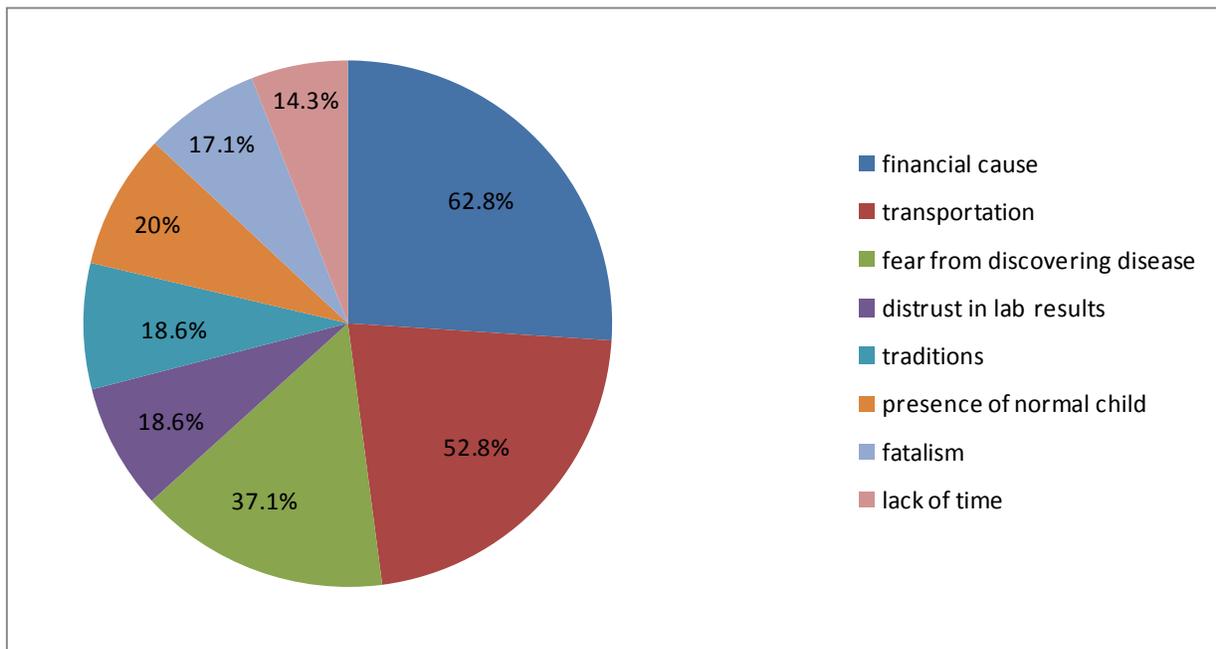
**Table (6): Difference between Knowledge of husbands and their wives pre and post genetic counseling. (n=100)**

Items	Mean	SD	t-test	P-value
<b>*Pre counseling</b>				
• Wives	14.5	29.5	1.33	.183 P> 0.05 NS
• Husbands	9.7	19.6		
<b>*Post counseling</b>				
• Wives	96.4	9.8	-1.98	0.0487 P< 0.05 S
• Husbands	98.8	7.5		
<b>*Post- Pre</b>				
• Wives	81.9	29.7	-1.99	0.0481 P< 0.05 S
• Husbands	89.9	20.4		

**Table (7): Percentage distribution of spouses according to their conducting of genetic investigations post counseling (n=30).**

Items	Wives		Husbands	
	No	%	No	%
<b>*Karyotype:</b>				
• Normal	21	70.0	25	83.3
• Abnormal structural aberrations	5	16.7	3	10.0
• Abnormal numerical aberrations	0	0.0	2	6.7
• Sex chromosome abnormality	4	13.3	0	0.0
<b>*Glucose tolerance test:</b>				
• Normal	26	86.7	29	96.7
• Impaired	3	10.0	0	0.0
• Diabetic	1	3.3	1	3.3
<b>*HB electrophoresis:</b>				
• Normal		86.7		86.7
• Hemolytic anemia	26	13.3	26	6.7
• Sickle cell anemia	4	0.0	2	3.3
• Thalassemia	0	0.0	1	3.3
<b>*Three-Dimension ultrasound of pregnant(n=18)</b>				
• Normal	0	53.3	1	0.0
• Intrauterine growth retardation	16	6.7	0	0.0
• <b>*Amniocentesis: (n=3)</b>	2	6.7	0	0.0
• Normal	2	3.3	0	0.0
• Abnormal	1	3.3	0	0.0

Items are not mutually exclusive.



