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Sickle Cell Disorder: Unraveling the Diverse Clinical Presentations

Dr. Y Veda Parimala[1], Dr. D Ravi Teja[2]

[1] Junior Resident, Department of General Medicine, MRIMS

[2] Junior Resident, Department of General Medicine, MRIMS

Abstract: ***This study investigates the clinical presentations of sickle cell disorders (SCD) among patients admitted to the General Medicine department at Mallareddy Institute of Medical Sciences. The research encompasses a cross - sectional hospital - based study involving ten patients, aged 15 to 49, diagnosed with sickle cell disease (SCD) through hemoglobin electrophoresis. The study reveals diverse clinical manifestations, with 70% of patients being male and 50% aged between 21 - 30 years. Among the participants, 80% were homozygous (HbSS) while 20% were heterozygous for SCD. The hemoglobin levels varied, with 33% having Hb <6.5 gm/dl, and 40% in the 6.5 - 10 gm/dl range. Clinical symptoms ranged from asymptomatic cases to severe conditions such as recurrent infections, leg ulcers, acute chest syndrome, avascular necrosis, and osteomyelitis. The findings highlight the varied severity and presentation of SCD, emphasizing the need for targeted medical interventions and continuous monitoring to manage this genetic disorder effectively. The study aligns with previous research, suggesting a higher prevalence of complications among male patients and the need for comprehensive care strategies.***

Keywords: sickle cell disorder, hemoglobinopathies, clinical features, anemia, patient study

1. Introduction

The discovery of sickle cell anemia can be traced back to the 19th century, when a dental student experiencing recurring fever, jaundice, leg ulcers, and shortness of breath underwent a routine blood test. The test revealed that his red blood cells were sickle - shaped, a finding that, combined with similar observations of abnormal red blood cells in other patients with varying symptoms, ultimately led to the identification of sickle cell anemia as a distinct medical condition.1

Sickle cell anemia is a kind of hemoglobinopathy.

The molecular biology of hemoglobinopathies is well understood, but clinical progress in treatment has been limited. The term 'sickle cell disorder' refers to conditions where red blood cells sickle in response to deoxygenation, rather than the specific genotype (such as HbSS, S/beta thal, or SE). These disorders are characterized by chronic hemolysis and anemia, with patients experiencing periods of relative well - being punctuated by episodes of illness. The severity of clinical manifestations varies widely among patients, although sickle cell anemia (HbSS) is the most severe form. Notably, there is significant overlap in clinical behavior among these diseases[3]

The main defect in SCA is the replacement of glutamate — valine on the 6 th position of globin chain.2

Sickle cell anemia, defined as homozygosity for the sickle hemoglobin mutation (α2 βS 2; glutamic acid [E] 7 valine [V] GAG - GTG), is the most common of these genotypes, followed by HbSC disease or compound heterozygosity for HbS and HbC (α2 βC 2; E 7 lysine [K] GAG - AAG) genes[2]
Sickle cell trait: Inheritance of only one HbS allele is termed sickle cell triat (HbAS). The percentage of HbA is always higher (60 percent) than HbS in sickle cell trait.2

Clinical presentation of sickle cell disease is varied ranging from asymptomatic, mild to being fatal involving anemia, acute chest syndrome, pulmonary edema, dactylitis, leg ulcers, osteonecrosis, osteomyelitis, CAD, Stroke, renal failure and many more.3

2. Aims and Objectives of the Study

To identify different clinical presentations of Sickle cell disorders.

3. Materials and Methods

Study design - Hospital based cross sectional study

Study Population: Patients admitted in the General Medicine department at Mallareddy Institute of Medical Sciences.

Place of Study: MALLAREDDY INSTITUTE OF MEDICAL SCIENCES

Sample Size: 10

Study Duration: 18 months from September 2022 to March 2024.

Inclusion Criteria:
1) Age between 15 - 49 years of both the genders.
2) Patients who have been diagnosed to have sickle cell disease by Hb electrophoresis.
3) Those who are willing to participate in the study.

Exclusion Criteria:
1) Patients age lesser than 15 years of age or older than 60 years of age.
2) Pregnant female

3) Patients with coexisting haemoglobinopathy along with sickle cell disease have been excluded.
4) Patients who are not willing to participate in the study.

Patients visiting the General medicine department of MRIMS were selected after taking written informed consent from the patient according to the inclusion and exclusion criteria, and a detailed history regarding the present, past and family history was taken and a thorough clinical examination was done and were subjected to necessary investigations depending upon the clinical profile of the patient.

