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Fetal Chromosome Abnormalities and Congenital Malformations: An Egyptian Study

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ABSTRACT

Objective: Our objective were to determine and evaluate the role of genetic counseling and amniocentesis in early detection of chromosomal abnormalities or congenital malformations among women at risk.

Patients and Methods: The study was performed on 784 pregnant women.

Results: The cause for seeking genetic counseling in 22.8% of the study cases was positive family history of CNS malformations, and in 17.9% was chromosomal abnormalities in previous child. Also, the results showed that the indications for amniocentesis in 60.8% were history of having previous child with Down syndrome, and in 15.3% were advanced maternal age.

The results of chromosomal analysis of amniotic fluid samples; 21 cases (19.3%) had chromosomal abnormalities, where trisomy 21 (Down syndrome) was detected in 10 cases (9.2%), unbalanced translocation Down syndrome was detected in 9 cases (8.3%) and one had 46 XX, del (13-q), one had 45, XX, t (13;14) and 2.8% was 46, XX, +21, der (14;21) (q10;q10). The risk of complications of amniocentesis was associated with performing amniocentesis early in pregnancy, and with increased number of attempts.

The results also showed that Multiple congenital anomalies (MCA) represented among 42.2%, congenital malformation of CNS represents 26.6%, congenital malformation of the skeletal system 20%, congenital polycystic kidney 8.8% and pyloric stenosis in 2.2%.

Among the 21 women with abnormal karyotype of amniotic fluid, the decision to terminate the pregnancy was made in 3 (14.3%). Among the 45 cases with abnormal findings suggesting fetal congenital malformation, 16 (35.6%) chose termination of their pregnancy.

In conclusion: Public awareness of the risks and difficulties facing a child with chromosomal anomalies or congenital malformations and the effect on their future health and living is of great importance for acceptance of prenatal screening.

Prenatal diagnosis may affect the reproductive decision after genetic counseling. It is essential that genetic counseling is noncoercive and nonjudjemental. The couples decision (Even if it is different from the counselor's views) should be respected.

Key Words:
Genetic counselling, antenatal screening, amniocentesis.

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INTRODUCTION

Genetic diseases can have devastating effects on both patients and their families alike, and on the community at large. Identifying people at risk of genetic disease will help to decrease the burden of such diseases on families and society. Early recognition also leads to greater success of treatment and improves outcome and prognosis.[1] Using screening testing will signifycantly reduce the impact of these disorders in our populations.[2]

A number of surveys have indicated that globally at least 2 per 1000 neonates have autosomal recessive disorders, 2-10 per 1000 have autosomal dominant disorders, 1-2 per 1000 have X-linked recessive disorders, 6-7 per 1000 have chromosomal abnormalities and about 20 per 1000 have congenital malformations.[3]

In Egypt, among patients with genetic diseases, the frequency of autosomal recessive disorders were 33.6%, autosomal dominant disorders were 13.4%, X-linked disorders were 6.7% and chromosomal abnormalities were 3.4%.[4] Chromosomal aberrations are among the most important causes of congenital malformation and mental handicap. The observed prevalence of Down syndrome among live births in the Eastern Mediterranean Region has been reported to vary from 1.15 per 1000 in the UAE to 2.5 per 1000 in Egypt.[5]

Genetic counseling is the process by which patients at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing and transmitting it and of the ways in which this may be prevented or ameliorated. So that they can take the appropriate decisions about marriage, reproduction and health management.[6]

Prenatal diagnosis is the diagnosis of disease or condition in a fetus before it is born, through amniocentesis and chorionic villus sampling. It provides the most appropriate approach to genetic diseases control and prevention. It can be of immense value, since diagnosis followed by genetic counseling could be preventing the birth of a child with genetic disease.[6] This strategy is prohibited among Muslims communities, where termination of a pregnancy may not be acceptable, because of ethical, psychosocial and religious considerations.[1] However, informing family members may protect future generations if they choose not to have children and allows them time to make arrangements for their care and the care of their family.[7]

OBJECTIVES

To determine the indications of genetic counseling among women at risk for giving an infant with chromosome abnormalities or congenital

malformations who attended the Medical Genetic Center, Ain Shams University.

