Digital Journal of Clinical Medicine

Volume 2 | Issue 1 Article 10

1-1-2020

# Sickle Cell Disease

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## Recommended Citation

Bhargava, Vibhu and Barve, Dr. Sandeep (2020) "Sickle Cell Disease," *Digital Journal of Clinical Medicine*: Vol. 2: Iss. 1, Article 10.

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# Sickle Cell Disease

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## CLINICAL HISTORY:

29 year old male patient came with the complaints of abdominal pain since 3 months, insidious in onset, non-progressive. He also gives history of low grade fever and cough. Cough is associated with expectoration, whitish to yellowish in color, mild in quantity, not blood tinged, not foul smelling. History of 2-3 episodes of vomiting since 1 week, non-bile tinged.

No history of joint pain, breathlessness and fatigue.

Past history: The patient is a known case of sickle cell disease diagnosed in childhood.
He has undergone multiple blood transfusions since childhood.
No history of diabetes mellitus, hypertension, thyroid disorders or cardiac disorders.

## EXAMINATION AND INVESTIGATIONS:

General Physical Examination:
Febrile. (99.2 F)
Pallor and icterus present.
No generalised lymphadenopathy.
Systemic Examination:

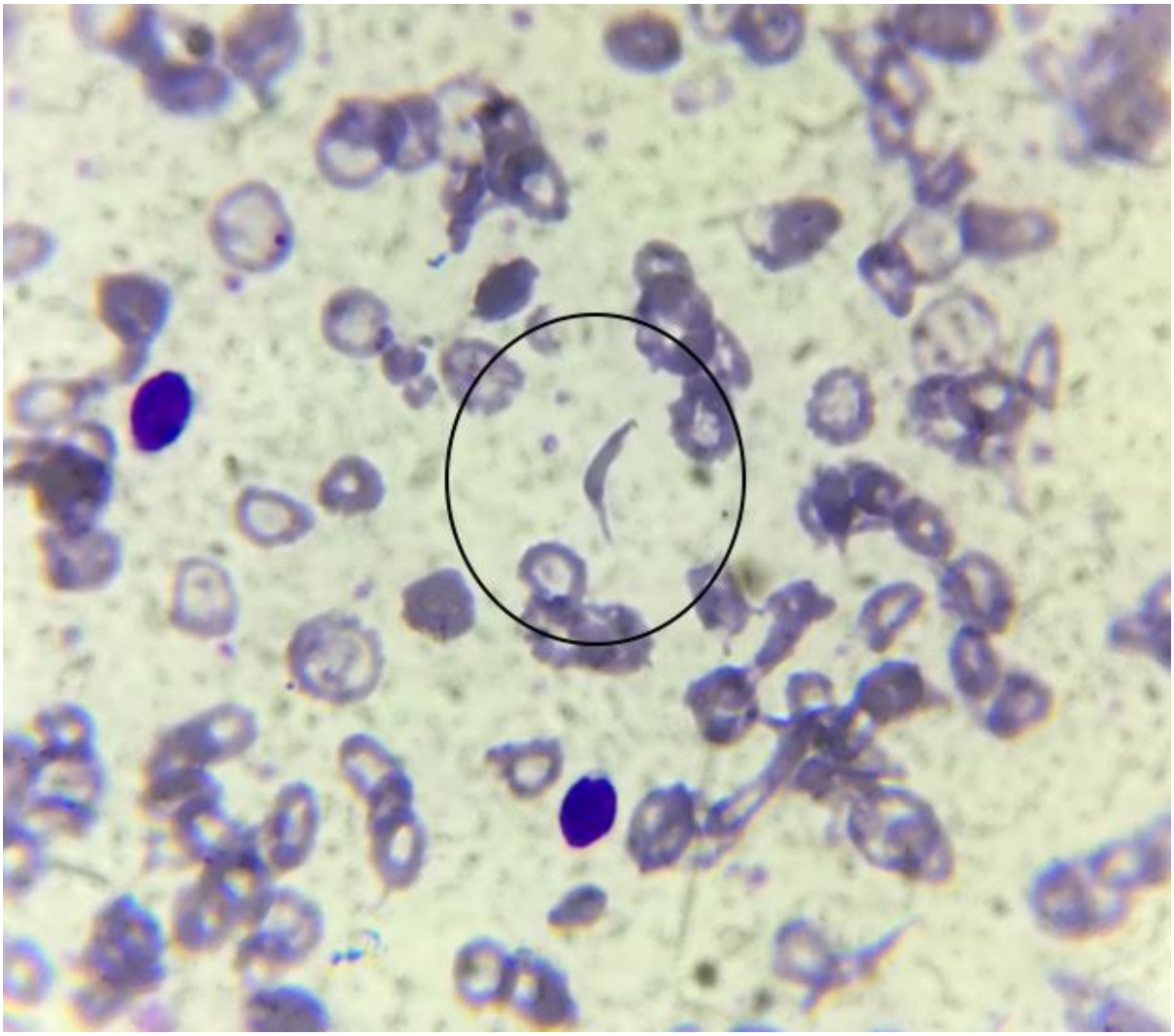
Abdomen:
Inspection: Umbilicus central. No distention visible
Palpation :
Tenderness present in the epigastric region. Mild tenderness present in RUQ on deep palpation.
Murphy’s sign negative.
Percussion: Normal. No Signs of free fluid.
Auscultation. Normal bowel sounds heard.
CVS: S1, S2 heard.No murmurs.