**Figure (1): Percentage distribution of spouses according to barriers against conducting genetic investigations post counseling (n=70). All items are not mutually exclusive.**

**Table (8): Percentage distribution of spouses according to their decision making toward reproductive health (n=100).**

Decision making	No.	%
<b>*Pregnant wives (n=18)</b>		
• Antenatal care	10	55.6
• Pregnancy termination due to growth retardation	2	11.1
<b>*Use of family planning method</b>	9	9.0

**Table (9-a): Relation between spouses knowledge post genetic counseling and their educational level (n=100).**

Items	Knowledge post genetic counseling		
	Mean	SD	ANOVA
<b>Wife education level</b>			
• Illiterate	88.81	16.52	<b>F = 8.614</b> <b>P = 0.00004</b> <b>P&lt;0.001 HS</b>
• Basic education	99.52	2.08	
• Secondary	98.48	3.97	
• University	100.0	0.00	
<b>Husband education level</b>			
• Illiterate	92.31	19.93	<b>F = 4.13</b> <b>P = 0.008</b> <b>P&lt;0.001 HS</b>
• Basic education	99.49	2.14	
• Secondary	99.81	1.33	
• university	100	0.00	

**Table (9-b): Relation between spouses knowledge pre - post genetic counseling and their consanguinity degree (n=100).**

consanguinity degree	Wives Knowledge post-pre genetic counseling		
	Mean	SD	ANOVA
• First	88.81	16.52	<b>F = 4.15</b> <b>P = 0.0186</b> <b>&lt; 0.05 S</b>
• Second	99.52	2.08	
• Third	98.48	3.97	
consanguinity degree	Husbands Knowledge post-pre genetic counseling		
	Mean	SD	ANOVA
• First	88.81	16.52	<b>F = 0.739</b> <b>P = 0.48</b> <b>&gt; 0.05 NS</b>
• Second	99.52	2.08	
• Third	98.48	3.97	

**Table (9-c): Relation between spouses knowledge pre - post genetic counseling and presence of genetic family history (n=100).**

Genetic family history	Wife's Knowledge			Husband's Knowledge		
	No.	Mean	SD	No.	Mean	SD
<b>Pre</b>						
• Not present	50	13.64	28.7	50	10.0	20.1
• Present	50	15.27	30.48	50	9.45	19.26
<b>t= -0.28 P=0.783 P&gt;0.05 NS</b>				<b>t=0.14 P = 0.89 P&gt;0.05 NS</b>		
<b>Post</b>						
• Not present	50	96.7	10.09	50	99.64	1.80
• Present	50	99.6	9.68	50	98.00	10.44
<b>t = -0.37 P=0.714 P&gt;0.05 NS</b>				<b>t=1.09 P =0.277 P&gt;0.05 NS</b>		
<b>Post-Pre</b>						
• Not present	50	82.36	28.94	50	89.63	20.03
• Present	50	81.45	30.78	50	88.54	21.00
<b>t = 0.15 P =0.88 P&gt;0.05 NS</b>				<b>t = 0.27 P =0.791 P&gt;0.05 NS</b>		

**Table (9-d): Relation between spouses knowledge pre - post genetic counseling and presence of children with genetic disorders (n=100).**

Children with generic disorder	Wife's Knowledge			Husband's Knowledge		
	No.	Mean	SD	No.	Mean	SD
<b>Pre</b>						
• Not present	24	17.05	32.75	24	9.85	19.50
• Present	76	13.64	28.53	76	9.69	19.77
<b>t = 0.49 P = 0.624 P &gt; 0.05 NS</b>			<b>t = 0.03 P = 0.972 P &gt; 0.05 NS</b>			
<b>Post</b>						
• Not present	24	94.70	12.26	24	99.62	1.86
• Present	76	96.89	8.98	76	98.56	8.53
<b>t = -0.95 P = 0.344 P &gt; 0.05 NS</b>			<b>t = 0.6 P = 0.55 P &gt; 0.05 NS</b>			
<b>Post-Pre</b>						
• Not present	24	77.65	32.16	24	89.77	19.38
• Present	76	83.25	29.02	76	88.87	20.86
<b>t = -0.8 P = 0.424 P &gt; 0.05 NS</b>			<b>t = 0.19 P = 0.852 P &gt; 0.05 NS</b>			