- To determine the indications of amniocentesis and its complications and the results of chromosomal analyses.
- To examine the parental decisions after diagnosis of chromosomal abnormalities or congenital malformations of their fetuses as regards continuation or termination of the pregnancy.

SUBJECTS AND METHODS

This cohort study was conducted during the period from (1st August 2006 to 31st October 2007) among pregnant women attended the Medical Genetic Center, Ain Shams University. This center is concerned with scientific researches and medical care for patients having genetic problems allover Egypt. The center provides preconception and prenatal genetic counseling for couples at genetic risk. Also, the center provides the prenatal screening through, non-invasive (Ultrasound) & invasive (Amniocentesis and karyotyping for diagnosis of chromosomal abnormalities).

Inclusion criteria:
All pregnant women at genetic risk who attended the outpatient clinic concerning with early detection of genetic diseases and congenital malformation of the fetuses at the Medical Genetic Center, Cairo, Ain Shams University.

Exclusion criteria:
The patient considered illegible if she refused to participate, or had incomplete data.

If a patient was eligible to participate, the researcher explained the purpose of the study and reassured the patient that all data would be kept anonymous and then asked her for participation after taking her written consent.

Tools of the study:

a) *History taking:* At first visit: the patient completed the genetic sheet which include the following data:
- Personal data: age, sex.
- Presence of consanguinity.
- Obtain family pedigree.
- Family history: of any genetic abnormalities.
- Past history of having previous births with genetic disorders.
- Current obstetric history: history of bleeding, drug intake and fever, history of having diabetes mellitus or hypertension.
- Reason for genetic counseling.

Each participant had her own file in the clinic which used for her regular follow up visits and routine antenatal investigations (CBC, RH group, FBS and PP, STORCH screening). Folic acid 5mg were given to all pregnant for the first 12 weeks gestation, and then replaced by iron.

b) *Chromosomal assay:*
(Karyotyping): for both couples.

c) *Ultrasound scanning for fetal congenital structural malformation:* the scanning was done during the follow up for all patients every 4-6 weeks. If a serious fetal defect as anencephaly, severe hydrocephalus, lethal skeletal dysphasia, polycystic kidney or multiple congenital anomalies (MCA) were diagnosed

by U/S and confirmed by 4 D U/S, the couples were advised to terminate the pregnancy and those accepted were referred to the maternity hospital of Ain Shams University.

d) *Counseling:* was done by genetic care providers. They discuss the details about the disorder in question with the parents including:

- Review the details about the disorder in question including.
- Expected course of the disease.
- Management issues, and possible treatments or interventions.
- Pattern of inheritance of the underlying condition.
- Describe risks to family members.
- The importance of prenatal diagnosis of some genetic disorders.

e) *Amniocentesis was done when indicated:* It was performed by the gynecologist after obtaining a written consent from the patient. They were given information about the importance of amniocentesis in prenatal diagnosis of chromosomal abnormalities and its complications.

Method of amniocentesis

Inserting a thin, hollow needle into the uterus and removing some of the amniotic fluid that surrounds the baby. During the procedure, the physician used ultrasound to determine the best location for placing the needle and to check for any signs of fetal abnormalities, the baby's heartbeat, to determine the position of the baby and of the placenta, to examine closely the main fetal structures, and to double check the baby's gestational age. Then he inserts a thin needle through the abdomen and uterus into the amniotic sac. About one mm for every gestational week of fluid were withdrawn then the needle was removed. Sometimes, many attempts were tried to give a fluid sample. After the sample is taken, the physician uses ultrasound to check that the fetal heartbeat is normal. The entire procedure takes just a few minutes.

- If the patient was RH negative, she must be taken anti-D IM injection. They were advised to take another one after delivery or after taken another sample if the culture was failed.
- The patient was under supervision for few hours to be sure that the procedure was successful. If a miscarriage occurred, a suitable management has been taken.

