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KNOWLEDGE REGARDING PREMARITAL THALASSEMIA SCREENING AMONG THE PEOPLE OF GAJJUMATTA (Community) LAHORE, PAKISTAN

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Abstract: **Thalassemia is a red blood cell (RBC) hereditary autosomal recessive disorder which includes the lack of, or defect in genes accountable for development of hemoglobin (Haque, A. T. M. E et al., 2015). Depending upon the globin chain involvement, thalassemia is divided into Alpha-thalassemia and Beta thalassemia. Beta- thalassemia is further classified as beta-thalassemia major, intermediate and minor, on the basis of clinical severity and inheritance pattern. (Stephens, A. D.et al., 2012) These patients even have substantial "hepatosplenomegaly due to hemolysis and extra medullary hematopoiesis". At this stage, urgent medical intervention is crucial for prevention of a number of complications. (Haque et al.,2015) Prenatal diagnosis and carrier detection is available in Pakistan for over a decade, but it's use remains limited due to lack of public awareness and the costs involved. It is reported that more than half of the families of the registered thalassemic patients did not request for prenatal diagnosis. (Naseem S, Ahmed S at el.,2008) Factors identified for this low utilization of services were lack of awareness about thalassemia, poor access to treatment, delay in seeking consultation and high cost of treatment.**

Objectives: **To determine the knowledge regarding premarital thalassemia screening among the people of Gajjumatta community Lahore, Pakistan.**

Methodology: **Quantitative descriptive cross sectional design was applied to access the contributing factors of premarital thalassemia screening. A cross-sectional research is the study that collects information about people at specific time of period (Lisa B 2014).This study was conducted at Gajjumatta community Lahore in natural setting. Data was collected by using demographic information and questionnaire related to premarital screening and knowledge regarding thalassemia. Data was collected from 103 people ranging from ages 18 years to 30 years. The data was analyzed by using Statistical Package of Social Science (SPSS) for Windows version 18.0**

Considerations: **The study was conducted after obtaining a permission letter from the Institution Review Board (IRB) and a permission letter from the society head along with an informed consent from the study participants.**

Results: **About 65% are unaware about the (genetic) nature of Thalassemia and 34% have the knowledge regarding the hereditary nature of Thalassemia. This concludes that people are not aware of the genetic nature of this disease. About 70% people do not have adequate information regarding Thalassemia and the factors that lead to the disease. Knowledge about the transmission of Thalassemia by blood was inadequate that was about 75%. Majority of participants have no knowledge about the role of consanguineous marriage that was almost 72%. About 84% people have no knowledge of signs and symptoms of this disease and 66% people do not know about the diagnostic criteria of the disease.**

Conclusions: **The findings in the current study offer a comprehensive understanding of people of knowledge regarding premarital thalassemia screening. This provides valuable evidence to assess that the Gajjumata community has insufficient knowledge regarding premarital Thalassemia screening. Screening should be performed on every individual before marriage to reduce the chances of Thalassemia. The control and prevention program for Thalassemia should be established through education counseling, screening and antenatal diagnosis.**

Keywords: **Thalassemia, Hereditary, Consanguineous, Hepatosplenomegaly, Screening.**

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1. INTRODUCTION

The word Thalassemia is descended from the Greek language Thalassa (sea) and haima (blood)". Thalassemia is a red blood cell (RBC) Hereditary Autosomal Recessive Disorder which includes the lack of or defect in genes accountable for development of hemoglobin (Haque, A. T. M. E et al., 2015)

Thalassemia is a hereditary haemoglobinopathy occurring from the absence, or decreased development of either alpha or beta globin chain (Olivieri, N. F.et al.,1999).

