VOLUME-9, ISSUE-6, JUNE-2020 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Pathology

PREVALENCE OF SICKLE CELL DISORDER IN JHARKHAND: A RETROSPECTIVE CASE STUDY

Dr. Sona Pathak* Post-Graduate Student, Depatment of Pathology, RIMS, Ranchi. *Corresponding Author

Dr. Mushtaque Ahmad Ansari Associate Professor, Department of Pathology, RIMS, Ranchi.

Dr. Suraj Sinha Post-Graduate Student, Department of Pathology, RIMS, Ranchi.

ABSTRACT **Background:** Sickle cell disease (SCD) is one of the most common monogenic disorders globally with an autosomal recessive inheritance and is most prevalent among people of African, Arabian and Indian origin. Sickle-cell anemia (SCA) is a neglected chronic disorder of increasing global health importance, with India estimated to have the second highest burden of the disease. **Material and Methods:** A retrospective study of 2023 cases was undertaken spanning a period of 20 months from July 2018 to February 2020 who has been screened positive for solubility test and/or NESTROF test and were sent for HPLC at RIMS, Ranchi. **Results:** Among all the cases prevalence of sickle cell disorder was highest i.e. approximately 64%. There is female gender preponderance in sickle cell disorders and Muslim population also contributes a significant number.

KEYWORDS : Sickle Cell Disorder, Solubility Test, Hereditary Disorder Of Haemoglobin , HPLC.

INTRODUCTION

Sickle cell disease (SCD) is one of the most common monogenic disorders globally with an autosomal recessive inheritance. James Herrick, a physician first described the characteristic sickle shaped red cells in a medical student from Grenada in 1910. Linus Pauling and his colleagues showed that sickle haemoglobin (HbS) had an altered electrophoretic mobility and they were the first to define it as a molecular disease in 1949. A few years later in 1957, Vernon Ingram discovered that sickle haemoglobin resulted from a single amino acid substitution in the haemoglobin molecule[1,2]. The disease results from a single base mutation (A to T) in the triplet encoding the sixth residue of the -globin chain, leading to a substitution of valine for glutamic acid and the abnormal haemoglobin S (HbS)[3]. Sickle haemoglobin (HbS) is abnormal haemoglobin, caused by a mutation in the HBB gene and inherited as an autosomal recessive Mendelian trait. Red blood cells (erythrocytes) with HbS become deformed under stress, forming a classic 'sickle' shape. Although heterozygotes (Sickle Cell Trait) are usually asymptomatic, patients who have inherited HbS alleles from both parents suffer from Sickle Cell Anaemia (SCA), the most common and severe form of SCD (a term that technically refers to any condition in which the production of HbS causes symptomatology, and can result from a broad range of inherited HBB mutations) . In patients with SCD, sickling of erythrocytes causes haemolysis, reduces the oxygen carrying capacity of erythrocytes, and can result in episodic microvascular occlusion leading to tissue ischaemia and painful 'crises' with serious and often life-threatening consequences[4,7].

To prevent complications, diagnosis of HbS has to be done as early as possible from the very first day of life. This diagnosis is usually done by isoelectric focusing, or cellulose acetate electrophoresis at alkaline pH, but more and more frequently by cation exchange High Performance Liquid Chromatography (HPLC)[5,9]. It should always be confirmed by a specific test for HbS, such as an electrophoresis on agar gel. In Sickle cell anaemia, it is important to evaluate the other factors that may modify the presentation (mostly HbF level and associated Alpha-Thalassemia). In a heterozygous subject presenting with pathological manifestations, more sophisticated biochemical tests or molecular biology investigations may be necessary to determine the cause of the disease[5].

FIG. 1:HPLC BIO-RAD VARIANT-II SYSTEM

MATERIALS AND METHODS

It was a retrospective record based study, performed in the Department of Pathology, RIMS, Ranchi. Study Population included all cases of Hereditary haemoglobin disorders presenting from July 2018 to February 2020 (20 months) screened positive by solubility test and NESTROFT (Naked Eye Single Tube Red Cell Osmotic Fragility Test) and subsequently subjected to High Performance Liquid Chromatography(HPLC BIO-RAD variant-II SYSTEM). Study Procedure involves case reports having patient sex, religion and type of hereditary haemoglobin disorder. The epidemiological data of hereditary haemoglobin disorder were compared and analyzed.

RESULT AND DISCUSSION

In our present study of 2023 cases which were found positive in screening test, 341 (16.85%) were found to be HPLC negative for hereditary haemoglobin disorders in HPLC. 1294(64%) were diagnosed as Sickle cell disorder. Data shows relative prevalence of other hereditary haemoglobin disorder as shown in Table-1. Among a total of 2023 patients, 301(15%) cases of Beta Thalassemia trait, 63(3%) cases of Beta Thalassemia major,16(0.8%) cases of HbF, 3(0.14%) cases of

HbE trait, 2(0.09%) cases of each HbD trait and HbE Beta Thalassemia and 1(0.05%) case of HbE disease.