**Table (10-a): Relation between spouse's performance genetic investigations and their age and family income post counseling (n=100)**

Items	Performance of genetic investigations			
	Yes(n=30)		No(n=70)	
	No.	%	No.	%
<b>Wife' age (Years):</b>				
• <20	10	33.3	10	14.2
• 20-	14	46.7	51	47.9
• 30-50	6	20.0	9	20.9
<b>Pearson <math>\chi^2 = 6.74</math> df= 2 P=0.034 P&lt;0.05 S</b>				
<b>Husband' age (Years):</b>				
• <30	22	73.3	43	61.4
• 30-50	8	26.7	27	38.6
<b>Pearson <math>\chi^2 = 1.32</math> df= 1 P = 0. 2 53 P&lt;0.05 S</b>				
<b>*Family monthly income (LE):</b>				
• Sufficient	18	60.0	56	80.0
• Not sufficient	12	40.0	14	20.0
<b>Pearson <math>\chi^2 = 4.37</math> df= 1 P = 0.0367 P&lt;0.05 S</b>				

**Table (10-b): Relation between spouse's performance genetic investigations and their education level post counseling. (n=100)**

Items	Performance of genetic investigations			
	Yes(n=30)		No(n=70)	
	No.	%	No.	%
<b>Wife education level</b>				
• Illiterate	6	20.0	20	28.6
• Basic education	5	16.7	14	20.0
• Secondary	16	53.3	26	37.1
• University	3	10.0	10	14.3
<b>Pearson <math>\chi^2 = 2.32</math> df= 3 P=0.508 P&gt; 0.05 NS</b>				
<b>Husband education level</b>				
• Illiterate	3	10.0	10	14.3
• Basic education	4	13.3	14	20.0
• Secondary	17	56.7	30	42.9
• University	6	20.0	16	22.9
<b>Pearson <math>\chi^2 = 1.75</math> df= 3 P =0.6269 P&gt; 0.05 NS</b>				

This table also shows that, the results of HB electrophoresis were 13.3% of wives and 6.7% of husbands had hemolytic anemia. Thalassemia and sickle cell anemia were found only in 3.3% of husbands. As for glucose tolerance test 3.3% of both wives and husbands had diabetes mellitus.

**Figure (1)** reveals that 62.8% of the spouses didn't perform genetic investigation due to financial causes, followed by 52.8% due to far distance, 37.1% due to fear from discovering disease, 18.6% due to traditions and distrust in lab results, 20.0% due to presence of normal child, 17.1% due to fatalism and 14.3% due to lack of time.

**Table (8)** demonstrates that 55.6% of pregnant wives decided ante-natal care, and 11.1% decided termination of pregnancy. This table shows also that 9% of wives used family planning methods.

**Table (9-a)** indicates that the relation between spouses educational level and their knowledge post genetic counseling is highly statistical significant  $P < 0.001$ .

**Table (9-b)** reflects statistical significant relation between consanguinity degrees and wives knowledge pre and post genetic counseling.  $P < 0.05$ . but it was not significant for the husbands.

**Table (9-c)** shows that there were no statistically significant associations between genetic family histories of the spouses and their knowledge pre or post genetic counseling ( $P > 0.05$ ).

**Table (9-d)** demonstrates that there were no statistically significant associations between presence of children with genetic disorders and spouses 'knowledge pre or post genetic counseling ( $P > 0.05$ ).

**Table (10-a)** shows that there were statistical significant relations between spouses genetic investigations and their ages and monthly income at  $P < 0.05$ .

**Table (10-b)** reflects that there were no statistically significant associations between spouses genetic investigations and their educational levels ( $P > 0.05$ ).

#### 4. Discussion:

The present study encompasses 100 spouses with consanguinity relation. It showed that the mean age of wives was  $24.7 \pm 5.13$  years and the mean age of husbands was  $29.5 \pm 6.08$ . Less than half of husbands and slightly more than two fifths of wives had secondary education. The current study finding demonstrated that all of husband and more than one quarter of wives were working. As regards consanguinity degree, more than half of spouses were first degree. The mean monthly income of spouses was  $358.3 \pm 110.06$ . In accordance with these study findings, **(table, 1)**. **Load et al., (2005)** stressed on importance of collecting data about sociodemographic characteristics because some disorders increase in incidence with mothers' age and

the education factor which affect knowledge acquisitions.

