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BETA THALASSEMIA PREVALENCE AND GENETIC AWARENESS

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ABSTRACT

To determine the prevalence of β thalassemia in Hyderabad, Sindh, and evaluation of risk factors and genetic awareness. Cross-sectional (prevalence study). 08 months. This study was conducted between April 2013 to November 2013 on different age groups from 1 to 40years old who have come to advance diagnostics and research laboratory LUMHS for their reports. Around 209 cases were screened for Thalassemia major and minor. Highest number (34 cases) with Thalassemia major was noted in children with age of (0-5 years) followed by 4 cases (6-10years), 1 case (21-40) 0 cases in (11-20 years). It is concluded that maximum percentage of Beta thalassemia major was seen in this study particularly in children with the age between 0-5 however knowing this disease have life threatening outcomes. Thus, we recommend that urgent measures to increase awareness and control the disease be taken.

Keywords: Prevalence, Beta Thalassemia, Genetic awareness

INTRODUCTION

Thalassemia is inherited autosomal recessive blood disease. There are two types Alpha and Beta thalassemia. The haemoglobin is made up of four protein chains, two α and two β globin chains. In thalassemia, patients have either defects in α or β globin chain.The β globin chains are encoded by a single gene on chromosome 11; α globin chains are encoded by two closely linked genes on chromosome 16 **[1].** In thalassemia, the disease is caused by the weakening and destruction of red blood cells. Haemoglobin is the protein in red blood cells that carries oxygen. Individuals with thalassemia produce less amount of haemoglobin and smaller amount of circulating red blood cells as compared to normal, which results in mild or severe anaemia.Thalassemia can cause major complications, including iron overload, bone abnormalities and cardiovascular disorder **[2, 3]**.

The total yearly incidence of symptomatic individuals is projected at 1 in 100,000 all over the world and 1 in 10,000 individuals in the European Union. Three main forms have been described: thalassemia major, thalassemia intermediate and thalassemia minor.Beta-thalassemia is predominant in Mediterranean countries, the Middle East, Central Asia, India, Southern China, and the Far East as well as countries along the north coast of Africa and in South America. The highest carrier frequency is reported in Cyprus (14%), Sardinia (10.3%), and Southeast Asia **[4]**. Cousin marriages, relocation of people and intermarriage between different ethnic groups has introduced thalassemia in nearly every country of the world, Including Northern Europe where thalassemia was previously absent. It has been estimated that about 1.5% of the global population (80 to 90 million people) are carriers of beta-thalassemia, with about 60,000 symptomatic individuals born annually, the great majority in the developing world **[5]**. It has been estimated around 5000-9000 children born every year with β-thalassemia.With gradual control of malnutrition and communicable diseases, β-thalassemia major patients who earlier died young are now surviving long enough to seek medical attention **[6].**

However in countries like Pakistan, raising problem for health-care services, only less amount of affected population can afford adequate blood transfusions with effective iron chelation and bone marrow transplantation. Hence, prevention has been demonstrated to be the way forward. It can be reduce by

prenatal diagnosis and genetic counselling to the carriers, effective measures for identification of different mutations should be taken more over prenatal diagnosis can open more door for it preventive medicine. The aim of this study is to evaluate the prevalence and public awareness level of beta thalassemia.

MATERIAL-METHOD

The Study was prospective all cases were collected from Advance Diagnostic laboratory Civil hospital, Liaquat University of Medical and Health Sciences, Hyderabad, Mirpurkhas and Jamshoro, study was conducted between April 2013 to November 2013 and between different age groups, moreover all cases were registered on Performa, after taking consent. Moreover data was collected according to family history of thalassemia, 6ml of blood was drawn in 2 separate EDTA tubes, one tube was used for Complete Blood Count (CBC) while on the other hand the 2nd sample was analysed for Hb Electrophoresis after haematology analysiswas confirmedon the basis of CBC results and microscopic examination where RBCs were increased and MCV and MCH level were found below then there normal values only those samples was further analysed on Alkaline cellulose acetate method. Apparatus was used for CBC Xe-2100 and for Hb Electrophoresis (HPLC Bio Rad) machine was used.All data was entered in computer and mean, frequencies andstandard deviation were calculated by using SPSS.

RESULTS

This study was conducted between April 2013 to November 2013 on different age groups from 1 to 40years old who have come to advance diagnostics and research laboratory LUMHS for their reports. Around 209 cases were screened for Thalassemia major and minor. Highest number (34 cases) with Thalassemia major was noted in children with age of (0-5 years) followed by 4 cases (6-10 years), 1 case (21-40) 0 case in (11-20 years) **Table 1 and Figure 1**. On the other hand highest number (17cases) of thalassemia minor was seen in patients with the age between (21-40 years) followed by 5 cases (0-5years), 5 case (11-20 years) 2 case in (6-10 years).

