ISSN- O: 2458 - 868X, ISSN–P: 2458 – 8687
Index Copernicus Value: 49. 23

International Journal of Medical Science and Innovative Research (IJMSIR)
IJMSIR : A Medical Publication Hub
Available Online at: www.ijmsir.com
Volume – 3, Issue –3, May - 2018, Page No. : 128 - 132

Retrospective Study of Sickle Cell Disease in Tribal District of Maharashtra: Gondia

Dr Suresh Ghangale[1] , Associate professor, Department of Biochemistry, Indira Gandhi Government Medical College, Nagpur

Dr Sanjivani Jadhao[2], Professor and Head, Department of Anatomy, DMACH&RC, Wanadongri, Nagpur.

Dr Pallavi Kamble[3], Assistant Professor, Department of Biochemistry, Government Medical College, Gondia.

Dr Ashok Jadhao[4], Professor and Head, Department of Community Medicine, Indira Gandhi Government Medical College, Nagpur.

Dr Durgesh Sahoo[5], Assistant Professor, Department of Community Medicine, Indira Gandhi Government Medical College, Nagpur

Correspondence Author: Dr Ashok Jadhao, Professor and Head, Department of Community Medicine, Indira Gandhi Government Medical College, Nagpur

Type of Publication: Original Research Paper

Conflicts of Interest: Nil

Abstract

Introduction: Sickle cell disease (SCD) is a genetic disorder of great epidemiological, clinical and public health relevance in developing countries like India. In Maharashtra, the prevalence of disease varies from 1.9% to 33.5% in different communities with an overall prevalence of 10%. This study was conducted to estimate the yield of HPLC in diagnosing Sickle cell disease on the basis of symptoms suggesting sickle cell disease in an endemic area of tribal district.

Material and Methods: This retrospective record based study was conducted in a tertiary care institute in Gondia district of Maharashtra among 767 participants during the period from October 2009 to February 2015. Patients with the symptoms of sickle cell disease were enrolled for HPLC testing in clinical biochemistry laboratory.

Results: Most of the cases were registered in 2011-12 year. The mean age of cases was 18.01±10.85 years with a range of 1-60 years. 5.35% of study subjects were affected in sickle cell disease while 4.43% of study subjects were sickle cell trait. Most of the Sickle cell disease cases were confirmed in males (61%) while most of the carrier was found in females (70.6%).

Conclusion: The overall yield of sickle cell disease and carrier was found to be 9.78% with higher prevalence of sickle cell disease among males and sickle cell carrier among females. HPLC is best tool for screening of sickle cell disease for early diagnosis and prevention of complications amongst them..

Keywords: Sickle cell disease, HPLC, Sickle cell carrier

Introduction

Sickle cell disorders are a group of autosomal recessive disorders where the normal round shape of red blood cells becomes like a crescent moon.[1] It is caused by point mutation at the sixth position in the beta globin chain resulting in substitution of glutamic acid by valine and the abnormal hemoglobin S (HbS). The HbS has poor solubility in the deoxygenated state; that leads to polymerization of deoxy HbS causing a distorted sickle shape and eventually increased hemolysis and vaso-occlusion of sickle red cells. [2]

It manifests in two forms. If a person inherits only one gene responsible for sickle hemoglobin from either of the parent, the condition is called carrier or trait. If a person receives two defective genes, one from each parent, the condition is called Sickle cell disease (SCD). Symptoms are rare before the first 6 months of life as infants are protected largely due to persistence of fetal hemoglobin (HbF). Person with sickle cell trait leads a normal life but the SCD person suffers from various complications throughout the life, such as anemia, recurrent infection, bone and joint pain, joint swelling, osteomyelitis, necrosis of bone, aplastic crises, abdominal pain, splenic sequestration crises, hepatosplenomegaly etc. [3]

Sickle cell anemia is common among people whose ancestors come from Sub Saharan Africa, South America, Central America, India, Saudi Arabia, Cuba and Mediterranean countries.[4]

