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A RARE CASE OF SICKLE CELL TRAIT WITH SEVERE ANEMIA WITH MEGALOBLASTOID CHANGES IN TRIPURA : CASE REPORT

Medical Biochemistry

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ABSTRACT

Sickle cell anemia is an inherited disorder caused by a point mutation in gene that encodes the β-globin chain of hemoglobin. The mutation results in replacement of glutamate by valine. The mutant hemoglobin (HbS) polymerizes red blood cells & cause blockage, resulting in acute, severe pain called a sickle cell crisis. Sickle cell diseases (SCDs) is an emerging public health challenge, not only in India but also globally. In India, SCD is distributed geographically in the central and western regions. Here we are presenting a rare case of Sickle Cell Trait with Severe Anemia with Megaloblastic changes in a middle aged man in Tripura. A Male patient in his forties came to the OPD with severe lethargy & generalised weakness. Patient had Hypoalbuminemia, Anemia with Marked Anisopoikilocytosis. Bone marrow showed Megaloblastoid changes. After evaluation of HPLC Chromatogram of Hemoglobin Electrophoresis with a Sickle Window of 15.7 %, HbA2 2.6%, HbF <0.8%, HbA0 71.6% was diagnosed as Sickle Cell Trait by the Authors. Although Sickle Cell Trait being the benign form of the disease, here it presented with severe Anemia. Hypoalbuminemia also supports the finding of Pedal Edema. Treatment provider we should always look for hematological status of the patient & encourage the patient for treatment of Anemia.

KEYWORDS

Sickle cell anemia, Sickle cell diseases, Sickle Cell Trait, Megaloblastoid changes, Bone Marrow Biopsy, Anemia, Complications.

INTRODUCTION

Sickle cell anemia is an inherited disorder caused by a point mutation (affecting a single nucleotide) in the gene that encodes the β-globin chain of hemoglobin (Hbβ). The mutation results in the replacement of negatively charged glutamate by a neutral, hydrophobic valine that produces sticky patches on the protein surface. Upon delivering oxygen to the tissues, the mutant hemoglobin (HbS) polymerizes into fibers, which distort ("sickle") red blood cells and cause blockage of the circulation, resulting in acute, severe pain called a sickle cell crisis. [1] Sickle cell diseases (SCDs) is an emerging public health challenge, not only in India but also globally. It has been estimated that, between 2010 and 2050, about 14.2 million babies will be born with sickle cell anemia (SCA). [2] In India, SCD is distributed geographically in the central and western regions. [3]

It has been estimated that about 5200 live births have SCD every year. The prevalence of sickle cell gene is as high as 5%–34% in various scheduled tribes (STs), who are socioeconomically disadvantaged and are frequently medically underserved. [4] Sickle cell trait is benign because patients do not get vaso-occlusive crisis; they have a better quality of life and mortality is the same as the rest of the general population. [5] Here we are presenting a rare case of Sickle Cell Trait with Severe Anemia with Megaloblastic changes in a middle aged man in Tripura.

METHODOLOGY

Blood samples were collected by Venepuncture for testing of Hemoglobin Electrophoresis, Biochemical parameters. Hemoglobin Electrophoresis was done by HPLC Method by BIORAD D-10, Biochemical tests were done by Full Automated Biochemistry Analyzer XL 640 by Erba & Serum electrolytes by ISE Method by Easylite analyzer. Other tests like Complete Blood Count, Bone Marrow Biopsy were done.

Case Study

A Male patient in his forties, daily labourer by profession came to the OPD of Agartala Govt. Medical College & GBP Hospital, Tripura with severe lethargy & generalised weakness. On examination he was found to be severely pale & was admitted in ward. The patient had a history of multiple blood transfusions in the past. Patient also complained of pain in his calves & Pedal edema.

RESULTS

Biochemical & Hematology Reports are given below:

Peak table - ID: 1

Peak	R.time	Height	Area	Area %
Unknown	0.14	4438	9238	0.3
A1a	0.20	4869	13133	0.5
A1b	0.28	5176	27692	1.0
F	0.46	2761	16291	< 0.8 *
LA1c/CHb-1	0.63	4519	33953	1.3
A1c	0.81	7437	75376	4.9
Unknown	1.17	1005	10552	0.4
P3	1.50	13673	98140	3.6
A0	1.69	341174	1932894	71.6
A2	3.25	3312	57706	2.6
S-Window	4.11	102981	423957	15.7
Total Area:		2698931		

