

Lymphangiomatosis with Extensive Skeletal Involvement

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Received: August 5, 2015

Revised: August 30, 2015

Accepted: September 3, 2015

Published online: October 23, 2015

ABSTRACT

Aim: To review a case presentation of systemic lymphangiomatosis—an uncommon medical condition that can be easily misdiagnosed in the clinical setting. **Abstract:** Generalized lymphangiomatosis is a rare condition with characteristic multi-centric angiomatous malformations of the skeletal system, with involvement of surrounding soft tissues and skin. It results from a congenital anomaly and presents predominantly in the paediatric age group. Systemic involvement includes not only the skeletal system, but also the pulmonary and gastrointestinal systems. Its diagnosis can be difficult to ascertain but is hinged on clinical presentation, imaging and histological confirmation. The condition carries a variable prognosis as treatment options are very limited. Pulmonary involvement is usually terminal, however. A case is presented to highlight the difficulties in diagnosing and managing this condition. **Conclusion:** The diagnosis of lymphangiomatosis involving the skeletal system is highly reliant on imaging studies; that depict well defined osteolytic changes of the bone. Management is supportive and no cure currently exists.

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Key words: Lymphangiomatosis; Systemic; Skeletal involvement

White K, Calderon C. Lymphangiomatosis with Extensive Skeletal Involvement. *International Journal of Orthopaedics* 2015; 2(5): 435-437
Available from: URL: <http://www.ghrnet.org/index.php/ijo/article/view/1341>

CASE REPORT

An 11 year old male of afro-caribbean descent presented with a two week history of a painful right hip with a limp, following a fall. Clinical history and examination revealed no constitutional symptoms and no deformities were noted.

Plain radiographs of the hip depicted generalised osteopenic changes, with small osteolytic lesions of both femurs and pelvis (Figure 1).

Further imaging - including contrast tomography displayed extensive, multiple, geographically well defined lucent lesions throughout the skeleton (Figures 2-4). Magnetic Resonance Imaging demonstrated a diffuse osteolytic process with medullary fat replacement and bone remodelling, suggestive of Langerhan' cell histiocytosis (Figure 5). With a differential diagnosis of leukaemia still to be excluded. Multiple bone biopsies from pelvis and femur proved inconclusive and insufficient.

The patient absconded from care early the following year, to re-present approximately 2 years later with a history of a serous discharge from the superior surface of the right buttock. A soft tissue mass was noted in the gluteal region with extension to posterior aspect of the thigh (Figure 6). Small multiple weeping bullous lesions were noted on the overlying skin of the gluteal mass. Contrast tomography at this time, depicted a right superficial gluteal lymphangioma with associated bony involvement and engorgement of retroperitoneal and posterior mediastinal lymphatic chain. Histology findings from punch biopsy of right gluteal mass were consistent with that of lymphangiomatosis (Figure7).

He subsequently developed a left sided chylothorax, with an associated right sided pleural effusion later on. Immediate management entailed thoracostomy and shortly after he underwent a two port video- assisted thoracoscopic pleurodesis.

The patient was commenced on interferon alpha treatment and conservative management of other medical complications, including electrolyte derangements, ensued. He subsequently succumbed to his medical condition at the age of thirteen.

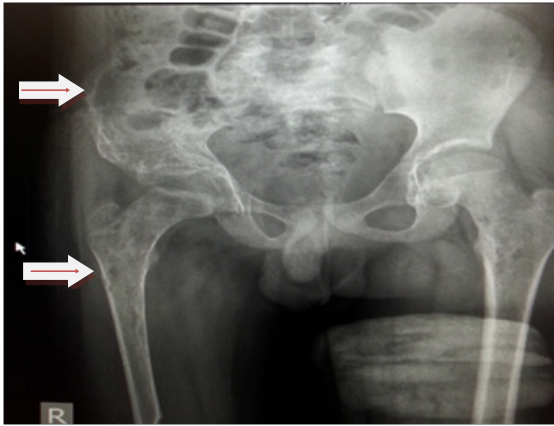


Figure 1 Plain radiograph of pelvis showing osteolytic lesions of the proximal femur and pelvis.

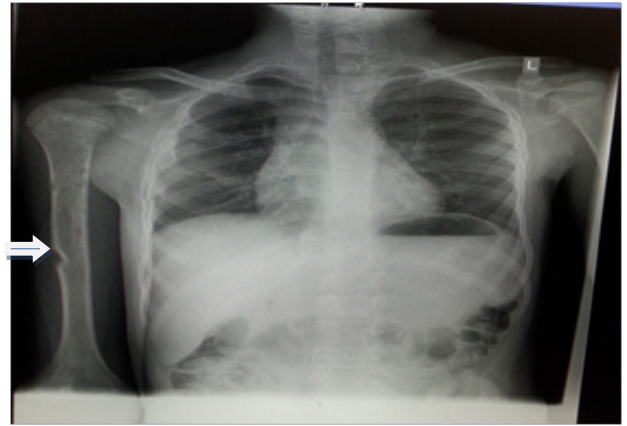


Figure 2 Plain radiograph of chest showing multiple osteolytic lesions of right humerus and ribs.