In India, the sickle cell gene was first described in the Nilgiri hills of Northern Tamilnadu in 1952[5]. Now it is known to be widespread in central and Southern parts of the country. This disease has remained a neglected field of research in India and most of the reports spread a misconception that this is confined to tribal population or some schedule casts only.[4] In Maharashtra, the prevalence of disease varies from 1.9% to 33.5% in different communities with an overall prevalence of 10%.[6] Lehman H et al reported prevalence of 15% among schedule cast population of Gondia District.[5]

This study was conducted to estimate the yield of HPLC (High Performance Liquid Chromatography) in diagnosing Sickle cell disease on the basis of symptoms suggesting sickle cell disease in an endemic area of tribal district of Maharashtra: Gondia.

Material and Method

This retrospective record based study was conducted in a tertiary care institute in Gondia district of Maharashtra. A total of 767 participants were included in this study of all ages and both sex during the period from October 2009 to February 2015. All the patients from Medicine OPD and Pediatric OPD suspected to have the symptoms of sickle cell disease like anemia, early fatigue paleness and shortness of breath, episodic pain, painful swelling of hands and feet, frequent infections, delayed growth, repeated fever, and repeated hospitalization were enrolled for HPLC testing performed in clinical biochemistry laboratory.

Procedure

Venous blood samples were collected in 2.0 ml tubes contacting EDTA and in a tube with a gel separator. The qualitative and quantitative Hemoglobin profiles were investigated by using automated HPLC apparatus. Newborn screening was conducted by dried heel prick blood spots tested by HPLC using the newborn screening machine (Bio-rad laboratories) according to manufacturer instructions.

Cation exchange HPLC has the advantage of quantifying HbF and HbA2 along with hemoglobin variant screening. Hemolysate is injected into a chromatography column containing negatively charged resin onto which the positively charged hemoglobin are adsorbed. As the ionic strength of liquid phrase increases, hemoglobin variants will come out at a specific retention time. This allows identification of Hemoglobin variant based on overall charge of protein and it is monitored by an optical detector. The chromatogram is stored in and analyzed by microcomputer.[7,8]

Data was entered in Microsoft excel and was analysed using SPSS version 20.0. Qualitative data was analysed by using frequency and percentage and quantitative data was represented with mean and standard deviation. Association between two qualitative data was analysed using chi-square test.

Results

In this study, 767 patients with symptoms of sickle cell disease were tested for HPLC for confirmation of sickle cell hemoglobin in a tertiary care center.

Table 1: Number of tests performed year-wise

Year	Frequency	Percentage (%)
2009-10	108	14.1
2010-11	139	18.1
2011-12	182	23.7
2012-13	150	19.6
2013-14	150	19.6
2014-15	38	4.9
Total	767	100.0

Most of the cases were registered in 2011-12 year. As only two month data were considered for the year 2014-15, hence the frequency was less.

Table 2: Distribution of study subjects according to demographic variable:

Variable	Frequency	Percentage (%)
Age group		
less than 15	328	42.8
15-45 years	425	55.4
more than 45 years	14	1.8
Gender		
Female	385	50.2
Male	382	49.8
Category		
SC	428	55.8
ST	79	10.3
Others	260	33.9

The mean age of all the cases were 18.01±10.85 years with a range of 1-60 years. Table 1 showed, majority of the patients belonged to the age group of 15-45 years (55.4%). Males and females have almost equal proportions. Majority of the patients were schedule caste (55.8%).

Table 3: Results of HPLC finding

HPLC Findings	Frequency	Percent
Sickle cell disease (SS)	41	5.35
Sickle cell trait (AS)	34	4.43
Normal	692	90.22

Table 2 showed out of all study subjects, 5.35% of study subjects were affected in sickle cell disease while 4.43% of study subjects were sickle cell trait, while 90.22% were Normal.

Table 4: Distribution Age and Sickle Cell Disease Status.