Testing the amniotic fluid for chromosomal abnormalities:

- After taking the sample of amniotic fluid, these cells were cultured in a laboratory for one to two weeks, and then tested for chromosomal abnormalities. Test results usually are available within 3 weeks by conventional chromosomal study.[8]
- After obtaining the results, those patients with abnormal results and their husbands were invited again to counseling: The staff explained to them the risk of keeping the pregnancy and the possible methods of management of the condition and the places that provide the heath service for the incoming child. The couples were informed that the center provides rehabilitation clinic for children with genetic disorders or with mental retardation as down sy-ndrome. Then, they were helped to make their own decisions.

Then, the parental decisions were examined by completing a questionnaire contains some possible factors that affect their decision for either continuation or termination of pregnancy.

f) *Follow up of the patients:*
All patients were followed till termination of their pregnancies (Either by normal delivery, delivery of genetically abnormal infants, spontaneous or induced abortions, intrauterine fetal deaths, still births). The outcomes of pregnancies were recorded. The data was obtained through regular visits follow up.

RESULTS

A total of 784 pregnant women attended the outpatient clinic for early detection of genetic diseases and congenital malformation of the fetuses at the Medical Genetic Center Ain Shams University for genetic counseling. Amniocentesis were done for 109 women who needed chromosomal analysis.

Table 1: Characteristics of the study group.

	Frequency	Percent
Age group:		
• 20-	72	9.2
• 25-	159	20.2
• 30-	323	41.3
• >=35	230	29.3
Education level:		
• Illiterate	240	30.6
• Primary school	195	24.9
• Preparatory school	107	13.6
• Secondary school	201	25.6
• University	41	5.3
Consanguinity:		
• Positive	140	17.9
• Negative	644	82.1
Family history		
• Positive	175	22.3
• Negative	609	77.7
Total	784	100.0

Table 2: Indications for genetic counseling among the pregnant women at genetic risk.

	Frequency	Percent
1.history of chromosomal abnormalities: (120) (15.3%)	120	15.3
2.history of congenital malformation		
a-CNS causes: (179) (22.8%)		
hydrocephalus	53	6.8
microcephaly	33	4.2
anencephaly	31	4.0
mental retardation	25	3.2
meningocele	22	2.8
Dandy Walker malformation	10	1.3
Agenesis of corpus calosum	5	0.6
b-Developmental causes (140) (17.9%)		
multiple congenital anomalies	108	13.8
cleft lip	15	1.9
cleft palate	17	2.2
c-Chondro-osseous defects: (59) (7.5%)		
achondroplasia	25	3.2
osteogenesis imperfecta	15	1.9
limb anomalies	10	1.3
skeletal dysplasia	3	0.4
hypochondroplasia	3	0.4
dislocation of the hip	3	0.4
d-other causes:(154) (19.6%)		
Congenital malformation of the respiratory system	45	5.7
Congenital heart disease	35	4.5
Metabolic disorders	15	1.9
Congenital polycystic kidney	12	1.5
Congenital malformation of alimentary tract	11	1.4
Duchenne muscular dystrophy	9	1.1
Genetodermatosis	9	1.1
Ambigious genitalia	5	0.6
Congenital eye anomalies	4	0.5
Deaf mutes	4	0.5
Speech defect	3	0.4
Malignancies	2	0.3
3.history of serious reproductive outcome (127) (16.2%):		
repeated abortions	51	6.5
repeated neonatal deaths	45	5.7
still births	31	3.9
Total	784	100.0

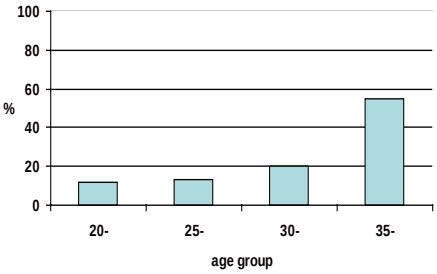

Fig. 1: Distribution of age among women required amniocentesis. Most women (66/120) who required amniocentesis were above 35 years (54.7%).

Table 3: Indications for amniocentesis.

	Frequency	Percent
1- Previous child with Down syndrome	73	60.8
2- Old maternal age > 35	19	15.8
3- Family history of Down Syndrome with anxious mother	10	8.3
4- Abnormal finding in U/S (fetal malformation)	10	8.3
5- History of repeated abortions due to chromosomal aberrations	8	6.7
Total	120	100.0

* 11 women didn't come in the proper time for amniocentesis.

