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Original Article

Evaluation of musculoskeletal manifestations of hemoglobinopathies by conventional imaging

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ABSTRACT

Introduction: The hemoglobinopathies are a group of genetic disorders in which there is alteration of hemoglobin structure. These hematologic disorders of the skeleton produce radiologic appearances related to marrow infiltration and/or replacement. Common findings are in place of such as osteopenia with trabecular prominence, growth disturbances, and extramedullary hematopoiesis. Conventional Radiography is required for knowing the imaging appearances of sickle cell disease, thalassemia, and other rare causes of anemia.

Objectives: To study the role of conventional imaging in detecting various radiological manifestations of hemaglobinopathies.

Materials & Methods: An observational descriptive study of 21 cases with hemaglobinopathies was done by conventional radiography & its varied musculoskeletal manifestations were studied.

Result: Most common site of involvement was head of femur which was seen in 16 cases out of which isolated involvement was seen in 10 cases. Rest of the cases was associated with spine, skull and upper limb manifestations.

Conclusion: Knowledge of the distribution and site of involvement and its musculoskeletal features is of considerable diagnostic importance in the evaluation and treatment of hemoglobinopathies. Conventional Radiography is required and its primary investigation modality It is important to know the extent of the range of radiologic appearances in order to accurately diagnose complications and to provide appropriate treatment.

Keywords: Hemoglobinopathies, Conventional Radiography,

INTRODUCTION

The hemoglobinopathies are a group of genetic disorders with a broad spectrum of clinical manifestations and findings. These hemoglobinopathies are associated with characteristic abnormalities of the skeleton. These include common findings, such as osteopenia with trabecular prominence, growth disturbances, and extramedullary hematopoiesis. The articular manifestations in the hemoglobinopathies can be attributed to epiphyseal osteonecrosis, growth disturbances, osseous weakening, infection, crystal deposition,

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hemarthrosis, and synovial membrane microvascular obstruction[1,2]

In India, the cumulative gene frequency of haemoglobinopathies is 4.2%. With a population of over one billion, there are over 42 million carriers and over12, 000 infants are born each year with a major and clinical significant haemoglobinopathy.[3]

Although these features are apparent in almost all the hemoglobin disorders but their severity varies, which in some cases allows differentiation among these disorders and aid their clinical management.

Classification of Hemoglobinopathies[4,5]

- Structural hemoglobinopathies such as sickle cell anemia.
- Thalassemia
- Thalassemic hemoglobin variants
- Hereditary persistence of fetal hemoglobin
- Acquired hemoglobinopathies

Aims & Objectives

To study the role of conventional imaging in detecting various musculoskeletal manifestations of hemaglobinopathies.

METHODOLOGY

Study area

The study was carried out in the Department of Radiodiagnosis, S.B.K.S. Medical Institute and Research Centre, Waghodia, Vadodara.

Study design

Type of the study: An Observational, Descriptive Hospital Based Study.

Sample size: 21 patients were taken up for study who were referred from clinical department and OPD of Dhiraj Hospital during the study period of 3 months.

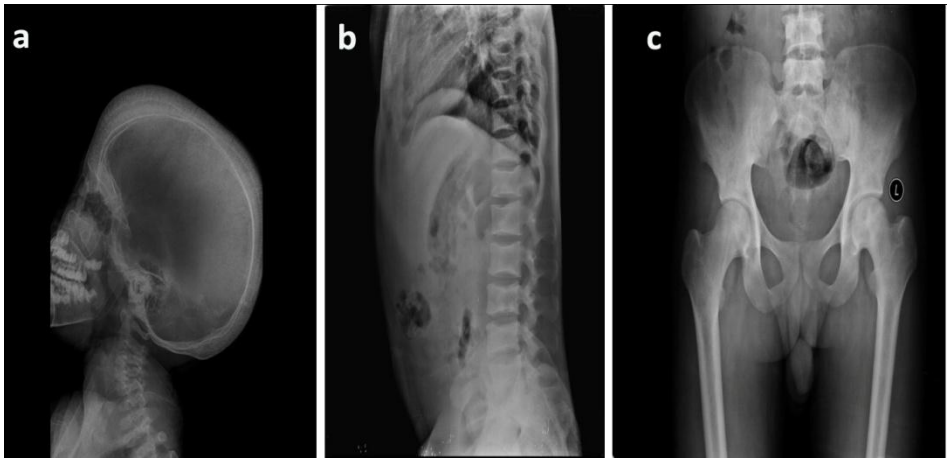

Figure 1. (a) Marked widening of vault of skull suggestive of classical hair on end appearance[17,18] (b) H- shaped vertebral body at multiple levels showing depression in mid part of superior & inferior endplates of body (c) Avascular necrosis of head of left femur in sickle cell disease.[19]

Selection of subject

Inclusion criteria

Patients referred to the radiology department for clinically suspicion of having hemoglobinpathies and found to have positive findings were included in this study.