4. Results

A total of 10 patients who were satisfying the inclusion and exclusion criteria and attending the department of general medicine were studied.

Out of 10, 7 were male and 3 were female.

Among 10 patients, 2 belonged to age group 15 - 20 years, 5 were from 21 - 30 years, 3 were from 31 - 40 years.
Out of 10 patients, 8 were homozygous (HbSS), 2 were heterozygous were sickle cell disease.

Anemia - 3 patients had Hb<6.5gm/dl, 4 patients had Hb 6.5 - 10gm/dl, 2 patients had Hb 10 - 12 gm/dl, while the other Hb>12gm/dl haemoglobin levels.

Among 10 patients,
2 - asymptomatic were incidentally detected
3 had history of recurrent infections and hospital admissions
2 had history of leg ulcers and dactylitis
2 had presented with acute chest syndrome
1 patient had avascular necrosis of the femur, osteomyelitis was heterozygous to sickle cell.

5. Discussion

Gender Distribution
The gender distribution of the study population reveals that about 70 % of the patients were males, while 30% females. Sickle cell disease is an autosomal recessive disorder having an equal probability of transmission to both males and female but studies have shown that there is slightly higher risk of male population developing complications therefore the disease is identified in males when compared to females who remain asymptomatic relatively.

The study also is in consistent with the study "*Do gender differences influence the prevalence of sickle cell disorder and related morbidities " done by* Dr Akre Charuhas and others and published in International journal of collabarative research on Internal Medicine and Public health in 2013 and "*Gender differences in complications of SCA" published in International journal of medicine and health development in JAN - MARCH 2022.* [4, 5]

Age Wise Distribution
In this study 20 % of the study population belonged to 15 - 20 years age group, 50% belonged to 21 - 30 years and the rest 30% belonged to 31 - 40 years.

Sickle Cell Anemia v/s Sickle Cell Trait
Out of 10 SCD patients of the study Hb electrophoresis revealed that 80 percentage were homozygous while 2 were heterozygous type. According to studies, it has been proven that although the incidence of sickle cell trait is more common when compared to sickle cell disease in the general population, the incidence of complications is more in sickle cell anemia (HbSS) population leading to SCA patients seeking medical care more frequently.

Hb levels in SCD patients:
The study revealed that out of 10,
Hb<6.5 gm/dl was there in 33% patients,
40% with Hb range 6.5 - 10 gm/dl
20% with Hb range 10 - 12 gm/dl
10% with Hb >12 gm/dl

The findings in the study is supported by an article "TITLE - Analysis of clinical profile, hematological factors burden in SCD patients" published in Lancet journal in JULY 2021.6

Clinical Presentation:
In our study population with SCD patients had varied clinical presentations ranging from

- Asymptomatic cases (no noticeable symptoms) - 20%
- Recurrent infections and hospital admissions - 30%
- Leg ulcers and dactylitis (inflammation of the fingers or toes) - 20%
- Acute chest syndrome (a serious condition that can cause chest pain, fever, and difficulty breathing) - 20%
- Avascular necrosis (death of bone tissue due to lack of blood supply) - 10%
- Osteomyelitis (infection of the bone) - 10%

Apart from the above mentioned symptoms SCD can present with various other complications as well which includes: [7, 8]

Renal medullary cancer
Hematuria
Renal papillary necrosis
Hyposthenuria

Splenic infarction

Exertional rhabdomyolysis
Exercise - related sudden death
Protection against severe falciparum malaria
Complicated hyphema

Venous thromboembolic events

Fetal loss/demise Low birth weight
Acute chest syndrome

Asymptomatic bacteriuria in pregnancy
Proliferative retinopathy

Unlikely or Unproven Associations Stroke
Cholelithiasis

Priapism
Leg ulcers

Avascular necrosis of the femoral head

For early detection and prevention of complications in SCD the Government of India has hiven few guidelines with the aim of elimination of SCD as a public health problem in India before 2047.9

6. Conclusion

Sickle cell disease is a genetic disorder that affects hemoglobin production and can cause a range of health issues.

Being heterozygous for sickle cell means that the patient has one copy of the mutated gene that causes sickle cell disease, but also has one normal copy. This can still cause some health issues, but typically not as severe as those experienced by people with two copies of the mutated gene (homozygous).

In this study, the study population was 10 with 70% males, around 50 % belonged to the age group 21 - 30 years. Almost all the patients except one had anemia, clinical presentation was varied from being asymptomatic, having history of recurrent infections, with hospital admissions, leg ulcers, dactylitis, acute chest syndrome, avascular necrosis of femur, osteomyelitis, nephrotic syndrome.

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