CHORIONIC VILLOUS SAMPLING TECHNIQUE; AWARENESS AMONG THE PARENTS OF THALASSEMIA CHILDREN AND ITS ROLE IN THE PREVENTION OF THALASSEMIA MAJOR

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Abstract

Objective: *To determine the awareness level of parents of Thalassemia children regarding CVS technique and the role of this technique in the prevention of thalassemia.*

Materials & Methods: *It was a cross sectional study conducted in Frontier blood transfusion services Peshawar on 130 subjects selected through a convenient sampling technique within six months from Jan; 2018 to Jun; 2018. A questionnaire was used for the collection of data. Those women who had at least one Thalassemia major child were included while women unwilling or having children with other genetic disorders were excluded from the study. Data were analyzed using SPSS version 20.*

Results: *Of the total 130 women, only 50 underwent CVS. The sensitivity of the test was 75 % while specificity was 91 %. Of the total 76 % were cousin marriages while 24 % were non-cousin marriages. Of the total 101 mothers were aware while only 29 mothers were unaware of the technique. The main source of awareness was the physicians who referred them for the procedure.*

Conclusion: *The general public and parents of thalassemia children have limited knowledge about the prevention of thalassemia through the CVS technique. Diagnostic centers need to be established at the divisional level.*

Keywords: *Chorionic villous sampling (CVS), β – thalassemia, Premarital screening, Sensitivity, and Specificity.*

INTRODUCTION

Beta thalassemia is the commonest genetic disorder worldwide (1). It is an autosomal recessive disorder in which one or both chains of hemoglobin are either missing or deficient (2). Peripheral blood smear examination and red cell indices are effective and valuable tools for screening and diagnosis of thalassemia (3). There are two main types of thalassemia; beta thalassemia and alpha thalassemia. If the beta chain of Hemoglobin is missing then it is called beta thalassemia (4). Thalassemia is a genetic disorder that is transmitted from parents to their children. It is more prevalent in those children whose parents are first cousins. Consanguinity is prevalent in South Asia and Middle East countries with rates exceeding 40% of all marriages (5). When both parents are carrier of Thalassemia, they have a 25% chance of having a thalassemia major child (with both genes for beta thalassemia), a 50% chance of having children with thalassemia minor (with only one gene

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for beta thalassemia), and a 25% chance of having a child without thalassemia major or minor (with both genes for normal beta chains) (6). Consanguineous marriages are practiced throughout the world with different ethnicity and socio-cultural background (7). The highest prevalence of consanguineous marriages is found in North Africa, the Middle East and central and south Asia (8). There are an estimated 60-80 million carriers of thalassemia worldwide. Each year 60,000 beta Thalassemia carriers are born. Approximately 1.5-3 % of the world population is suffering from beta thalassemia (9). About 50 % of thalassemia trait in the world is in South East Asia (10). 60,000 children in Pakistan are suffering from thalassemia. Approximately 5% of our population is the carrier of thalassemia (11). The prevalence of thalassemia in Khyber Pakhtunkhwa Province is approximately 4 % (two million) (12). Premarital screening for thalassemia is an effective tool for its prevention as commonly practiced in Iran (13). Thalassemia can be prevented by prenatal diagnosis in pregnant ladies during the first trimester of pregnancy in families positive for Thalassemia. CVS technique is widely used for in vitro diagnosis of Thalassemia. If turn out to positive, pregnancy can be terminated on medical grounds in almost all religions of the world (14). Awareness about the CVS technique is limited in the general population (15). The rationale of this study is to highlight the importance of prenatal diagnosis of Thalassemia through the Chorionic Villous Sampling technique for the prevention of this commonest genetic disorder.

MATERIALS AND METHODS

It was a cross sectional study conducted in Frontier Foundation Blood Transfusion center Peshawar within six months from Jan; 2018 to Jun; 2018. A total of 130 subjects were selected through convenient sampling technique. A questionnaire was used for the collection of data. Variables like cousin marriages, awareness, sources of awareness and validity of the technique were analyzed using SPSS Version 20. Those women who had at least one Thalassemia major child were included while women unwilling or having children with other genetic disorders were excluded from the study.

RESULTS

A total of 130 mothers were interviewed and the data were analyzed. Out of 130 couples, 99 were cousins while 31 were non-cousin marriages. Of the total, 78 % of mothers were aware while 22 % were unaware of the technique. The source of awareness was mainly their referring physicians. The sensitivity of the test was 75 % and Specificity was 91 %.

A total of 50 mothers underwent the CVS technique. Of the total 50, 15 were positive for CVS while 35 were negative for CVS. 11 out of 15 positive were counseled for abortion, however, 04 out of 15 refused abortion. Sensitivity was calculated among those who didn't go for abortion. The specificity of the test was calculated among those who were negative on the CVS test.

Sensitivity of CVS calculated was 75 % (¾ x 100 = 75 %).

Specificity was 91 % (32/35 x 100 = 91 %)

Table 1: Frequency of Consanguinity

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

Table 2: Frequency of awareness about CVS in Mothers

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

Table 3: Sources of Awareness

Source	Number	% age
By physician	116	89
Family members	05	3.8
Friends	05	3.8
Media	00	00
Other sources	04	3.7

DISCUSSION

My study was a cross sectional study which was conducted in Peshawar in which my aim was to determine the prevalence of cousin marriages in the study population and to determine the validity of CVS technique which is not yet commonly used in Pakistan. As the prevalence of thalassemia major is quite high in the province of Khyber Pakhtunkhwa (16), there was a strong possibility that the prevalence of cousin marriages would be equally high in this province. In our study frequency of consanguineous marriages was 76.15 % which is compatible with international study results (17). Knowledge of parents in cases

and controls about thalassemia as well as chorionic villous sampling (CVS) during pregnancy if they have any future planning for more children was also checked. The awareness level was 78 % among mothers of Thalassemia children. Our results match the results of an international study (18). Prenatal diagnosis of thalassemia major in the 1st trimester using Chorionic Villous sampling (CVS) technique had a sensitivity of 75 % and specificity of 91 %. Low sensitivity may be due to the low number of women who underwent abortion or due to the limited number of CVS centers. The results of our study are compatible with an international study (19).