RS: NVBS heard on the right side. Decreased breath sounds over the left lung. No added sounds.

CNS: Normal.

Blood Investigations:

Hb:9.1gms/dl
Total WBC:
15,220/mcl
N:71%, L:20%
RBC:4.16 million/mcl
MCV:69.8%
MCH:21.9%
MCHC:31.4%
RDW-CV:17.5%
Platelet:53000/mcl
Peripheral Blood Smear:
Red blood cells are normocytic normochromic to mildly hypochromic.
Moderate degree of anisopoikilocytosis noted.
Sickle cells seen. (Marked in black)
(Insert Image 1)

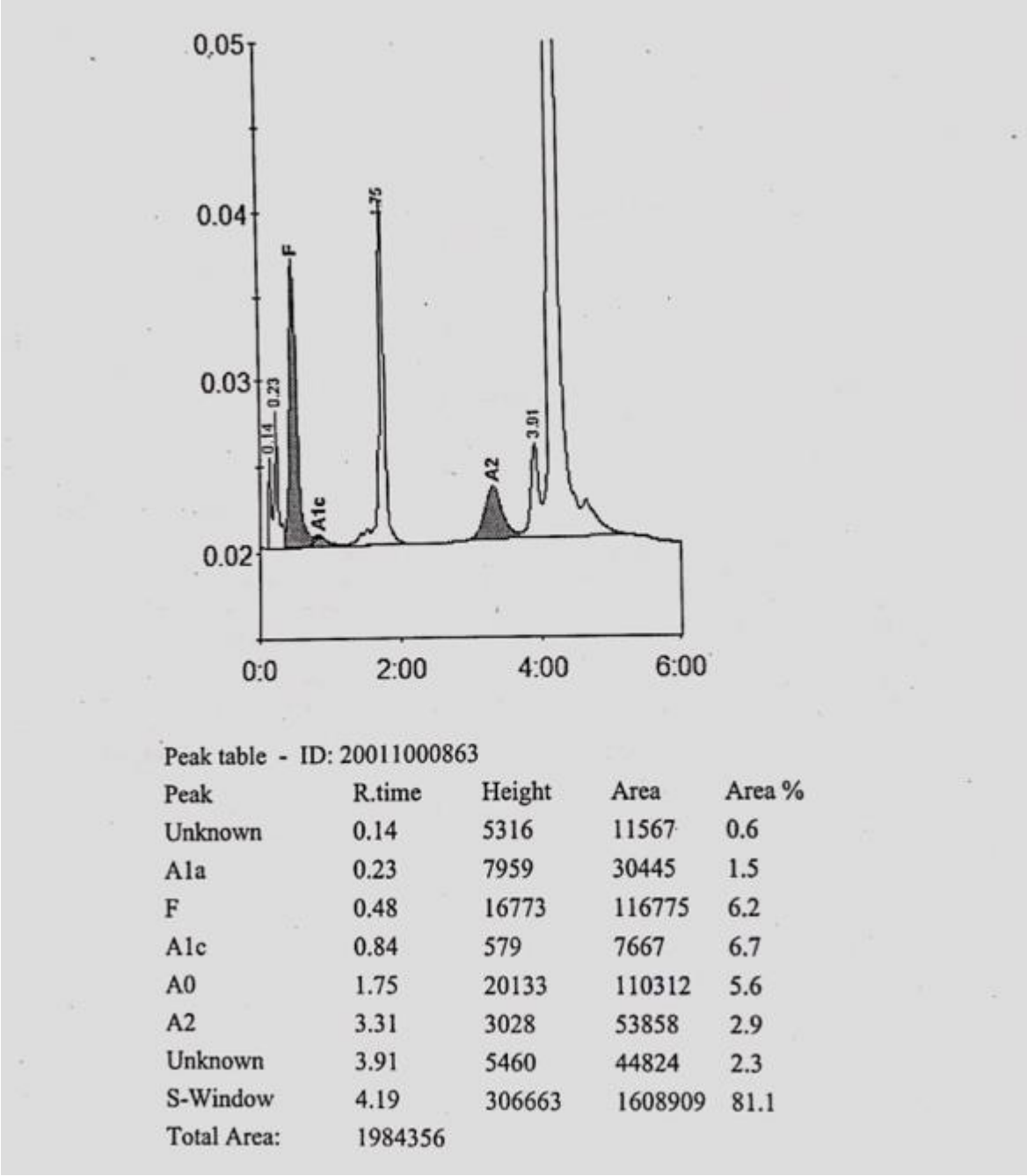


Solubility Sickling Test:
Positive
(Insert Image 2)

1.

Hb analysis by HPLC