Depending upon the globin chain involvement, Thalassemia is divided into Alpha-Thalassemia and Beta-Thalassemia. Beta-Thalassemia is further classified as Beta-Thalassemia major, intermediate and minor, on the basis of clinical severity and inheritance pattern. (Stephens, A. D.et al.,2012)

Thalassemia major is an inherited hematological disorder causing anemia in affected children. It is an autosomal recessive disease in which chromosome 11 is involved. It affects building of the B globin chain of hemoglobin which is either decreased or absent leading to early turnover of Red Blood Cell (RBC). When one of the beta globin chain gene is normal and other abnormal, it is thalassemia minor. If both the genes are involved and disorder manifests late in life (older children and adults), it is called as Thalassemia intermediate. If both the genes are involved and disorder is manifested early in life (infants) it is called as Thalassemia major which is a more aggressive disease. Abnormal shaped RBCs are rapidly destroyed by the reticula endothelial system particularly spleen leading to microcytic hypo chromic anemia and iron overload (Galanello, R.,et al.,2010)

Majority group are of moderate severity and often do not need blood transfusion. Two genes are involved in making the beta hemoglobin chain but only one gene will be affected in minor and these individuals are of the intermediate carriers. In β-thalassemia intermedia or major (Cooley's anemia), both the genes possess anomaly. β-thalassemia major is the most severe form and results from the inheritance of the homozygous state for the phenotype. Pediatric patients of thalassemia present as early as 3 months. Patients become increasingly "pale, difficulty in feeding and irritable". These patients even have substantial "hepatosplenomegaly due to hemolysis and extra medullary hematopoiesis". At this stage urgent medical intervention is absolutely necessary for prevention of a number of complications. (Haque et al.,2015)

According to (Bryan et al., 2011) Thalassemia was not recognized as a clinical entity until 1925, when a syndrome was described as occurring early in life that was associated with splenomegaly and bony deformities. There are approximately 240 million people worldwide who are heterozygous for β-thalassemia and approximately 200,000 affected homozygotes are born yearly.

The main prevention strategies comprise of providing appropriate information to the public and professionals, screening and counseling of families at risk and screening of couples prior to marriage.

Prenatal diagnosis and carrier detection is available in Pakistan over a decade but its use remains limited due to lack of public awareness and the cost involved and reported that more than half of the families of the registered thalassemic patients did not request for prenatal diagnosis. Factors identified for this low utilization of services were lack of awareness about Thalassemia, poor access to treatment, delay in seeking consultation and high cost of treatment. (Naseem S, Ahmed S at el.,2008)

Like many other developing Asian countries, βthalassemia poses an increasing burden for healthcare services in Pakistan and it is not possible to provide blood transfusion and iron chelation therapy to all patients with limited available national resources. Bone marrow transplantation is highly expensive and unaffordable for most Pakistani patients. In countries like Pakistan, prevention is the least expensive and most effective means to deal with β-thalassemia. The best possibility of preventing thalassemia major from occurring is to detect the carriers at a pre-marital stage and prevent them from getting married by giving them proper marriage counseling. It is thus essential to carry out screening programs for detection of Thalassemia carriers and to provide them the advantage of marriage counseling to reduce the incidence of new Thalassemia major cases. There have previously been few studies on β-thalassemia mutations in various regions and ethnic groups of Pakistan.

The basic prevention strategies comprise of providing appropriate information for the public and professionals, screening and counselling of families at risk and screening of general population prior to marriage. Appropriate and extensive screening, accurate detection and counselling of at risk couples along with prenatal diagnosis are promising strategies for the reduction of mortality and morbidity from thalassemia in countries, where it is prevalent. Premarital screening alone has tremendously decreased the birth prevalence of β-thalassemia major (Ishaq, F et al.,2015)

Keeping in mind all the available treatment choices, we are definitely left with no other alternatives but prevention of this disease because it remains very relevant as for as Thalassemia is concerned that "prevention is better than cure". The rationale of the study is based upon the research question. What is the level of medical and social awareness for this debilitating disease among people of the community, as so far no study has been conducted in the present set up? The results of this study will help determine the areas needing further improvement in public awareness campaigns as part of national Thalassemia prevention programs. This study was conucted to determine the level of awareness among people of the community regarding prevention, options and complications of the disease. The low literacy rate in Pakistan is the main obstacles in improving the level of awareness which has been supported in various studies.(Ghafoor, M. B.at el., 2016)

Premarital screening programs have been introduced worldwide in many β-thalassemia affected countries. Several countries have benefitted from such programs. For example, Cyprus, which reduced the prevalence of β-thalassemia from extremely high to almost negligible levels following the introduction of a screening program.