The current study findings revealed that, the mean age of wives at first pregnancy was  $23.0 \pm 3.86$  and one quarter of them had a history of spontaneous abortion less than three times. More than half of wives delivered once or twice, and half of pregnant wives were in the second trimester (**table, 2**). An Egyptian study conducted by **Mokhtar and Abdel-Fattah (2006)**, revealed that, consanguinity among couples with repeated abortion constituted 60% with 44% first cousin. In Egypt chromosomal abnormalities are present in nearly half of all spontaneous miscarriages. In accordance with **Abdel Meguid et al., (2005)**, the aggregation of abortions, stillbirth, neonatal and infant deaths is stronger in consanguineous families than those with unrelated marriage. **Ward et al., (2008)**, stated that the reproductive history includes any type of pregnancy loss, spontaneous abortion, stillbirth and prenatal death. For those couples with three or more miscarriages for which no maternal anatomic or physiologic explanation can be found, cytogenic analysis should be considered.

These study findings demonstrated that, first consanguinity degree had eight genetic disorders. Diabetes mellitus was the commonest followed by renal diseases, Down syndrome, cystic fibrosis, hearing loss, blindness, and muscle atrophy. Spouses with second degree relation had ten genetic disorders. Diabetes mellitus was also the commonest followed by blindness, Down syndrome, cystic fibrosis, muscle atrophy, epilepsy, hearing loss, renal disease, hemophilia and sickle cell anemia. Regarding to third consanguinity degree, thalassemia and sickle cell anemia were the two only genetic diseases found (**table 3**). These findings were incongruence with **Mokhtar and Abdel-Fattah (2006)**, who found that family history of the previous genetic disease was present in approximately the majority of the couples, while **Albar (2004)**, reported that first cousins have sixteenth of their genes in common because all individual are carriers of five to seven recessive gene. **Dale et al., (2007)** emphasized that one objective in medical genetic counseling is to identify the family genetic risk. Family tree is a powerful diagnostic tool for this purpose, taking and interpreting a basic family history to identify reproductive risk as an important element in preconception and early antenatal care.

The present study findings showed that about one quarter of the spouses had children with thirteen types of genetic disorders and congenital defects. The most common were Down syndrome representing five percent, followed by cerebral palsy accounting for three percent and equal percentage of

two percent for sickle cell anemia, juvenile diabetes, G6-PD deficiency, color blindness and hydrocephalus. The least equal percentages representing 1% of children were suffered from Thalassemia, PKU, meningocel, imperforated anus, hemophilia and congenital bilateral hip dislocation (**table, 4**).

In accordance with this study results of **Hamame et al.,(2006)**, stated that the rate of children with Down syndrome in some Arab countries exceeds the 1.2 to 1.7 per 1000 typical for industrial countries. This may be related to the relatively high proportion of births to older mothers in the region. Up to 50% of children with Down syndrome in the region are estimated to be born to mothers aged 40 or over. In study conducted by **Reynold et al., (2006)**, spouses had family history of different genetic disorders, genetic investigations revealed chromosomal abnormalities in 15%.

The finding of pre counseling showed that, unsatisfactory knowledge among approximately all spouses even the university educated. On the other hand, post counseling, there was a highly significant increase in spouses' knowledge (**table, 5**). In a study carried out by **Abo-Baker (2008)**, Three quarters of parents had deficit knowledge about genetic disorders; the majority did not know the meaning of genetic disorders and its causes. However, in spite of long term instructions, about one fifth of parents had adequate understanding about nature of the genetic disorders. This could be due to using different methods of counseling as face to face, interactive laptop, discussion and demonstration supported by using real objects, posters, models and handouts which are effective approaches for conveying information.

The study results emphasized a great improvement in the mean score knowledge post counseling for both husbands and their wives (**table, 6**).