Known cases of various hemoglobinopathies came for follow up.

21 Patients were evaluated. Where the Patients had presented with anemia with some associated symptoms like bone and joint pain, abdominal pain, icterus, fever, weakness etc. X-rays of the affected part along with total skeletal survey was conducted.

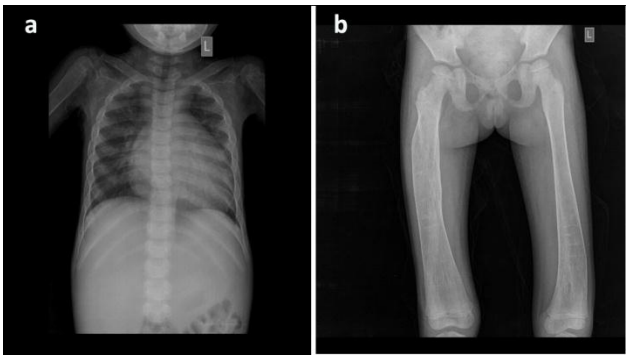

Figure 2. a,b Expansion of ribs showing prominent trabecular pattern with small left paravertebral shadow in lower dorsal region,s/o extramedullary hematopoiesis20 and widening of diaphysis with prominent trabecular pattern in both femur in k/c/o thalassemia[21]

RESULT

Most common age group in this study was 11-20 yrs (42.8%) followed by 1-10yrs and 21-30yrs having an incidence rate of 33.3% and 19.2% respectively. In this study hemoglobinopahties were more common in males (61.9%) than in females (38.1%). Sickle cell anemia was more common than thalassemia with 15 cases of sickle cell anemia (2-sickle cell trait) and 6 of thalassemia.

Most common site of involvement was head of femur which was seen in 16 cases out of which isolated involvement was seen in nine cases[4]. Rest of the cases were associated with spine, skull and upper limb manifestations.

Spine involvement was seen in seven cases and were all associated with other site manifestations. Skull was involved in two and ribs were involved in one case.

Sickle cell anemia was associated with lower limb changes in nine cases, spine in five cases, upper limb in two cases and skull in one case. Thalassemia was associated with lower limb changes in five cases, upper limb in 2, spine in 2 cases, skull and rib in 1 case each.