Table 4: Results of chromosomal analysis of the amniotic fluid.

Karyotype	No.	%
Normal karyotype	88	80.7
Male (46,XY)	55	50.5
Female (46,XX)	33	30.2
Karyotype with numerical aberrations	**10**	**9.2**
Male (47,XY+21)	4	3.7
Female (47,XX+21)	6	5.5
Karyotype with structural aberrations	**11**	**10**
Translocation	10	9.2
Balanced [45,XX,t (13;14)]	1	0.9
Unbalanced	9	8.2
Male	3	2.7
46,XY, +21, der (15;21) (q10;q10)	2	1.8
46,XY, +21, der (21;21) (q10;q10)	1	0.9
Female	6	5.5
46,XX, +21, der (14;21) (q10;q10)	3	2.8
46,XX, +21, der (21;21) (q10;q10)	2	1.8
46,XX, +21, der (13;21) (q10;q10)	1	0.9
Deletion [46,XX,del (13q$^-$)]	1	0.9

Table 5: Factors affecting the occurrence of complications of amniocentesis.

	Occurrence of complication No. (%)		Absence of complication No. (%)		Total
Time of amniocentesis:					
- <14 weeks	12	(36.4)	21	(63.6)	33
- 14-18 weeks	3	(3.9)	73	(96.1)	76
Types of tab					
- Clear	5	(6.0)	78	(94.0)	83
- Bloody	10	(38.5)	16	(61.5)	26
Attempt:					
- 1st	1	(2.0)	47	(98.0)	48
- 2nd	3	(9.7)	28	(90.3)	31
- 3rd	7	(31.8)	15	(68.2)	22
- 4th	4	(50.0)	4	(50.0)	8

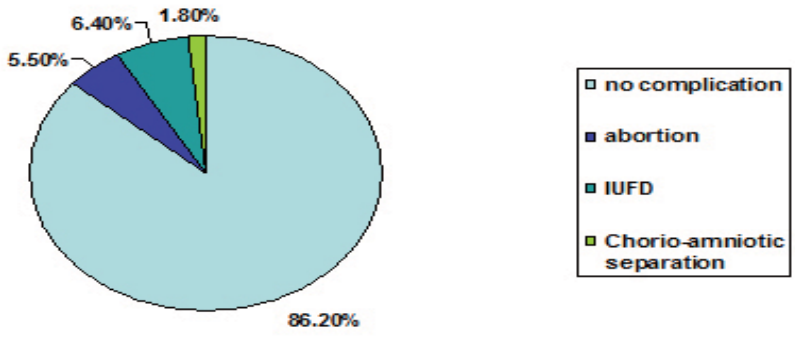

Fig. 2: The frequency of complications of amniocenesis.

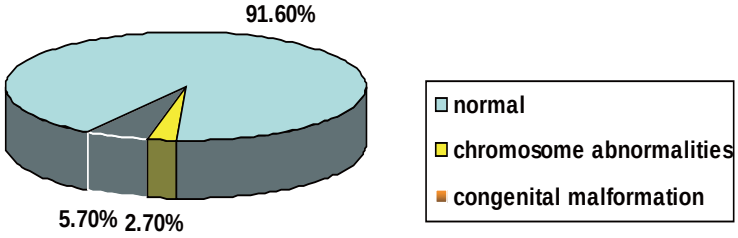

Fig. 3: The frequency of fetal chromosome abnormalities and congenital malformation detected by antenatal screening.

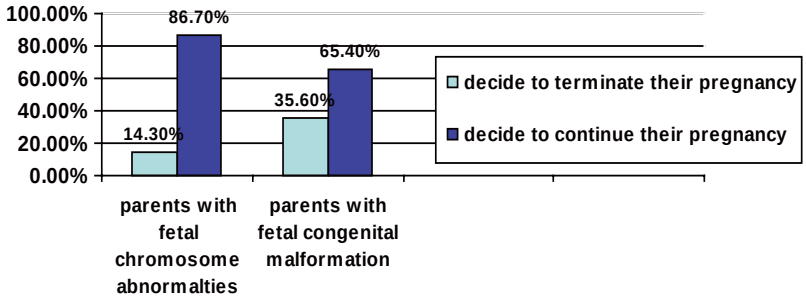

Fig. 4: Among the 21 women with abnormal karyotype of amniotic fluid, the decision to terminate the pregnancy was made in 3 (14.3%). Among the 45 cases with abnormal findings suggesting fetal congenital malformation, 16 (35.6%) chose termination of their pregnancy.