Hb A:9.8%
Hb A2: 2.9%
Hb F: 6.2%
Hb S: 81.1%
Findings are suggestive of SICKLE CELL SYNDROME.



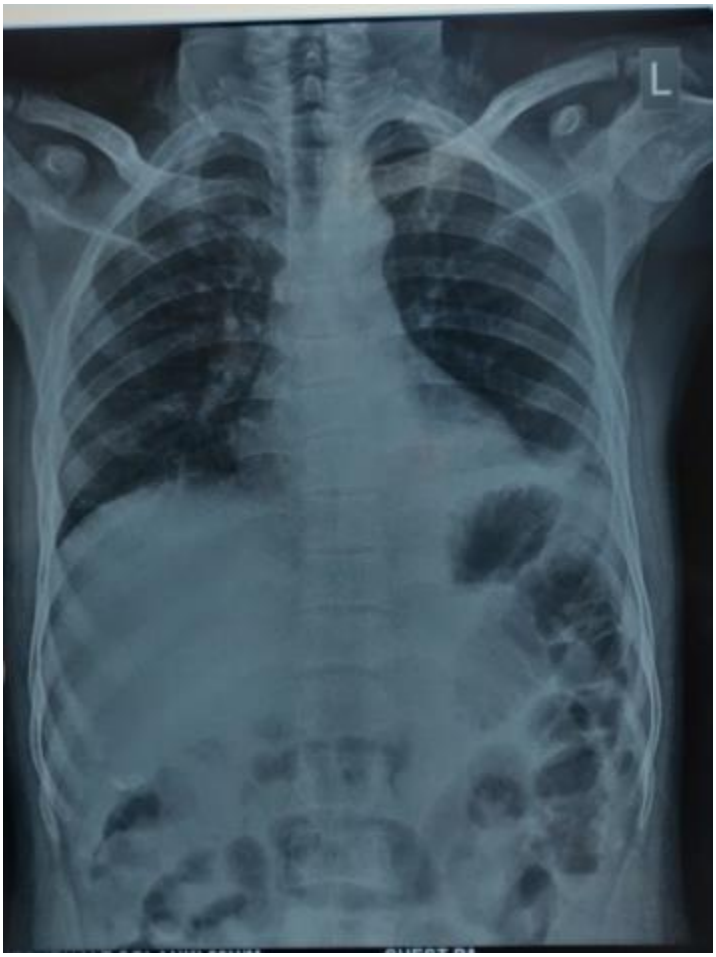

Peak table - ID: 20011000863

| Peak | R.time | Height | Area | Area % |
|---|---|---|---|---|
| Unknown | 0.14 | 5316 | 11567 | 0.6 |
| A1a | 0.23 | 7959 | 30445 | 1.5 |
| F | 0.48 | 16773 | 116775 | 6.2 |
| A1c | 0.84 | 579 | 7667 | 6.7 |
| A0 | 1.75 | 20133 | 110312 | 5.6 |
| A2 | 3.31 | 3028 | 53858 | 2.9 |
| Unknown | 3.91 | 5460 | 44824 | 2.3 |
| S-Window | 4.19 | 306663 | 1608909 | 81.1 |
| Total Area: | | 1984356 | | |