CONCLUSION

Keeping in view the high prevalence rate of thalassemia in Pakistan, premarital screening and prenatal diagnosis of thalassemia through CVS technique can be used as key strategies in the prevention of this commonest genetic disorder.

REFERENCES

1. Malik S, Syed S, Ahmed N. Complications in transfusion–dependent patients of ß-thalassemia major. Pak J Med Sci Jul - Sep 2009; 25(4):678-82.
2. Ramezanzadeh M, Salehi M, Salehi R. Assessment of high resolution melt analysis feasibility for evaluation of beta-globin gene mutations as a reproducible, cost-efficient and fast alternative to the present conventional method. Adv Biomed Res. Apr 192016; 5 (3):71-4.
3. Aman A, Zehra N, Haider G, Anjum F, Rani S, Munir A A. Role of mean corpuscular volume as screening test for Thalassemia in pregnant women at Isra University. Pak J Med Sci April – June 2010; 26(2): 390-3.
4. Boon NA, Colledge NR, Walker BR, Hunter JA. Davidson principle and practice of medicine: blood disorders. 20th ed. New Delhi: Churchill Livingstone (Elsevier) 2006; 1038-1039.
5. Abdalla, B., & Zaher, A. Consanguineous marriages in the Middle East: Nature versus Nurture. The Open Complementary Medicine Journal, 2014; 5(1): 1-11.
6. Neufeld EJ. Mendelian inheritance in man. J hemtaol, 2016; 90 (5) : 649-54
7. Pakistan observer Saturday 07, 2013. Inter cousin marriages is the major cause of thalassemia major; a report presented by Lt. General (R) Moeen ud Din Haider (President Pakistan Thalassemia Federation) in 8th thalassemia conference at RIPHAH International University Al mezan campus Peshawar road Rawalpindi on Sunday 06 May, 2012.
8. Sirdah MM. Consanguinity profile in the Gaza Strip of Palestine: large-scale community-based study. Eur J Med Genet, Feb 2014; 57(2-3):90-4.
9. Ul ain Q, Ahmed L,Hassan M, Rana S M, Jabeen F. Prevalence of beta thalassemic patients associated with consanguinity and Anti HCV Antibody positivity- A cross sectional study. Pak J.zool 2011;43(1):29-36,.
10. Niazi M, Tahir M, Raziq F, Hameed A. Usefulness of red cell indices in differentiating microcytic hypochromic anemia. Gomal J Med sci 2010; 8(2): 41-43.
11. Pakistan Observer Saturday September 07, 2013. Intercousin Marriges is the major cause of Thalassemia major; a report presented by Lt. Gen. (R) Moeen ud din Haider (president Pakistan Thalassemia federation) in 8th national thalassemia conference at RIPHAH International University Al mizan campus Peshawar road Rawalpindi on Sunday 06, May 2012.
12. Tasleem S, Tasleem H, Siddiqui MA, Adil MM, Rashid Y. Prenatal diagnosis of beta thalassemia by chorionic villous sampling. J Pak Med Assoc, Nov 2007; 57(11):528-31.
13. Samavat A, Modell B. Iranian national thalassaemia screening programme. BMJ, November 2004; 329 (7475): 1134–7.
14. Beulen L, Van den Berg M, Faas BH, Feenstra I, Hageman M, Van Vugt JM, Bekker MN. The effect of a decision aid on informed decision-making in the era of non-invasive prenatal testing: a randomized controlled trial. Eur J Hum Genet, May 2016; 40 (4): 70-5.
15. Rudra S, Chakrabarty P, Hossain MA, Ripon MJ, Rudra M, Mirza TT. Awareness among parents of β-thalassemia Major patients regarding prenatal diagnosis and premarital screening in day care center of transfusion medicine department. Mymensingh Med J,2016 Jan; 25 (1) : 12-17
16. Pakistan Observer Saturday September 07, 2013. Intercousin Marriges is the major cause of Thalassemia major; a report presented by Lt. Gen. (R) Moeen ud din Haider (president Pakistan Thalassemia federation) in 8th national thalassemia conference at RIPHAH International University Al mizan campus Peshawar road Rawalpindi on Sunday 06, May 2012.
17. Kremastinos T D, Farmakis D, Aessopos A, Hagalis G, Hamodraka E, Tsiapras D, Karen A. Beta Thalassemia cardiomyopathy: History, present consideration and future perspective. Circ Heart Fail, May 2013; 3: 451-458
18. Rudra S, Chakrabarty P, Hossain MA, Ripon MJ. Awareness among parents of beta thalassemia major patients regarding prenatal diagnosis and premarital screening in day care center of transfusion medicine department. Mymensingh med j, January 2016; 25 (1) : 12-17.
19. Ghahramani F, Alimohamadi Y, Mahboubi M. Negative predictive value of CVS in diagnosis of thalassemia in genetic laboratory of Dastgheib Hospital, Shiraz Iran, 2012. Arch Iran Med, 2014 July ; 17 (7) : 483-5.