SIGNIFICANCE OF STUDY:

The result of this study will help determine the areas needing further improvement in public awareness campaign as part of national Thalassemia prevention program. The study will help people to gain the knowledge regarding Thalassemia. Early knowledge regarding premarital Thalassemia screening will reduce the risk of Thalassemia.

2. METHODOLOGY

STUDY POPULATION

People of Gajjumatta Community Lahore, Pakistan.

RESEARCH STUDY DESIGN:

Quantitative descriptive cross sectional design was applied to access the contributing factors of premarital thalassemia screening.

RESEARCH SETTING:

Research setting was the Gajjumatta community Lahore, Pakistan. The data was collected in natural setting.

RESEARCH SITE:

Gajjumatta community Lahore was the site for data collection.

TARGET POPULATION:

People above 18 to30 years of age and married couples

SAMPLE SIZE:

The total population of Gajjumata is 20,000. By taking into consideration, the Taro Yamni sample size calculation formula and keeping precision level 10%. By using 90% confidence level, the sample size was 03 subjects, out of the total study population.

SAMPLING TECHNIQUE:

Convenient sampling technique was used.

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INCLUSIVE CRITERIA:

All young females and males who were waiting for marriage and their guardians.

EXCLUSIVE CRITERIA:

- Old people and those who did not consent
- People below 18 years of age were excluded from this study.
- People who were not willing to participate in research study.

DURATION OF THE STUDY:

Duration of study was 3 to 4 months.

STUDY TOOL:

The instrument of data collection was a research questionnaire, which was already checked for validity and reliability. Data was collected by using demographic information and questionnaire related to premarital screening and knowledge regarding Thalassemia and response was documented.

ETHICAL CONSIDERATION:

The study was conducted after obtaining a permission letter from the Institution Review Board (IRB). A permission letter from society head and informed consent from the study participants was also collected after giving complete information about the study purpose.

3. DATA ANALYSIS METHOD

The data was analyzed by using Statistical Package of Social Science (SPSS) for windows version 18.0 Frequency, percentage, chi-square and correlation was used to assess the relationship between variables. Demographic variables like age and monthly income was presented by mean and standard deviation. Demographical categorical data like gender qualified and marital status will be presented by frequency and percentage. Specific questions about the knowledge regarding Thalassemia will be presented by categorizing knowledge as poor, average and good knowledge.

SIGNIFICANCE LEVEL:

The significance level for all statistical analysis was set at 0.05.

STUDY TIMELINE:

The data was collected from DEC, 2019 to MAR, 2020

ETHICAL CONSIDERATION:

The study was conducted after taking permission letter from the Institution Review Board (IRB). A permission letter from society head and informed consent from the study participants was also collected after giving complete information about the study purpose.

Table 1: Demographic variables,

Demographic variables	
1: Age	**18-30 years**
2: Gender	**Male and females**
3: Marital status	**Married and unmarried**