Table 6: Types of fetal congenital malformation that detected by antenatal screening among the study group.

	No of Cases	Percent
Multiple congenital anomalies (MCA):	19	2.4
Single defect:		
***Congenital malformation of central nervous system (CNS):**		
-hydrocephalus	5	0.6
-anencephaly	4	0.5
-meningocele	2	0.3
-Dandy Walker malformation	1	0.1
***Congenital malformation of skeletal system:**		
-Lethal skeletal dysplasia		
-achondroplasia	3	0.4
-limb anomalies	3	0.4
-osteogenesis imperfecta	2	0.3
***Pyloric stenosis**	1	0.1
***Congenital polycystic kidney**	1	0.1
	4	0.5
Total	**45**	**100.0**
Total no of Cases with congenital malformation	**45/787**	**5.7%**

N.B.: Total number of cases detected with congenital malformation 45/787 (5.7% of the study group). Where multiple congenital anomalies (MCA) represented among 42.2% (19/45), congenital malformation of (CNS) represents 26.6% (12/45), congenital malformation of the skeletal system 20% (9/45), congenital pyloric kidney 8.8% (4/45) and pyloric stenosis in 2.2% (1/45).

Table 7: Factors affecting the parental decisions to continue or to terminate their pregnancies inspite of presence of chromosomal abnormalities or congenital malformations.

Decided to continue		
- Due to religious causes	38	80.9
- Wanted baby even with handicapping	9	19.1
- **Total**	**47**	**100.0**
Decided to terminate the pregnancy		
- Due to sad feelings to have another baby with the same problem	8	42.1
- Due to economic causes	6	31.6
- For benefit of our fetuses	5	26.3
- **Total**	**19**	**100.0**

DISCUSSION

Prenatal diagnosis of fetal disorders is a very wide range of noninvasive and invasive methods. In this study, we includes ultrasound examination as an non-invasive tool for detecting structural abnormalities and performed prenatal amniocentesis to obtain definitive diagnosis of chromosome disorders in high risk pregnancies.

The indications of genetic counseling among the 784 women in this study were: 22.8% of them had previous family history or history of having child with the congenital malformation of CNS, 13.8% having previous child with the of multiple congenital malformations, 16.2% due to previous history of serious reproductive outcome, 15.3% for prenatal diagnosis of chromosome abnormalities.

In India, Verma et al.[9] reported that out of 3500 subjects provided genetic counseling at a tertiary genetic center in India, 28.7% were for prenatal diagnosis for chromosome abnormalities, 13.7% for mental retardation±malformations, 11.5% for thalassemia, hemophilia and leukemia, 8.5% for neural tube defects and other malformations, and 8% for muscle dystrophy and spinal muscle atrophy.

The difference in result between two studies raises up the role of genetic sonography and amniocentesis in the detection of the major and minor fetal malformations which may associated with chromosomal abnormalities in our country.

Amniocentesis is the most common prenatal test used to diagnose a large

number of genetic and chromosomal abnormalities in the fetus. Amniocentesis is offered when there is an increased risk of chromosomal or genetic birth defects, or certain malformations.[10]

In this study, the indications for amniotic fluid among the 109 pregnant women at risk for chromosome abnormalities, 73 of them (60.8%) due to history of having previous child with Down syndrome, 19 (15.8%) were due to advanced maternal age, 10 (8.3%) due to Family history of Down Syndrome, 10 (8.3%) due to Abnormal finding in US and 8 (6.7%) due to history of repeated abortions due to chromosomal aberrations. This is differ than the findings of Turhan et al.[11] where they observed that the indications of amniocentesis among the 131 genetic amniocentesis performed in Turkey were: advanced maternal age>35 in 24 (18.3%), suspicion of genetic abnormality on ultrasound in 15 (11.5%), history of siblings with down syndrome in 2 (1.5%) and abnormal triple screen in 90 patients (68.7%), respectively. Also, Verma et al.[9] reported that indications for amniotic fluid studies (n=835) were advanced maternal age (35.7%), high risk result on triple test (21.3%), previous child with trisomy 21 (21.3%) and abnormalities seen on ultrasound (11.1%).