Table 1: Shows the number of cases of thalassemia major in different age groups

Numbers	Thalassemia major with age 0-5 years	Thalassemia major with age 6-10 years	Thalassemia major with age 21-40 years
34 Cases	29 cases	4 cases	1 cases

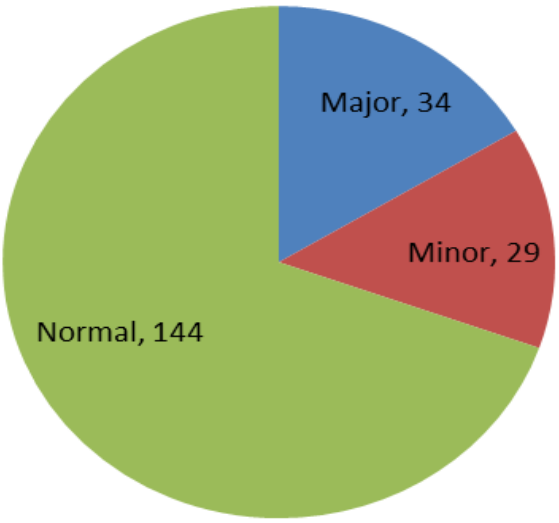

Figure 1: Distribution of patients with major minor cases

DISCUSSION

It is estimated that carrier rate of Beta-thalassemia in Pakistan is 5.4%, however world wide the carrier rate for this disease is 1.4-7.96% **[7]**. One out of every twenty individuals carries a gene for B-thalassemia, making it one of the commonest inheritable disorders in Pakistan **[8, 9]**. In a developing country like ours, prevention of Beta-thalassemia through carrier screening, genetic counselling and pre-natal diagnosis can be the best choice to control the affected births **[10]**.

The service of pre-natal diagnosis is available in the major cities of Pakistan where patients from neighbouring rural areas are referred. The patients seen at our centre usually come from interior Sindh, with a strong rural background and low literacy and income rate. To make matters worse these people also strongly adhere to the age old customs of consanguineous marriages within the same family. High prevalence of this disease in this area with such traditional practices can consequence in explosion of disease in the area. Prompt measures are required to control the disease in these high risk populations and pre-natal diagnosis can be one of the most effective tools when used in a vigilantly selected group of patients to decrease the spread of the disease. More over a better emphasis needs to be placed about the awareness of the factors leading to the spread of the disease in our target population. On the other hand Clinicians should be trained on when to suspect these disorders, how to refer them for genetic counselling and pre-natal testing so that healthier outcomes can be ensured. These patients are commonly very worried and

concerned between two boundaries of potentially bad outcomes, the possibility of having a baby with a serious genetic disorder and the chance of losing an otherwise normal and wanted pregnancy. Their concerns and doubts need to be addressed in a very helpful manner to improve patient security.

The assessment of the comparatively incidence of beta thalassemia, people seem to have a lack of awareness about the disease. These results show a significant possibility for improving the basic awareness of beta thalassemia among our population. We recommend further studies to test beta thalassemia patients' awareness and attitude toward their illness.

CONCLUSION

It is concluded that maximum percentage of Beta thalassemia major was seen in this study mostly in children with the age between 0-5 however knowing this disease have life threatening outcomes. Moreover affected families have social stigma. This might be due family forcing cousin marriages or prenatal screening not done for such diseases, Having all this in mind proper measures should be taken by doctors, and other health workers such as carrier counselling, for prenatal screening and information about life threatening diseases. This may help to identify disease early which in turn reduces the burden of diseases in our population.

REFERENCES

[1] Galanello, Renzo; Origa, Raffaella (21 May 2010). *Beta-thalassemia*. Orphanet Journal of Rare Diseases.

[2] Isma'eel, Hussain , Maria D Cappellini, and Ali Taher. "Chronic transfusion, iron overload and cardiac dysfunction: a multi-dimensional perspective." The British Journal of Cardiology 15.1 (2008): n. pag. BJC. Web. 16 May 2013.

[3] Tanner, Mark A, J Malcolm Walker, Sunil V Nair, Martina Pibiri, Annalisa Agus, Mark A Westwood, Gillian C Smith, Carlo Dessi, Renzo Galanello, and Dudley J Pennell. "Combined Chelation Therapy In Thalassemia Major For The Treatment Of Severe Myocardial Siderosis With Left Ventricular Dysfunction." Journal of Cardiovascular Magnetic Resonance 10.1 (2008).

[4] Flint J, Harding RM, Boyce AJ, Clegg JB: The population genetics of the

hemoglobinopathies. *Bailliere's Clinical Hematology* 1998, **11:**1-50.

[5] Vichinsky EP: Changing patterns of thalassemia worldwide. *Ann N Y Acad Sci* 2005, **1054:**18-24.

[6] Thalassemia International Federation: *Guidelines for the clinical management of thalassemia*. 2nd edition. 2008 [http://www.thalassemia.org.cy]

[7] Hashmi JA, Farzana F.Thalassemia Trait,abnormal haemoglobins and raised fetal haemoglobin in Karachi (letter). Lancet .1976; 1:206

[8] Khateeb B, Moatter T, Shaghil AM, Haroon S, Kakepoto GN. Genetic diversity of beta-thalassemia mutations in Pakistani population. J Pak Med Assoc. 2000; 50:293-96.

[9] Lodhi Y. Economics of thalassemia management in Pakistan. In Thalassemia Awareness Week, Ed. Ahmed S. Friends of Thalassemia 2003.

[10] Saller, DevereUX N. Jr MS, MD; Canick, Jacob A. PhD. Current Methods of Prenatal Screening for Down Syndrome and Other Fetal Abnormalities. Clinical Obstetrics and Gynecology: March 2008 - Volume 51 - Issue 1 - pp 24-36.