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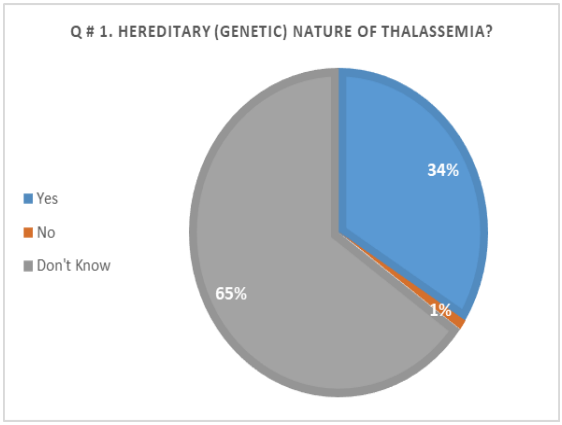
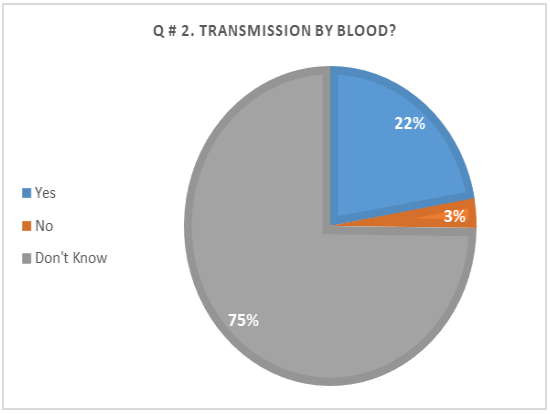
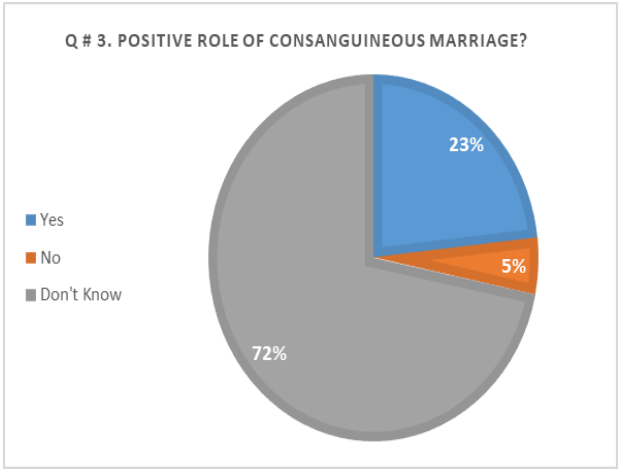
4. RESULTS



- As evidences, about 65% are unaware about the (genetic) nature of Thalassemia and 34% have knowledge regarding the hereditary nature of Thalassemia that concludes that people are not aware of the nature of this genetic disease.



- Knowledge about the transmission by blood can cause Thalassemia was inadequate. About 75% people have no knowledge, 22% have knowledge of Thalassemia transmission by blood, 3% did not reply.



- 72% have no knowledge about the role of consanguineous marriage almost 23% were aware of it and 5% have not answered this question.

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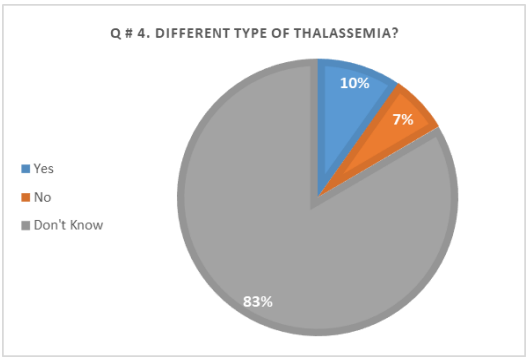
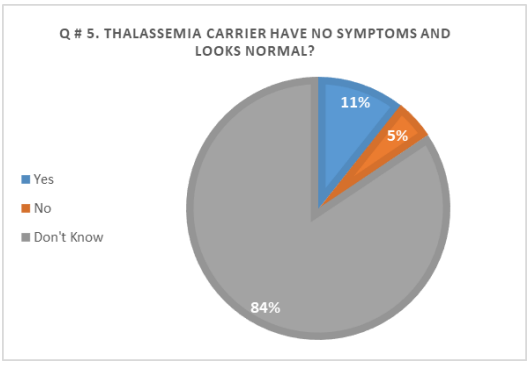
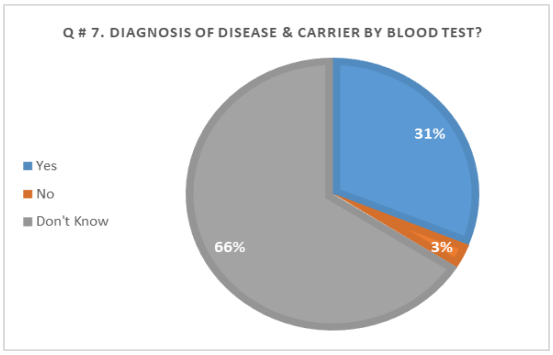