Although, most of our sample required amniocentesis was above 35 years (54.7%), only 19 (15.8%) of them asked to do prenatal diagnosis due to their old age.

However, our finding is not consistent with Preis et al.[12]. They analyzed all the 721 amniocenteses carried out in the Department of Obstetrics in Gdansk in 1996-2002. Amniocenteses were performed due to: advanced maternal age in 553 (76.7%) cases, fetal malformation in current pregnancy in 39 (5.4%) cases, inherited disease in previous pregnancies in 80 (11.1%) cases, maternal balanced translocation in 6 (0.83%) cases, psychological reasons in 15 (2.1%) cases, inherited diseases in the family in 8 (1.1%) cases and serious obstetric history in 9 (1.2%) cases, abnormal results of triple test in 11 (1.5%) cases. Nassar et al. published that[13] out of the 1,347 genetic amniocentesis performed at a tertiary care institution in USA, the most common indications were advanced maternal age (72.3%) and abnormal triple screen (20.3%). The difference may be explained by the difference in educational level, social and cultural factors.

Amniocentesis is reasonably safe and the risk of losing a baby is nearer 1 in 1600 than the more traditionally cited 1 in 200.[10]

In the current study, pregnancy losses were observed in (13.8%) of our cases: where abortion, IUFD and Chorioamniotic separation represented by (5.5%), (6.4%), (1.8%), respectively. These complications of amniocentesis were higher than reported in the previous studies. According to the Centers for Disease Control and Prevention (CDC), 1995[14], the rate of miscarriage is between one in 400 and one in 200 procedures. The risk of miscarriage was 2.6% after early amniocentesis, compared to 0.8% after secondtrimester amniocentesis.[15] A systematic review of complications related to genetic amniocentesis was done by Mujezinovic and Alfirevic.[16] They searched the MEDLINE database

for articles published after January 1st, 1995. Pregnancy loss was 1.9% for total pregnancy loss. Cavallotti et al.[17], found that the miscarriage incidence was 1.7% for amniocentesis. While the total fetal loss rates including spontaneous abortions and intrauterine fetal deaths/still births were 2.3 and 2% in the study groups, in Turkey.[18] The difference may be explained by to some extent to lack of strict measure of infection control during the procedure which may increase the susceptibility to infections.

Our results showed that the risk of complications is significantly associated with: performing amniocentesis early in pregnancy, with bloody fluid attempt and with increasing the number of attempts. These findings was consistent with Cavallotti et al.[17] where they found that the incidence of miscarriage increases with bloody fluid in comparison to clear attempt. However, Müngen et al.[18] reported there was no statistically significant difference in fetal loss rate between women requiring two needle insertions to obtain amniotic fluid and those having only one insertion (p=1.00; OR, 0.75; 95% CI, 0.10 to 5.53).

Amniocentesis has an accuracy rate of between 99.4 and 100% in diagnosing chromosomal abnormalities.[15]

In our study, among the 109 women examined by amniocentesis, 21 cases (19.3%) had chromosomal abnormalities.

Nassar et al.[13] reported that an abnormal karyotype was detected in 34 (2.5%) fetuses from 1,347 genetic amniocentesis performed at a tertiary care institution in USA. Quadrelli et al.[19] reported that Chromosomal anomalies were found in 2.16% of studied cases included: Down syndrome, aneuploidies in which a severe prognosis was expected. The difference between the results of our study and the previous two studies may be due to difference in sample size, and this shows the importance of considering fetal karyotype in early detection of fetal malformation.

After obtaining the abnormal results, the genetic counselors discussed with the parents their options and they helped to make their own decisions as regards continuation or interruption of pregnancy.

The couple was given the choice of abortion when a serious congenital or hereditary condition is discovered. If a fetus has a condition for which prenatal treatment is not yet possible, prenatal diagnosis may help parents to prepare emotionally for the birth and to plan how to take care of this child in the rehabilitation center.

