Multiple complications in sickle cell anaemia

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Abstract

Sickle Cell Disease (SCD) is a structural haemoglobinopathy which is extremely diverse in its presentation regarding disease severity and organ involved. The homozygous form if poorly managed gives rise to numerous life threatening conditions which are otherwise avoidable. Here we report the case of a male adolescent with homozygous SCD who presented with haemolytic anaemia, massive ascites, hepatomegaly and multiple fractures secondary to severe malnourishment associated with the disease.

Keywords: Hemoglobinopathies, Sickle cell anemia, Hepatomegaly.

Introduction

Sickle Cell Disease (SCD) is a chronic progressively debilitating disease with haemolytic anaemia and recurrent acute vaso-occlusive events. An estimated 3.2 million people have sickle-cell disease with about 80% cases occurring in Africa.[1] About 0.5 to 1 percent of the Pakistani population carries haemoglobin S or haemoglobin E; nevertheless the exact prevalence is not established.[2]

Local literature contains few case reports about compound heterozygous states like Sickle-β thalassemia and Sickle-Haemoglobin D Punjab. Here we report the case of a 13 yr old adolescent with homozygous SCD who presented with multiple pathologies and severe malnutrition seen in poorly managed cases.

Case Report

A 13 year old male child, Balochi by ethnicity, resident of Thatta, interior Sindh, diagnosed with Sickle Cell Disease with genotype SS, at the age of 2 years, presented to our Emergency Department in August 2016 with a 12-days history of severe abdominal distension, shortness of breath and yellow discoloration of sclera. Since the diagnosis the child was on irregular follow ups. There was history of multiple falls since 2013 leading to limited activity. Patient's two siblings diagnosed with SCD had died earlier. His parents had a consanguineous marriage.

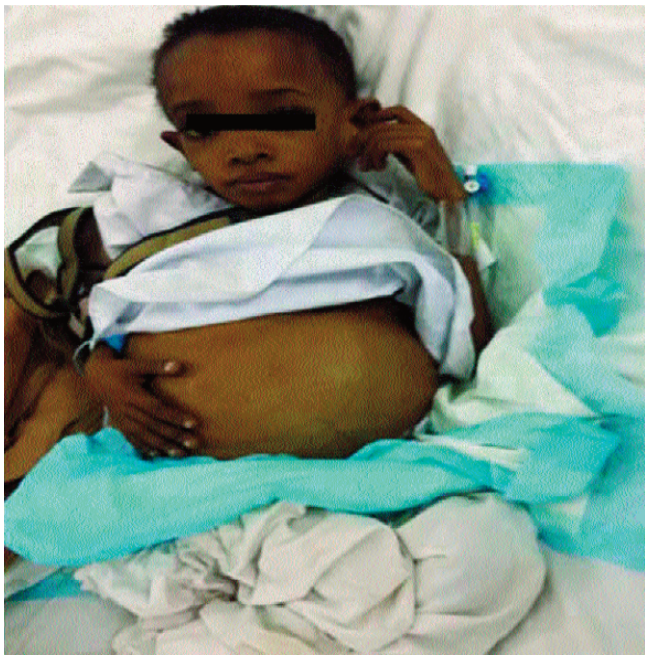

Figure-1: Picture shows marked abdominal distension and prominent abdominal veins. Severe malnourishment can be seen in muscle wasting and lower limb contractures. Digital clubbing and dactylitis is also.

On examination the child was lethargic, irritable and severely malnourished. He was markedly pale, deeply jaundiced, with grade III digital clubbing and massive abdominal distension His vitals showed blood pressure: 100/50mm Hg, pulse rate: 120 beats/min, respiratory rate: 100/min and fever was documented at 100°F.

Abdominal examination revealed a markedly distended abdomen with prominent dilated veins, a positive fluid thrill and enlarged liver was palpable approximately 10 cm the right sub costal margin. Review of musculoskeletal system showed prominent muscle wasting in both upper and lower limbs with bilateral knee joint contractures. Further musculoskeletal examination was compromised due to the above mentioned limitations. Chest

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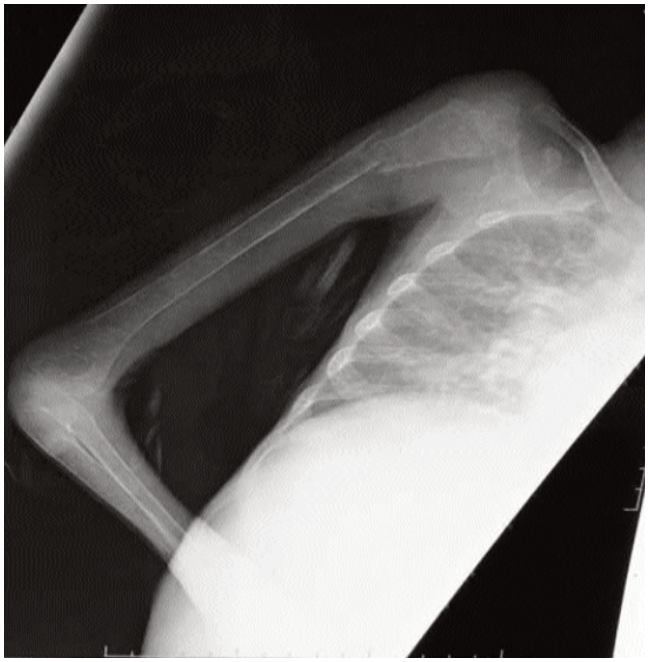

Figure-2: X-ray right arm shows displaced fracture of shaft of humerus with severe osteopenia.

examination revealed bilateral reduced air entry in lower zone of the lungs. Cardiovascular examination and Central Nervous System examination was unremarkable.