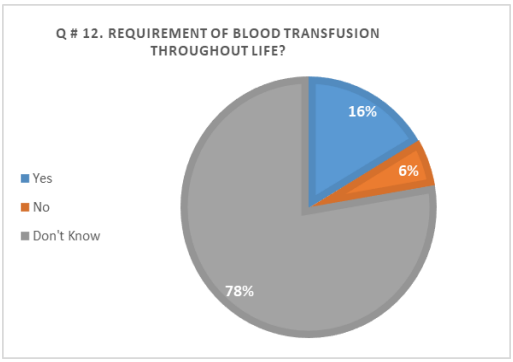
- Only 10% of respondents have sufficient knowledge about the types of thalassemia, 83% did not know and 7% did not give any answer.



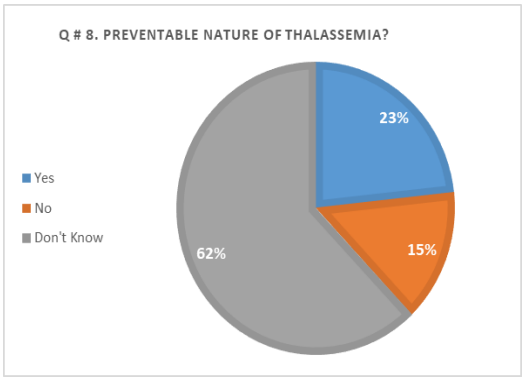
- About 84% of the people have no knowledge about signs and symptoms of Thalassemia, only 11% were aware of it, 5% was did not.



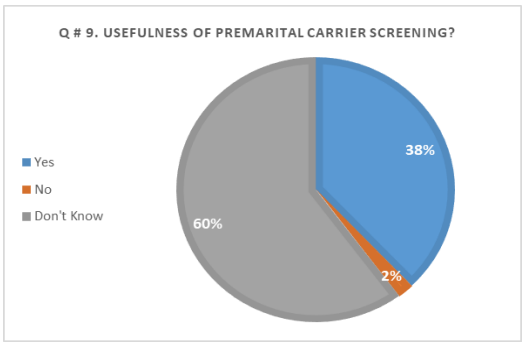
- Diagnostic criteria of the disease was known by only 31%, 66% people were not aware and 3% did not answer.



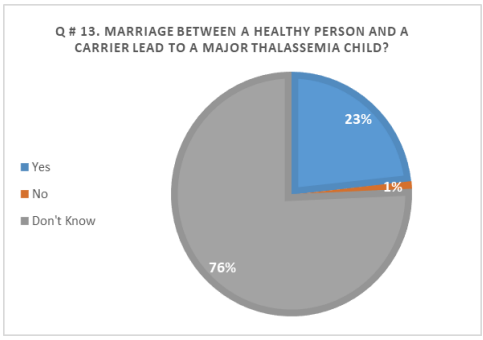
- 78% did not know about the requirement of blood transfusions for the patients suffering from Thalassemia, only 16% were answered - yes, 6% did not reply.



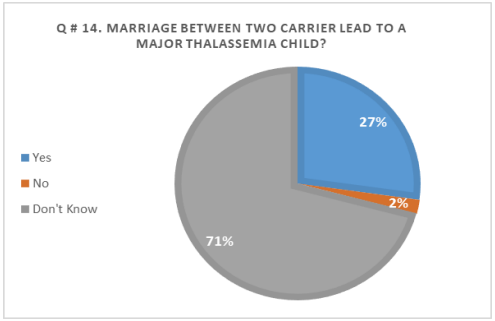
- 62% participants have no knowledge about the preventable nature of Thalassemia, 23% have sufficient knowledge, 15% did not reply.



- 60% were not aware about the practicality of Thalassemia carrier screening, 38% were aware about the usefulness of this test, only 2% did not reply.



- 76% said that marriage between a healthy person and a carrier cannot lead to a major Thalassemia child, only 23% said yes,1% did not answer.



- 71% said No in response to the question that can marriage between two carrier lead to a major Thalassemia child. 27% replied in Yes and 2% declined to answer.