Acceptance of prenatal diagnosis and termination of pregnancy in the case of an affected fetus may vary from one country to another, depending on the health system, religious belief, cultural and educational backgrounds of the population.

In the current study: Among the 21 women with abnormal karyotype of amniotic fluid, the decision to terminate the pregnancy was made in 3 (14.3%). Among the 45 cases with abnormal findings suggesting fetal congenital malformation, 16 (35.6%) chose termination of their pregnancy. These

findings were not consistent with other studies in developed countries. Where Quadrelli et al.[19] reported that when the parents faced with an anomaly such as Down syndrome and aneuploidies in which a severe prognosis was expected, 89% and 96% of them, respectively, decided to terminate the pregnancy although termination of pregnancy is not legally available in Uruguay. Drugan et al.[20] reported that 93% of patients with severe prognosis opted for pregnancy termination. In Israel, Zlotogora[21] examined the decisions of 1467 among both Jews and non-Jews women who had an abnormal result after an invasive prenatal test The main factor in the decision to terminate or continue the pregnancy is the severity of the disorder diagnosed. However, among Arabs other factors are important, in particular the time at which the diagnosis is made.

Vincent et al.[22] examined 378 singleton pregnancies in which a cytogenetic abnormality was reported and for which information regarding pregnancy outcome was available, the decision to terminate the pregnancy was made in 276 (73.02%). Pregnancies involving the most common autosomal trisomies (21, 18, and 13) were terminated at a rate of 92% to 95%. Vincent et al.[22] Kramer et al.[23] examined 145 cases with prenatal diagnosis of Down syndrome, they found that 19 (13.1%) of women chose continuation of pregnancy, while 126 (86.9%) chose termination. Patients who chose termination were significantly older and earlier in gestation than those electing to continue their pregnancy.

In the UK, four hundred and twenty white and Pakistani women living were surveyed about their attitudes to prenatal testing and termination for 30 different fetal conditions. Pakistani women held more favorable attitudes to prenatal testing, but less favorable attitudes to termination than their counterparts.[24] Both groups were most in favour of termination for the same four conditions: anencephaly, trisomy 13 or 18, quadriplegia, Duchene muscular dystrophy. The rank ordering of conditions was also similar.[1]

In Lebanon, Zahed et al.[25] interviewed 90 couples at risk for a variety of genetic disorders, in order to assess their acceptance of prenatal diagnosis and the variables that might influence their choice. Overall, 54% of couples said they would request diagnosis in their next pregnancy, while 26% were opposed to such a procedure. In 87.5% of cases, the reason for refusal was because of religious conviction against termination of pregnancy. Refusal of prenatal diagnosis was also related to a lower socioeconomic background and poorer education.

The acceptance rate of invasive procedures among highrisk group of pregnant Saudi Arabian women to prenatal screening for chromosomal anomalies was (34.2%) probably because as it carries the risk of abortion. On the other hand, 29.1% of the women did not accept the idea of screening; the main reason was that they did not accept termination of pregnancy as a treatment option.[26]

This strategy is used in several countries, but among Muslims, the termination of pregnancy is prohibited and hence such as strategy raises several ethical issues.[27]

The fatwa of the Islamic Jurisprudence Council of the World Islamic League at its 12th session (10-17 February, 1990) in Mecca, agreed by a majority vote to allow for the option of abortion under certain specific conditions. The fatwa determined that an abortion may take place only if a committee of specialized, competent physicians has decided the fetus is grossly malformed, and that its life would be a calamity for both the family and itself. The malformation must be untreatable, unmanageable and very serious, and the abortion may only be carried out prior to the 120th day of conception (Computed from the date of fertilization, not the last menstrual cycle). On the basis of this fatwa, abortions of serious congenital disease are carried out in the hospitals of Saudi Arabia.[28]

So, we recommended encouraging couples to undergo premarital examination for infectious and hereditary diseases and also discouragement of close-relation consanguineous marriage in our communities and good genetic counseling as primary prevention level. Secondary prevention entails either the prevention of the birth of affected babies through prenatal diagnosis and selective abortion.

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