

Sickle Cell Disorder Persistence in Poor Resource Regions: Failure of a Screening Tool

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Abstract: Sickle cell is a well-recognized and preventable genetic disorder. Although, extinct in some developed climes and persist in third world countries where its orthopedic manifestations poses a diagnostic dilemma. Its persistence may connote failure in our medical screening or societal misplaced belief regarding conjugal courtship. We extracted data from patient medical journal with clinical examination in the clinic and review of relevant literatures. The case was that of a twenty months old child who had orthopedic manifestation of sickle cell disorder but was misdiagnosed probably due to wrong genotype results from both parents at presentation. Sickle cell disorder should be considered in cases of atypical multifocal chronic osteomyelitis in Nigeria. Medical screening for sickle cell should be done with confirmation to avoid error.

Keywords: Sickle cell disorder, orthopedic manifestation, diagnostic dilemma, atypical chronic osteomyelitis, failure of medical screening.

INTRODUCTION

Sickle cell disease (SCD) is an inherited genetic disorder of haemoglobin. It has a worldwide distribution due to population shift or migration with an incidence of 1 / 500 live birth in African Americans. The sickle cell trait persist in about 20 – 40% of the population, this varies in different climes with 8% of African Americans having the trait [1].

It was first reported in 1910 by two separate reports by Herrick and Washburn. Hahn and Gillespie in 1927 showed that sickling is induced by deoxygenation and reverted by oxygenation. A musculoskeletal manifestation was described by Danford who reported osteonecrosis of the femur in 1940. Pauling working with his colleagues in 1949 demonstrated an abnormal electrophoretic pattern in sickle cell haemoglobin, this has formed the basis for diagnosis and screening. In 1957, Ingram elucidated the biochemical defect in sickle cell disease [2, 3].

SCD patients are susceptible to infections including chronic osteomyelitis which usually presents without classical features (atypical). This sometimes poses a diagnostic dilemma and requires a high index of suspicion. Musculoskeletal manifestations are a major cause of disability and morbidity in West and Central Africa with the West Indies. The morbidity of chronic osteomyelitis combined with other effects of SCD decreases the quality of life [4].

Diagnosis of SCD is made by haemoglobin electrophoresis and skeletal survey to confirm

multifocal chronic osteomyelitis. In some third world countries most of the equipment used for this electrophoresis is archaic and often fabricated locally with no quality control. I have search the literature for documented evidence of failure rate of this equipments but found none. In our index case we report a case of failure of this screening tool leading to persistence of SCD in our environment.

CASE REPORT

Our patient is twenty months old child who presented initially to a private facility with a history of fever, pain in both upper and lower limbs with no discharging sinuses and passage of dark coloured urine all of 4 weeks duration.

Patient was referred to University of Calabar Teaching Hospital where she was seen in the Children emergency room (CHER) and examined following which, a diagnosis of malaria to rule out upper respiratory tract infection was made. She commenced antimalarial, antibiotics (intravenous ceftriaxone and gentamicin) and analgesics. Blood was taken for full blood

count and genotype which is usually done routinely in CHER.

Above medications relieved symptoms mildly but lower limb pains continued despite medications. This necessitated further investigations and referral to orthopaedic unit.

On presentation at the orthopaedic unit, child was irritable with difficulty bearing weight on both lower limbs and worse on the left lower limb. Radiographs of both lower limb was requested which revealed features in keeping with multifocal atypical chronic osteomyelitis of the left femur, left tibia and right femur. See Fig. 1 below.



Fig-1: Lower limb radiograph depicting chronic multifocal osteomyelitis

Following the above, enquiry into parents' genotype was done which revealed (separate results from different laboratories)

Father – HBAS
Mother – HBAA

Child's genotype result revealed HBSS. Repeat test of parents' genotype revealed both parents had genotype HBAS. She was commenced on syrup clindamycin and limb immobilized bilaterally with hip spica. Patient referred to the hematology Department for counseling and further management.

DISCUSSION

SCD may be difficult to eradicate among blacks especially from Africa due to errors in laboratory test, poor quality control regarding equipment used despite advancement in technology, poorly trained laboratory technicians and family pressures with respect to marriage. The age and symptoms at presentation of the index case is in keeping with findings reported by other authors that SCD patients with osteomyelitis present at an earlier age and the symptoms are similar to non SCD patients [4-6]. Haematogenous spread was the

commonest route of infection which is in keeping with observations studies reported by other authors [4, 5]. The child has been in and out of a health facility before being referred to our facility which is a tertiary hospital maybe due to ambiguity or dilemma of making a diagnosis probably due to wrong results presented by the parents.

It is also important to note (especially in poor resource areas) where results of investigations cannot be trusted when emanating from some private laboratories, it is pertinent for patients to carry out investigations especially genotype in two or more centers to rule out laboratory errors. This is however expensive to the patient especially if patient does not have health insurance cover. In advanced and well developed countries where laboratory errors are minimal, this may not be a problem for the managing physician.

During marital counseling usually by the clergy, intending couples are asked to bring basic screening results for HIV, Genotype, Blood group and Hepatitis B virus. Where they obtaining it from are usually not specified. In this case probably one of the parties went to a friendly laboratory, where oblivious to them, they manipulated the test results in other to save her wedding and maintain cordial relationship with their client. In this situation professionalism is sacrificed for customer relationship. Unlike other inherited genetic disorders like haemophilia, SCD can be eradicated because it has a readily available screening tool and there is increased awareness to its existence [7-9].

We use this case to demonstrate how citizen connivance / selfish interest amongst other reasons can cause SCD to persist despite increased awareness and educational level.

CONCLUSION

SCD is a cause of multifocal atypical chronic osteomyelitis. The researcher is of the opinion that, the eradication of SCD is unthinkable without strong regulatory institutions, inter-racial marriage and political will of Government. Conscientious effort should be made to identify the reason for the persistence. Treatment is multidisciplinary involving the genetist.

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