Fig: Chromatogram of HPLC-Hb Electrophoresis

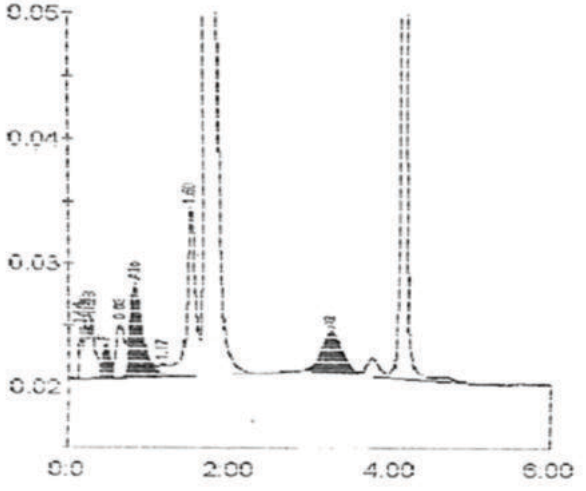

Fig: Hemoglobin Electrophoresis

Biochemical Tests:

Test Name	21/12/2023	22/12/2023	Reference range
Blood Urea	35	36	Upto 50
Sr. Creatinine	0.9	0.8	Upto 1.5

26/12/2024		
Test Name	Result	Reference range
Random Blood Sugar	109	Upto 140

Liver Function Test				
Test Name	21/12/2023	22/12/2023	26/12/2023	Reference Range
Total Bilirubin	0.8	0.9	0.9	0.2 - 1.2 mg/dL
Conj. Bilirubin	0.2	0.1	0.2	0 - 0.30 mg/dL
Unconj. Bilirubin	0.6	0.8	0.7	0.2 - 1.0 mg/dL
SGOT (AST)	10	09	13	Upto 40 IU/L
SGPT (ALT)	11	18	22	Upto 40 IU/L
GGT	20	20	18	3 - 22 IU/L
ALP	223	240	228	54 - 369 IU/L
Total Protein	6.8	6.0	6.7	6 - 8 gm/dL
Albumin	2.8	2.6	2.7	3 - 5 gm/dL
Globulin	4.0	3.4	4.0	2 - 4 gm/dL
A:G Ratio	0.6	0.76	0.7	0.9 - 2.5

Lipid Profile			
	21/12/2023	30/10/2023	Reference Range
Total Cholesterol	70	71	120 - 200 mg/dL
Serum Triglyceride	34	30	06 - 175 mg/dL
Serum HDL	20	22	30 - 70 mg/dl
Serum LDL	42	40	70 - 130 mg/dl
Serum VLDL	08	09	12 - 35 mg/dl

Electrolytes			
	21/12/2023	26/12/2023	Reference Range
Sr. Sodium	138	136	135-142 mmol/L
Sr. Potassium	3.4	4.0	3.5-5.5 mmol/L

Complete Blood Count			
	21/12/2023	22/12/2023	26/12/2023
Hb%	1.6 gm%	1.7 gm%	3.1
PCV	5.6	6.3	11.4
Platelet Count	1 Lakh	0.8 Lakh	1.3
TLC	2800	3200	3100
N	53	75	68
L	45	25	32
M	1	0	0
E	1	0	0
B	0	0	0
RBC Morphology	Marked Anisopoikilocytosis	Anisopoikilocytosis & Hypochromic	Moderate Anisopoikilocytosis

27/12/2023

Mone Marrow Biopsy:

Megaloblastoid changes with Erythroid Hyperplasia with predominance of early & intermediate normoblast.

DISCUSSION

After evaluation of the HPLC Chromatogram of the Hemoglobin Electrophoresis with a Sickle Window of 15.7 %, HbA2 2.6%, HbF <0.8%, HbA0 71.6% with Marked anisopoikilocytosis & without any Sickle cell in the peripheral blood smear was diagnosed as Sickle Cell Trait by the Authors. Although this much severe Anemia is also rare in such cases.[5] This much severe anemia may be attributed to Megaloblastic changes in marrow. There are instances of Folic Acid Deficiency causing Anemic Sickle Cell Crisis.[6] Studies have also shown that sickle cell is associated with exertional rhabdomyolysis. [5] which supports the diagnosis as per patient's complaint. Hypoalbuminemia also supports the finding of Pedal Edema. Patient belonged to the Tribal community among whom these hemoglobinopathies are common [4] , supporting the diagnosis of Sickle Cell Trait.

CONCLUSION

Although Sickle Cell Trait being the benign form of the disease, here it presented with severe Anemia. Even after getting multiple blood transfusions, hemoglobin level rise was pretty slow. So, as the Treatment provider we should always look for hematological status of the patient & encourage the patient for treatment of Anemia through Oral Iron supplementation to Blood transfusion in rare cases.

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