TABLE – 1 Case-wise Prevalence

Disease	No. Of patient	Percentage (%)
Sickle cell trait	1120	55.36
Sickle cell disease	64	3.16
Sickle Beta Thalassemia	110	5.43
Beta thalassemia trait	301	14.87
Beta thalassemia major	63	3.11
HbE Trait	3	0.14
HbE beta Thalassemia	2	0.09
HbE disease	1	0.05
HbD Trait	2	0.09
HbF	16	0.8
Normal	341	16.85
Total	2023	100

Out of 1294 cases of sickle cell disorders, 1120 (87%) suffered from sickle cell trait, 64 (5%) suffered from sickle cell disease while 110(8.5%) were sickle beta thalassemia.

Table – 2 Sex-wise Prevalence

Disease	Prevalence (no.)		
	Male	Female	Total
Sickle Cell Trait	217	903	1120
Sickle Cell Disease	21	43	64
Sickle beta Thalassemia	35	75	110
Total	273	1021	1294

Further result showed that among 1294 overall sickle cell patients female contributed 1021 (79%) and male 273 (21%) cases as shown in Table-2. It can be seen that female prevalence of Sickle cell trait, Sickle cell disease as well as Sickle Beta Thalassemia is higheri.e. 80%, 67% and 68% respectively as compared to males.

TABLE – 3 Prevalence In Muslims

Disease	Prevalence(no.)		
	Muslim	Non-Muslim	Total
Sickle Cell Trait	304	816	1120
Sickle Cell Disease	17	47	64
Sickle beta Thalassemia	27	83	110
Total	348	946	1294

In our study, Muslim population comprises 27% cases of Sickle Cell disorder which is significantly high proportion as compared to their share of 14.5% in total population of Jharkhand[6].This could be attributed to the consanguineous marriages prevalent in Muslim community. Ahsana et. al.[8] also reported high prevalence of sickle cell disorder in female and muslim community.

CONCLUSION:

Sickle cell disorders are most common type of hereditary haemoglobin disorder comprising of 64% of all cases. Sickle cell trait alone makes 55% of all of hereditary haemoglobin disorder. In our study female gender was more commonly affected than males. Similarly prevalence among Muslims was more common than non-Muslims as compared to their population statistics. High prevalence communities can be screened for sickle cell disorders and counseled so that consanguineous marriages as well as marriage among carriers can be avoided.

REFERENCES:

[1] Stuart, M. J., Nagel, R. L.(2004). Sickle cell disease. Lancet. 364, 1343–60. [PubMed] [Google Scholar]

[2] Rees, D., C., Williams, T., M., Gladwin, M.,T(2010). Sickle cell disease. Lancet. 376, 2018–31.[PubMed] [Google Scholar]

[3] Colah, R. B., Mukherjee, M. B., Martin, S., & Ghosh, K. (2015). Sickle cell disease in tribal populations in India. The Indian journal of medical research, 141(5), 509–515. https://doi.org/10.4103/0971-5916.159492

[4] Wastnedge, E., Waters, D., Patel, S., Morrison, K., Goh, M. Y., Adeloye, D., & Rudan, I. (2018). The global burden of sickle cell disease in children under five years of age: a systematic review and meta-analysis. Journal of global health, 8 (2),021103.m https://doi.org/10.7189/jogh.08.021103

[5] Wajcman. H.(2004). Diagnostic et dépistage de la drépanocystose [Diagnosis and screening of sickle cell disease]. La Revue du praticien. 54(14), 1543-7.

[6] Census of India 2011. Office of the Registrar General and Census Commissioner. Ministry of Home Affairs, Govt of India. [accessed on March 27,2015]. Available from: http://www.censusindia.gov.in .

[7] Chakravorty S., Williams T.,N. (2015). Sickle cell disease: a neglected chronic disease of increasing global health importance. Arch Dis Child. 100. 48–53. doi: 10.1136/archdischild-2013-303773. [PMC free article] [PubMed] [CrossRef] [Google Scholar]

[8] Shah, A., Hussaain, R., Fareed, M., Afzal, M. (2012). Gene frequency of sickle cell trait among Muslim populations in a malaria belt of India,i.e., Manipur. Egyptian journal of medical human genetics, 13(3),323-330. doi.org/10.1016/j.ejmhg.2012.04.001

[9] Surve, R. R., Mukherjee, M. B., Kate, S. L., Nagtilak, S. B., Wadia, M., Tamankar, A. A., Ghosh, K., Colah, R. B., & Mohanty D. (2000). Detection of the beta S gene: an evaluation of the solubility test against automated chromatography and haemoglobin electrophoresis. Br J Biomed Sci. 57(4), 292–4. [PubMed] [Google Scholar]