Baseline laboratory investigations showed severe anaemia with haemoglobin of 5.1 gm/dl, White Blood Cell Count: 20.3 x 10^9/L, Platelet Count: 92 x 10^9/L. Corrected Reticulocyte count was elevated at 23.5%. Prothrombin Time 14 seconds(s), Activated Partial Thromboplastin Time 33 seconds, with a control of 11s and 23 s respectively. International Normalized Ratio (INR) was 1.3. Serum Urea, Creatinine and Electrolytes were normal. However both serum Calcium and Albumin were low at 6.6mg/dl and 2.26 gm/dl respectively.

Liver Function Tests showed a total Bilirubin of 11.27mg/dl, with Direct Bilirubin as 9.76 mg/dl and Indirect Bilirubin as 1.51mg/dl with normal liver enzymes. Serum Lactase Dehydrogenase was raised to 574 U/L. Pan cultures including blood and urine cultures along with Malarial Parasite smear and Dengue serology were sent which were all negative.

Ultrasound revealed a compromised scan due to massive ascites; part of liver visualized was enlarged. Echocardiography was normal.

The patient underwent diagnostic and therapeutic Ascitic tap; 1300ml of fluid was drained undercover of Fresh Frozen Plasma to provide immediate relief in the ER. Transfusion with packed red cells (PRBCs) was started and he was shifted to High Dependency Unit (HDU) for close monitoring. He underwent repeat ascitic tap and a further 500 cc of fluid was drained. A repeat Ultrasound scan showed a liver span of 18cm with minimal ascites. He received three units of PRBCs, intravenous broad spectrum antibiotics and was adequately hydrated over the next four days.

Once the patient was vitally stable, X-rays of limbs was done which revealed right humerus fracture and bilateral old healed femur fractures. Orthopedic consult was taken; they advised an arm sling and analgesics with instructions for follow up visits for fracture repair. Paediatric team was taken on board for his malnourished state. Anthropometric measurements showed Body Mass Index (BMI) of 16.67 kg/m^2 and Weight for Height z-score <-3SD. He was given a rigorous diet plan with a combination of macro and micronutrients for the next 6 months.

Our efforts to further review and manage the patient were truncated by the patients request for discharge against medical advice because of serious financial constraint.

On discharge the child's Haemoglobin was 8gm/dl, TLC 10x 10^9/L, platelets 70 x 10^9/L. He was prescribed calcium and folate suppliments with a high protein diet and further two units of PRBCs transfusions over the next 7 days. The family was advised for strict follow-up to Haematology, Paedriatric and Orthopaedic OPDs in the coming months.

Discussion

This case report highlights the complications that arise in sickle cell disease patients with poor awareness and limited financial and social support experienced in developing countries.

Our patient belonged to a family with strong history of consanguineous marriages and was a Baloch who have the highest rate of SCD among different ethnic groups in Pakistan.[3]

This patient presented with massive ascites and hepatomegaly, severe malnutrition resulting in growth retardation, short stature, muscle wasting, fixed flexion deformity and radiologically confirmed fracture of left humerus.

The trigger for exacerbation of chronic liver failure remained elusive as blood and urine cultures were negative. In this case, the possibility of chronic, fluctuating sequestration without cholestasis reported as the most frequent hepatic manifestation in SCD

cannot be ruled out.

Liver biopsy was not performed due to deranged coagulation profile.

Our patient was extremely emaciated. Studies have shown that SCD causes slowed growth, delayed sexual maturity, and poor immunologic function partly due to under-nutrition associated with the disease.[4] The BMI of our patient was significantly lower than the reported 20.0 kg/m^2 mean BMI in male adults of SCD. The current approach is to provide additional calories in a low-bulk but high-calorie format that is appealing to children including micro and macro nutrients.

This subject showed multiple fractures both old and new on radiological examination. Avascular necrosis of long bones, persistence of red marrow, intra-medullary bone hyperplasia and bone infarcts are some of the common musculoskeletal abnormalities seen in SCD.MRI is indicated for detailed assessment of musculoskeletal abnormalities but was not done due to cost issues.

Currently Haemopoietic Stem Cell Transplant and Hydroxyurea treatment have improved life expectancy from 20 years to more than 40 years.[5] Hydroxyurea therapy has a proven efficacy when started before the onset of end-organ damage with close laboratory monitoring and dose adherence. Our patient was an unlikely candidate for this therapy due to poor medical compliance in past, presence of end organ damage and a low platelet counts.

Conclusion

SCD with genotype SS has a debilitating outcome if timely management is not done. Through this case report we have highlighted some of the complications seen in a poorly managed case of advanced SCD. A weak health care infrastructure and low level of literacy contribute to the plight of these children. A multidisciplinary approach is needed in these patients to minimize future disabilities.

Consent: Informed consent was taken from the parents for reporting this case.

Disclaimer: None to declare.

Conflict of Interest: None to declare.

Funding Disclosure: None to declare.

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