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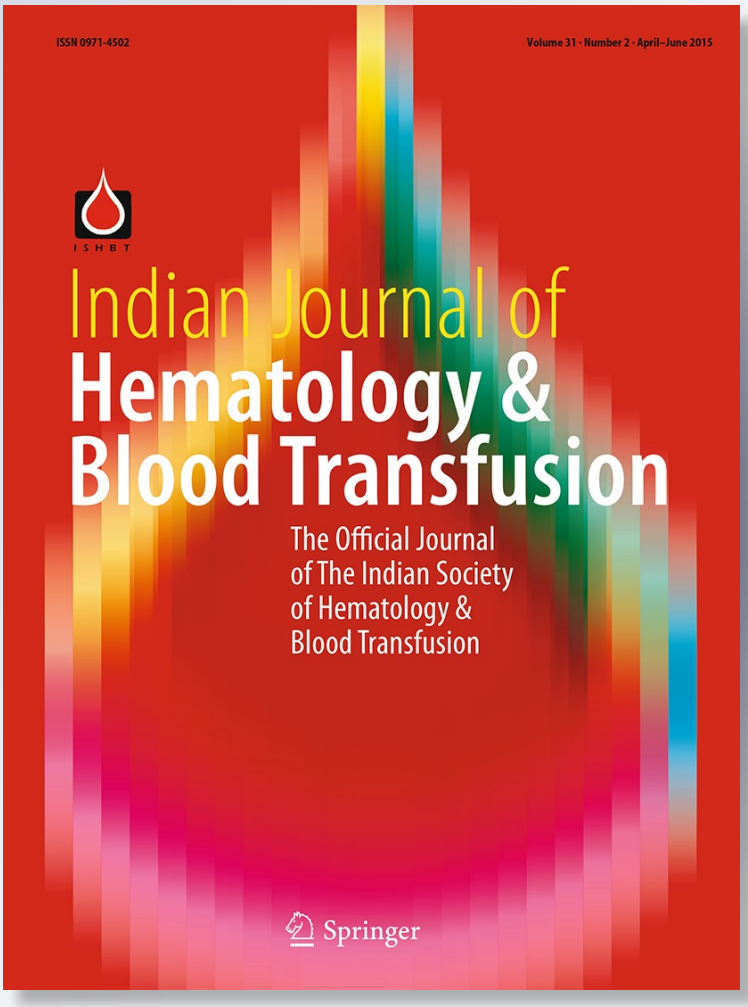
Indian Journal of Hematology and Blood Transfusion

ISSN 0971-4502
Volume 31
Number 2

Indian J Hematol Blood Transfus (2015)
31:295-296
DOI 10.1007/s12288-014-0422-0



Springer

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CASE REPORT

An Adolescent Case of Osteopetrosis with Portal Hypertension as well as Mandibula Osteomyelitis

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Received: 18 December 2013 / Accepted: 10 June 2014 / Published online: 25 September 2014

Abstract Osteopetrosis is a clinical syndrome characterized by the failure of osteoclasts to resorb bone. Excessive bone density can interfere with vital tissues and structures, causing serious problems of the body. Hematopoietic insufficiency, disturbed tooth eruption, nerve entrapment syndromes, and growth impairment may develop in a patient with osteopetrosis. Herein, we present an adolescent girl diagnosed with non-infantile type of osteopetrosis with rare complications of the disease like mandibular osteomyelitis and portal hypertension (PHT) without liver cirrhosis. To our knowledge, this is the first pediatric case with osteopetrosis related PHT.

Introduction

Osteopetrosis is a hereditary disorder characterized by generalized osteosclerosis as a result of decreased osteoclastic activity and unequal balance between new bone growth and elimination of old bone [1]. Increased intraosseous bone growth results in reduced marrow space with hematologic complications as well as external bone growth with narrowed foramina as resulting in cranial nerve palsies and visual complications beside other bone shape changes. We present an adolescent girl diagnosed with non-infantile type osteopetrosis with rare complications of the disease like mandibular osteomyelitis and PHT without liver cirrhosis.

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Case Summary

A 15-year-old girl with osteopetrosis was admitted to our pediatric emergency department with the complaints of cough, sore throat and abdominal pain. Her past medical history included multiple erythrocyte and thrombocyte transfusions because of chronic anemia and thrombocytopenia, multiple bone fractures (including pelvic bones) and recurrent hospitalizations for surgical and medical treatment of mandibular osteomyelitis and periorbital abscess.

