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PREVENTION OF THALASSEMIA;

ROLE OF CHORIONIC VILLOUS SAMPLING TECHNIQUE MAJOR IN FAMILIES WITH THALASSEMIA POSITIVE HISTORY

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ABSTRACT… Objectives: The study was conducted to determine role of CVS in prevention of thalassemia and its sensitivity and specificity in those families with thalassemia positive history. **Period:** Three months. **Study Design:** Cross sectional study. **Material and Methods:** 130 women who had thalassemic children and had come to transfusion centers for blood transfusion. Questionnaire was used for interview. **Results:** out of the total 130 women, 101 were aware of CVS technique in which 50 had CVS. In 50 CVS, 15 had positive result while 35 had negative result. In 15 positive cases 11 had abortion while four positive women refused to abort. Out of the four women who completed their term, three were true positive while one case was false positive. In 35 negative case results, 32 were true negative and three were false negative. **Conclusion:** Thalassemia is the commonest genetic disorder worldwide. It is transmitted from those parents who are carrier of thalassemia. Chorionic villous sampling plays important role in prevention of thalassemia during 1st trimester of pregnancy.

Key words: Thalassemia, Chorionic villous sampling (CVS), sensitivity, specificity

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INTRODUCTION

Thalassemia was first described by cooley in 1915.[1] It is the commonest genetic disorder worldwide. It is an autosomal recessive disorder in which one or both chains of hemoglobin are either missing or deficient.[2] Worldwide 60,000 thalassemia carriers are born each year. More than 5 % of the world population is carrier of thalassemia.[3] In Pakistan about 60000 children are suffering from thalassemia. About 5 % of our population is carriers of thalassemia.[4] It is mainly transmitted to those children whose parents are 1st degree cousins. Thalassemia is non-curable but preventable disease through premarital screening or genetic studies during 1st trimester of pregnancy.[5] Chorionic villous sampling technique is widely used test for in vitro diagnosis of thalassemia in those parents who are carriers of thalassemia or who have one or more children with thalassemia and plan to have further children.[6] Consanguineous marriages are practiced throughout the world with different ethnicity and sociocultural background.[7] Highest prevalence of consanguineous marriages is found in North Africa, Middle East and central and south Asia.[8] Awareness about prenatal diagnosis of thalassemia major is limited in general population.[9] Objective of the study was to determine role of CVS technique in prevention of thalassemia and sensitivity and specificity of this technique.

MATERIAL AND METHODS

It was a cross sectional study conducted in Frontier, Hamza and Fatmid transfusion centres of Peshawar in three months from January 2016 to March 2016. Total 130 mothers were included in the study through convenience sampling technique. Questionnaire was used to collect data regarding frequency of consanguinity, knowledge and practice of chorionic villous sampling technique and sensitivity and specificity of CVS. Women with one or more Thalassemic children were included while those women who were unwilling to appear in the interview or having children with other genetic disorders were

excluded from the study.

RESULTS

Total 130 mothers were interviewed and the data was analyzed in the following tables.

Total mothers	Cousin marriages	Non-cousin
130 (100 %)	99 (76.15 %)	31 (23.84 %)

Table-I. Frequency of consanguinity

Total mothers	Aware	Unaware
130 (100 %)	101 (78 %)	29 (22 %)

Table-II. Frequency of awareness about CVS in Mothers

Source	Number	% age
By physician	89	88 %
Family members	05	5 %
Friends	05	5 %
Media	00	0 %
Other sources	02	2 %

Table-III. Sources of awareness

Out of the total 130 women who were interviewed, only 50 (38.46 %) had undergone CVS.

Sensitivity of CVS calculated from the above data was 75 % while specificity was 91 %.

<table>
<tr><th colspan="4">Total subjects who went for CVS = 50</th></tr>
<tr><td colspan="3">Positive CVS results = 15</td><td>- ive CVS results = 35</td></tr>
<tr><td rowspan="2">Did abortion = 11</td><td colspan="2">Didn't abort = 4</td><td rowspan="2">True –ive after birth = 32</td><td rowspan="2">False +ive after birth = 03</td></tr>
<tr><td>True +ive after birth = 03</td><td>False +ive after birth = 01</td></tr>
</table>

Table-IV. CVS results

DISCUSSION

Total 130 women who had thalassemia children were interviewed. About 76.15 % children had parents with cousin marriages which is almost identical to an international study.[10] Out of the total mothers who were interviewed, 101 (77.69 %) mothers were aware of chorionic villous sampling (CVS) technique as a tool of prevention of thalassemia in their future children. Most of the mothers were counseled by their physicians sitting in the respective transfusion centers while no mother was aware through media. Of the total 101 aware mothers only 50 (38 %) underwent CVS during their 1st trimester of pregnancy. Socio cultural factors were the main obstacles followed by religious reasons for not opting for CVS for prevention of thalassemia in their future children. CVS technique had a sensitivity of 75 % and specificity of 91 %. Low sensitivity in our study may be due low number of cases who did not go for abortion or it may be due to the fact that only limited centers are available for CVS technique in Pakistan. Results of CVS technique are compatible with international studies.[11]

CONCLUSION

Prevalence of thalassemia is high in Pakistan which can be prevented using premarital screening programs or CVS technique during 1st trimester of pregnancy in those women who have thalassemic children or those couples in which both the partners are carriers of thalassemia. Awareness programs need to be established to give awareness about thalassemia.

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"When my absence doesn't affect your life, then may presence has no meaning in it."

Unknown

AUTHORSHIP AND CONTRIBUTION DECLARATION

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1	Dr. Romana Ayub	Main cincept and study conduction	
2	Hayat Muhammad Khan	Data collection & entry	
3	Dr. Zia ur Rehman	Data collection	
4	Dr. Junaid Ahsan	Discussion & results interpratation	
5	Dr. Rubina Gul	Methodology & write up	
6	Dr. Uzair Khan	Data collection	
7	Dr. Shama Khan	Data collection	