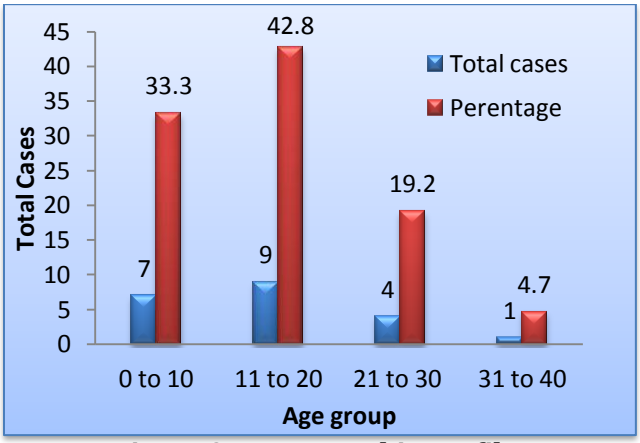

Figure 3. Demographic Profile

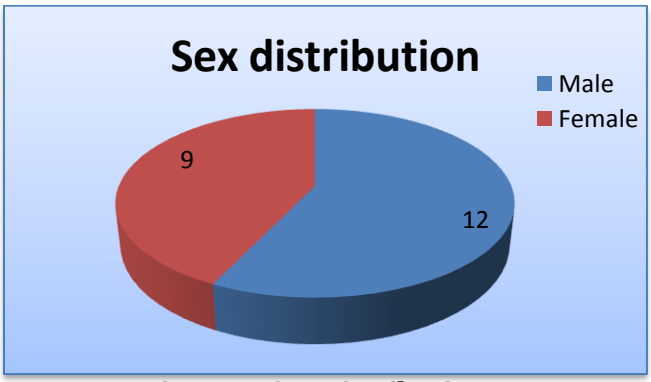

Figure 4. Sex Distribution

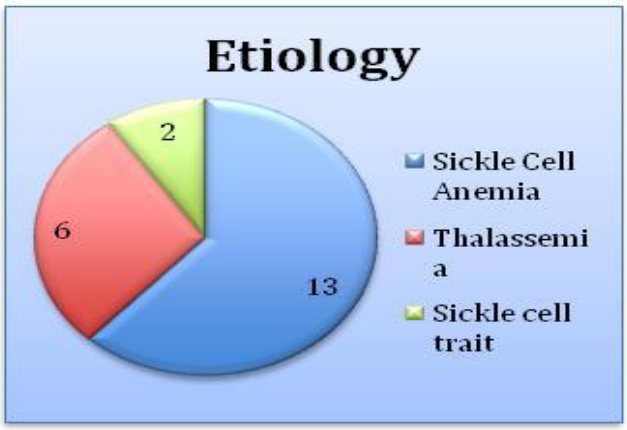

Figure 5. Classification according to etiology

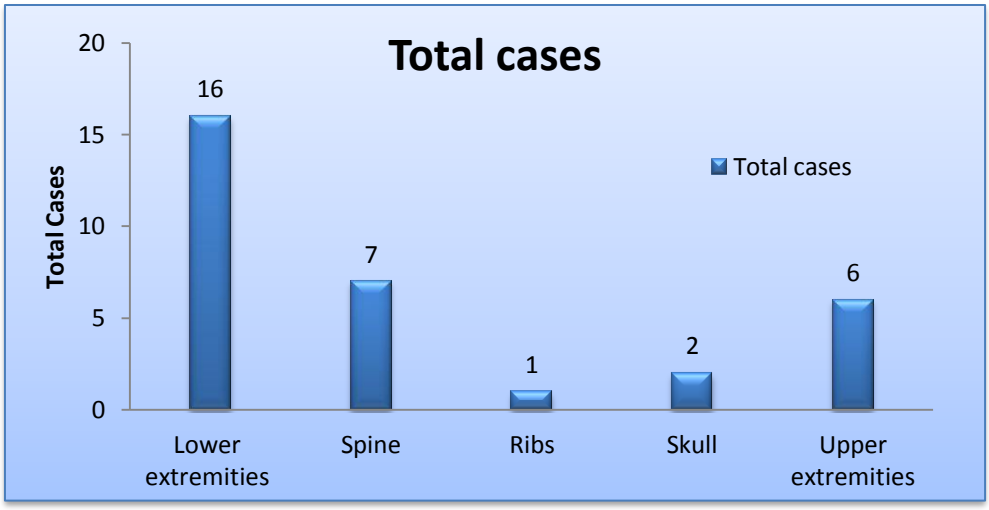

Figure 6. Classification according to site involved

DISCUSSION

Avascular necrosis of the bone is a relatively common problem among patients with Sickle cell disease.[6,7] Both in children and in adults, the long bones are commonly affected and the femoral head is the most common area of bone destruction in these patients. These findings were consistent with findings of previous studies.[8,9] Milner et al (1991) in a study of 2590 cases found that osteonecrosis of the femoral head is common in patients with sickle cell disease, with an incidence ranging from about 2 to 4.5 cases per 100 patient-years with overall prevelance of 10%.[10,11,12]

In a similar study done by M Mukisi-Mukaza et al (2000) found that out of 113 adults with sickle cell disease Forty-two (37.2%) patients had osteonecrosis of one or both hips.[13,14]

As the patients are chronically anaemic there is persistence of red marrow in both the axial and appendicular skeleton into adulthood together with bone marrow hyperplasia which results in widening of the medulla and subsequent cortical thinning, resulting in coarsening of the normal trabecular pattern in long and flat bones.[15] Common examples of abnormal intramedullary haematopoiesis include .include skull and vertebral bodies.[16]

CONCLUSION

Knowledge of the distribution and its musculoskeletal features is of considerable diagnostic importance in the evaluation and treatment of hemoglobinopathies. It is important to be aware of the range of radiologic appearances in order to accurately diagnose complications and expedite appropriate treatment.

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