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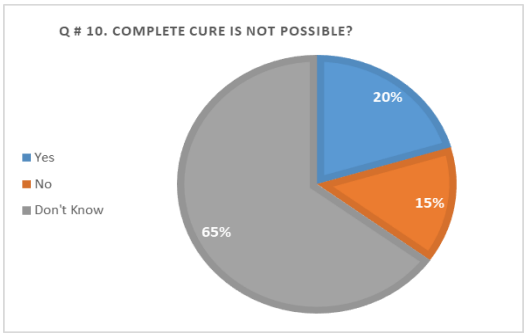
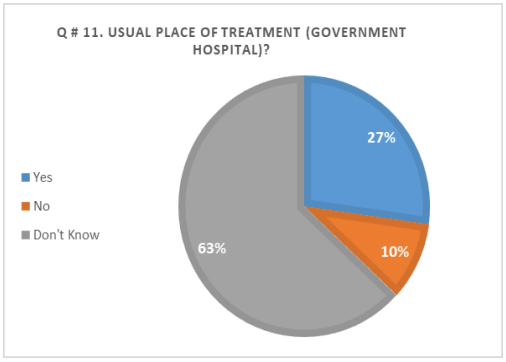
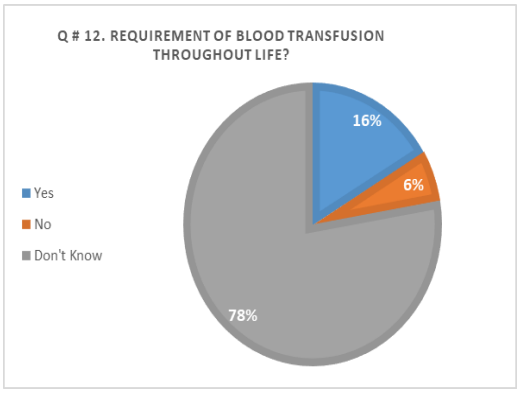


- 20% said that complete cure is not possible, 65% said that it is possible, 15% gave no answer.



- Only 27% participants agreed that the usual place for the treatment of Thalassemia are the government hospitals, 63% denied and 10% did not reply.



78% said that blood transfusion is not required for thalassemia patient throughout life ,16%said that it is needed ,6% didn't reply.

5. DISCUSSION

Thalassemia is a disorder due to an autosomal inheritance. Its prevention depends on awareness which is frequently contributed by the educational level of the community. In the present study people have no formal education and have low educational level regarding premarital Thalassemia screening. There is inadequate knowledge of accurate frequency and distribution of Thalassemia disorder in the developing countries. In the present study it was observed that due to frequent visits, to the community, we can increase knowledge regarding Thalassemia screening.

This Study was conducted on pre designed and pre tested questionnaire and a cross sectional study design was selected, assessment of the knowledge regarding premarital Thalassemia screening. 103 samples were collected from Gajjumatta community Lahore, Pakistan. Exclusion and Inclusion criteria mentioned in the methodology were followed. The findings in the current study offer a comprehensive understanding of people of knowledge regarding premarital Thalassemia

screening. This provided valuable evidence to assess that Gajjumatta community has insufficient knowledge regarding premarital Thalassemia screening. Screening should be conducted on every individual before marriage, in order to reduce the chances of Thalassemia. The control and prevention program for Thalassemia should be established through education counseling, screening and antenatal diagnosis.

All persons in the community should be invited to participate since the participant's responses indicate their specific knowledge regarding the prognosis. The pattern of inheritance was poor. Early education can provide prevention of the disease. Lack of knowledge regarding premarital Thalassemia screening leads to increasing incidences. It is necessary to check the level of awareness of premarital screening regarding early detection and prevention of Thalassemia.

6. CONCLUSION

The present study concluded that community knowledge was limited. However due to regular visits and genetic counseling at Thalassemia Centers in the community can reduce the burden of Thalassemia in Pakistan. There is need for education and awareness among the carriers. The mass media should be encouraged to make more programs such as films, plays, dramas and talk shows to help increase the awareness of Thalassemia. The control and prevention program for Thalassemia should be established through education counseling, screening and antenatal diagnosis.

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