			HPLC Results			Total
			SS	AS	Normal	
Age group	less than 15	Number	23	10	294	327
		%	7.0	3.1	89.9	100.0
	15-45 years	Number	17	22	387	426
		%	4.0	5.2	90.8	100.0
	more than 45 years	Number	1	2	11	14
		%	7.1	14.3	78.6	100.0
Total		number	41	34	692	767
		%	5.35	4.43	90.22	100.0
Chi square= 8.498, p value= 0.075, non-significant						

Though most of the Sickle cell disease cases were found in the age of less than 15 years (7.0%) but the association between age and HPLC finding was statistically insignificant.

Table 5: Distribution of Gender and Sickle Cell Disease Status.

			HPLC Results			Total
			SS	AS	Normal	
Sex	Female	Number	16	24	345	385
		%	39.0	70.6	49.9	50.2
	Male	Number	25	10	347	382
		%	61.0	29.4	50.1	49.8
Total		Number	41	34	692	767
		%	100.0	100.0	100.0	100.0
Chi square= 7.734, p value= 0.021, significant						

Most of the Sickle cell disease cases were confirmed in males 25(61%) out of 41, while most of the carrier were found in females 24(70.6%) out of 34. This association was statistically significant.

Discussion

Sickle cell disease and hemoglobinopathies are endemic in Central India region. Hemoglobinopathies can present as from, mild anemia to severe complications which can lead to disability and mortality. Potential interactions between two sickle cell carriers may lead to serious homozygous state of sickle cell disease in offspring. The use of cation-exchange high performance liquid chromatography (CE-HPLC) to separate and quantify various normal and abnormal Hb fractions has been increasing day by day. [5] It offers a reliable tool for early, accurate detection.

The present study emphasized the yield of HPLC testing in diagnosing Sickle cell disease on the basis of symptoms suggesting sickle cell disease in a tertiary care center of Gondia, tribal district. A total of 9.78% were found to be having sickle cell disease or trait on HPLC finding.

As regards to sex distribution of the disorder out of all Sickle cell disease cases, 61% were males and 39 % were females. Sickle cell trait was more common in females 70.6% as compared to males 29.4%. Similar results were observed by Deshmukh P et al[4] where sickle cell trait was more common in females (3.0%) as compared to males (2.8%).

Conclusion

The overall yield of sickle cell disease and carrier was found to be 9.78% with higher prevalence of sickle cell disease among males and sickle cell carrier among females. HPLC is best tool for screening of sickle cell disease for early diagnosis and prevention of complications amongst them. However, DNA studies in selected cases are essential for confirmation.

Funding: Nil

Conflict of Interest: Nil

References

1. Sickle Cell Disease | WHO | Regional Office for Africa [Internet]. [cited 2017 Dec 27]. Available from: http://www.afro.who.int/health-topics/sickle-cell-disease
2. Arab H. Sickle Cell Disease and Sickle Cell Anaemia. Am J Med. 2010;1–8.
3. Jain DL, Sarathi V, Desai S, Bhatnagar M AL. Low Fixed-Dose Hydroxyurea in Severely Affected Indian Children With Sickle Cell Disease. Hemoglobin. 2012;36(4):323–32.
4. Garg B, Garg N, Prajapati N, Bharambe M, Deshmukh P. Prevalence of sickle cell disorders in rural Wardha. Indian J Community Med [Internet]. 2006;31(1):26. Available from: http://www.ijcm.org.in/text.asp?2006/31/1/26/54928
5. Lehmann H. Sickle-cell Trait in Southern India. Br Med J. 1952;1(4755):404–5.
6. Neville B, Damm DD, Allen C, Bouquot J. Oral and Maxillofacial Pathology. 3rd ed. International Journal of Human Genetics. Saunders; 2002. 610–8 p.
7. da Fonseca SF, Amorim T, Purificação A,

Gonçalves M, Boa-Sorte N. Hemoglobin A2 values in sickle cell disease patients quantified by high performance liquid chromatography and the influence of alpha thalassemia. Rev Bras Hematol Hemoter [Internet]. 2015 Sep 1 [cited 2018 Jan 22];37(5):296–301. Available from: https://www.sciencedirect.com/science/article/pii/S1516848415000869

8. Sickle Cell Institute Chhattisgarh R. A Hand Book on Sickle Cell Disease. 2014;(771). Available from: www.scic.co.in