1.

(Insert Image 3)

X-Ray Chest (PA view)
Blunting of left costophrenic angle. s/o left pleural effusion.
(Insert image 4)




1.

MDCT Abdomen & Pelvis:
Acute pancreatitis with intrapancreatic pseudocysts.
Cholelithiasis.
Left mild to moderate pleural effusion..

**FINAL DIAGNOSIS:**

SICKLE CELL DISEASE WITH PLEURAL EFFUSION AND CHOLELITHIASIS

**DISCUSSION:**

NJ CEFTRIAXONE 1 GM IV x 7 days
TAB FOLVIT (Folic acid) x 5 days
TAB PANTOPRAZOLE 1-0-1 X 20 days
Pleural tapping was advised SOS.

The patient refused further interventions, hereafter.

Sickle cell disease is a common hereditary hemoglobinopathy caused by a point mutation in B globin that promotes the polymerisation of deoxygenated haemoglobin leading to red cell distortion, microvascular obstruction, haemolytic anaemia and ischemic tissue damage.1

The replacement of β6 glutamic acid with valine results in a hydrophobic interaction of sickle hemoglobin (hemoglobin S) with another hemoglobin molecule in conditions of low oxygen availability, trigger an aggregation into large polymers. The polymerization of deoxygenated hemoglobin S is the primary event in the molecular pathogenesis of sickle cell disease, resulting in a distortion of the shape of the red cell (banana or sickle shaped) and a marked decrease in its deformability. These rigid cells are responsible for the vaso-occlusive phenomena that are the hallmark of the disease.2

SCD is caused by genetic and non-genetic factors. The genetic cause is characterized by the presence of abnormal erythrocytes damaged by HbS, a variant of normal adult hemoglobin (HbA), inherited either from both parents (homozygous for the HbS gene) or with β-thalassemia (compound heterozygosity) or from one parent, along with another hemoglobin variant, such as hemoglobin C (HbC). The non-genetic factors include climate, air quality, as well as socioeconomic factors. 2,3

Dactylitis is the most common initial symptom, noted in children in the first 2 years of life whereas painful crisis is the first symptom frequently seen in patients after the age of 2 years. The most common nonspecific symptom is pneumonia.4

Infection is a major determinant of the outcome in patients with sickle cell disease. Splenic dysfunction has a key role in the increased susceptibility to bacterial infections seen in children with sickle cell disease, and pneumococcal and haemophilus infections.3

The complications of sickle cell disease are varied ranging from vaso-occlusive crisis to acute chest syndrome, and pulmonary hypertension.5 A dreaded complication of formation of silent infarcts due to adherence of sickle cells to the endothelium, causing activation of the inflammatory cascade and clotting factors resulting in in a nidus for thrombus formation can lead to severe motor and cognitive impairment.6

Clinical outcomes have gradually improved over the years, mostly as a result of developments in supportive care and treatment with hydroxyurea, which stimulates hemoglobin F production.2 Penicillin prophylaxis in children, primary stroke prevention, regular blood transfusions are main modalities of treatment and prevention. 4
Hematopoietic stem-cell transplantation is potentially curative, although its use

is restricted by the high cost, toxicity, and limited availability of suitable donors.3

## CONCLUSION:

We reviewed a case of a 29-year male, adult who presented with acute pain abdomen and additional history of low grade fever . This report illustrates the importance of having valid differentials for pain abdomen in an acute setting and performing the required valid blood investigations.prompt treatment by medical line of management at diagnosis to improve outcome and prevent complications recurrent infection and vaso occlusive complications.

**ACKNOWLEDGEMENTS:**None

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