In order to achieve screening tests, spouses under study were provided with adequate information regarding purpose, types of testing and Place of genetic investigations. However, the present study findings showed that less than one third only of spouses performed genetic investigations (**table, 7**). As regards ultrasound done for pregnant wives in this study, two out of eighteen had intrauterine growth retardation whereas; one out of three had abnormal amniocentesis. Investigation of HB electrophoresis done for spouses were six out of thirty had hemolytic anemia, one out of thirty husbands had sickle cell anemia and thalassemia. This table showed also that about one third of wives and more than one tenth of husbands had abnormality karyotype. The investigators found that spouses faced difficult

decision about whether to terminate or to continue the pregnancy. This could be due to religion and cultural perception of rural community which refuse prenatal diagnosis and selective abortion. In accordance with **Abdel Meguid et al., (2005)**, in their study about premarital genetic investigation; effect of genetic counseling found chromosomal analysis of cases revealed about 15% of the studied sample with chromosomal abnormalities, either structural aberrations or numerical aberrations or both. These results indicate the importance of chromosomal analysis as part of genetic investigations in premarital counseling to identify couples who may require post conception amniocentesis.

More than two thirds of spouses in this study did not perform genetic investigations which can be attributed to financial causes, transportation, fear from discovering diseases, tradition, distrust in laboratory results, presence of normal child, fatalism and lack of time (**figure,1**). These results were consistency with those of **Saleem et al.,(2004)**, who reported that parental attitude to antenatal diagnosis and pregnancy termination is a consequence of a balance between parental understanding of the disease, its mode of inheritance and the prenatal diagnostic options available on the one hand; and traditional belief in fate, religious, social value and economic factors on the other hand. Genetic services are focused on those who are most able to pay for services.

The present study findings demonstrated that ten of the pregnant wives who received antenatal care, minority terminated their pregnancy due to intrauterine growth retardation (**table,8**) In accordance with this study findings, **Reynold et al.,(2006)**, indicated that good antenatal care is essential to anticipate difficulties and complications of labour and to ensure the birth of healthy baby. Results in this table showed also that a very little percent of the studied sample used family planning methods. This could be due to some of spouses had no children or other problems as spontaneous abortion or still births or desire of large family size.

The present study finding revealed that the relation between spouses educational level and their satisfactory knowledge post genetic counseling was highly significant (**table 9-a**). Incongruent to this results, **Tiller et al., (2005)**, reported that women who reported lower level of education may be most likely to benefit from educational strategies designed to supplement genetic counseling to improve their knowledge level.

Concerning the consanguinity degree, the present study finding demonstrated that there was statistically significant correlation between first degree consanguinity and wives' knowledge pre and post genetic counseling (**table, 9-b**). This may be

attributed to first degree consanguinity associated with many genetic abnormalities which lead to increasing spouses' interest and experiences.

The current study finding showed insignificant associations between genetic family history of the spouses, and their knowledge pre and post genetic counseling (**table, 9-c**).

This study showed statistically insignificant associations between presence of children with genetic disorders and spouses' knowledge pre and post counseling (**table, 9-d**). In a similar study, **Antley and Hartlage (2004)**, found that mothers of children with Down syndrome had a considerable amount of information regarding the nature of condition before counseling that was related to their educational background, but they lacked knowledge on recurrence risks. As for the post counseling results of the mothers, it was revealed that they understood the nature of the condition, understood the recurrence risk, and had a good understanding of both topics.

The finding of this study showed that there were no statistical significant correlation between spouses performance of genetic investigations and their age, or education, (**table 10 a ,b**). These results could be due to that more than quarter of wives were illiterate ,and the highest percent that performed investigations were among secondary level of education. But there was statistical significant correlation with monthly income where the mean of the income was  $358 \pm 110.06$  L.E. as it puts them beyond their capabilities to conduct genetic investigations which are more than 500 L.E.

### 5. Conclusion:

In conclusion, this study proved that genetic counseling helps in increasing spouses' knowledge regarding purpose of genetic counseling, genetic disorders, how it will be inherited and the risk of having an affected child. Also, it helped to have decision for their reproductive health related to antenatal care and family planning. In addition, the study showed significant statistical association between spouses' performances of genetic investigations and their monthly income.

### Recommendations:

The finding of this study projected the need for:

Genetic clinics should be accessible in all maternal and child health centers especially in rural areas due to high incidence of consanguineous marriage- supplied with necessary facilities and coordinated team work for conducting genetic investigations and genetic counseling program about:

- Risk of consanguineous marriage.
- Children with genetic disorders.

- Early detection of genetic disorders.
- Management of genetic disorders.
- Reproductive health, antenatal care and termination of pregnancy.
- Family planning.

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