On admission, the patient was alert, awake and cooperative but she had some degree of deafness and blindness. Her vital signs were within normal limits, except tachycardia, exophthalmic right eye, and mandibular purulant drainage. Spleen was palpated about 10 cm and liver about 3 cm below the costal margin. Some deformities related with old bone fractures were also remarkable at the extremities. She was hospitalized with diagnosis of osteopetrosis, mandibular osteomyelitis and anemia. Laboratory investigation revelaed the following: Hb: 7, 9 g/dl, Plt: 1,35,000/mm^3, WBC: 4,800/mm^3, PT: 14 s, fibrinogen: 224 mg/dl, total protein-albumin: 6, 3-2, 5 g/dl, Erythrocyte transfusion and apropriate antibiotic treatment was started. Generalized ostesclerosis was seen in the chest radiograph (Fig. 1). PHT and ascites were detected in the abdominal doppler ultrasonography. Thrombus or obstruction was not detected in the portal vein and liver paranchyme was homogeneous. Spironolactone was added to the treatment and antibiotics were continued. During the treatment, she suddenly died of pulmonary hemorrhage.

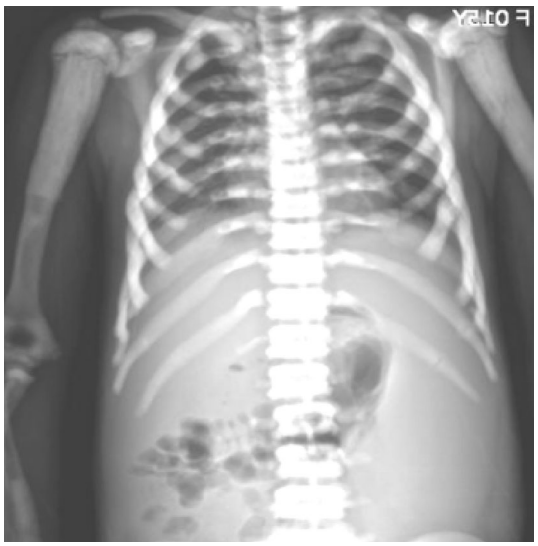

Fig. 1 Chest radiograph showing generalized osteosclerosis and shadow of massive spleen

Discussion

Portal venous pressure is normally aproximately 7–10 mmHg and levels above 10–12 mmHg is defined as PHT [1]. There are numerous causes of PHT; most of them are related to the obstruction of the portal circulation. Non-obstructive PHT is rare. These causes include congestive heart failure, arteriovenous fistulas and splenomegaly. Hepatosplenomegaly may be present with osteopetrosis due to extramedullary hematopoiesis in the reticular system. Increased inflow to the portal venous system, usually because of splenomegaly, causes the changes that lead to clinical findings of portal hypertension, also called "Forward Flow Portal Hypertension" [2].

Therapeutic approach to the infantile type of osteopetrosis mainly stays on bone marrow transplantation. Our patient did not receive BMT since a donor was not available. Also, she did not receive enough transfusions for anemia although she was remarkably anemic since the family was not compliant to treatment and medical follow-up. These factors may have contributed to her splenomegaly and ascites formation with forward flow PHT.

In a previous case report in 1971, a 56-year-old woman with osteopetrosis and PHT was presented [3]. This is the only report of this association we can find in the English literature. The presented patient in this report has almost the same clinical findings as in our patient like exophthalmos, visual loss, multiple old fractures, a draining sinus and splenomegaly, except ascites. Ascites formation and presentation at an adolescent age is unique to our patient.

Osteomyelitis related to osteopetrosis is not common although it is a well known entity. It is seen mostly as a result of odontogenic infections and generally is polymicrobial [4]. It is a resistant infection because of the decreased blood flow to these locations and neutropenia. As a result of this fact, oral hygiene is very important in these patients to prevent dental complications. All patients must also be followed by a dentist even when they have no complaints. Our patient was treated with appropriate broad spectrum antibiotics. As the literature revealed, most of the patients are unresponsive to treatment. Our patient died of pulmonary hemorrhage of unknown origin while she was getting treatment. Pulmonary hypertension may develop prior to or a result of bone marrow transplantation in these patients [6]. Bleeding may have developed due to pulmonary hypertension as a complication.

In conclusion, splenomegaly related to forward flow PHT is a rare entity in osteopetrosis. Awareness of this condition is important to take necessary precautions to prevent massive splenomegaly and ascites.

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