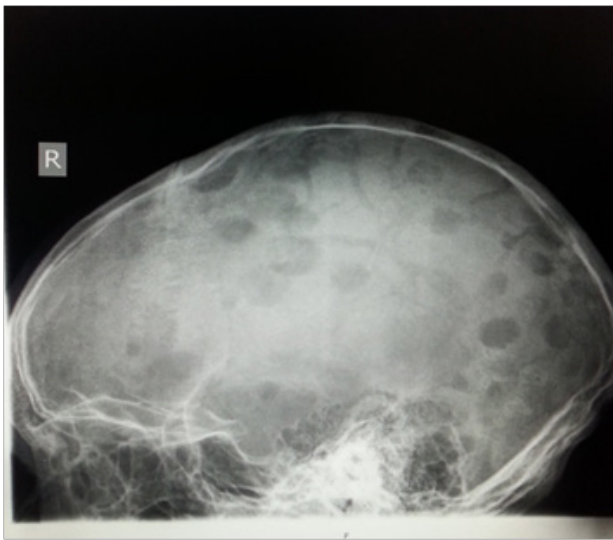


Figure 3 Plain radiograph of skull - "soap bubble appearance"



Figure 4 Plain Radiograph displaying osteolytic lesions of distal femurs bilaterally.



Figure 5 MRI of right hip showing large soft tissue mass (blue arrow) and osteolysis of ipsilateral femur shaft (red arrow)



Figure 6 Soft tissue mass overlying right gluteal/hip region.

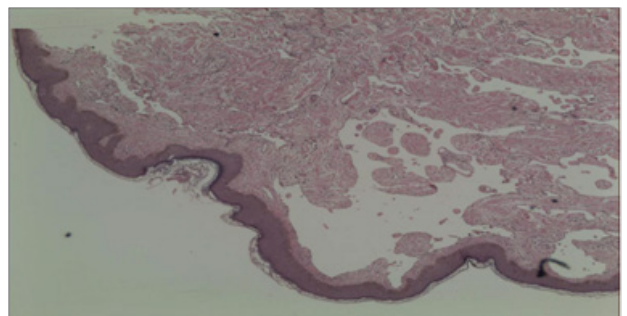


Figure 7 Intra-lesional biopsy over gluteal region - histology showing increase in size and number of thin-walled lymphatic channels.

DISCUSSION

Generalised lymphangiomas are a rare multi-system affecting medical condition arising from the lymphatic system^[1]. This progressive disorder, results from a proliferation of lymphatic channels and an increase number of complex anastomoses forming between them, during intrauterine life^[1,2]. The end product is a continuous dilation of these interconnected thin-walled lymphatic vessels, containing benign endothelial cells^[3]. The age group affected is highly varied, although most are associated with the paediatric age group, with no predilection to any gender, and no known inheritance pattern^[4].

Lymphangiomas may be categorised macroscopically as either localised or systemic. The systemic form which was noted in this case above differs from the localised forms of the disease – which are isolated to a single anatomic location, classically bone, and have a good prognosis. Although seventy-five percent (75%) of cases in the systemic form include skeletal manifestations, the occurrence of multifocal lymphangiomas involving the gastrointestinal tract, lung, and pleura creates additional management challenges. As was highlighted in the case, thoracic involvement accelerated the clinical course and may have hastened his demise.

Symptoms, clinical presentation and progression of disease contrast broadly, adding to the difficulty in making a definitive diagnosis.

Most cases of skeletal involvement are found incidentally on plain radiographic imaging, or as a pathological fracture, a consequence of an internally weakened bone structure^[5]. These appear on imaging as central areas of lucency, with or without reactive borders, giving a multi-loculated “soap bubble” appearance^[1,6]. The exterior cortex of the bone is typically maintained, however, reports of vertebral collapse have been made. As noted from the case report, although extensive bone involvement was noted initially, symptoms were relatively mild, and linked only to the unilateral weight bearing limb.

A diagnosis of this condition requires a high index of suspicion, as it may be easily misdiagnosed, even with appropriate imaging, and biopsy findings. Radiological investigations of importance include high resolution computed tomography and more so, magnetic resonance imaging^[2]. These are pivotal in determining the extent of disease and hence excluding critical organ involvement. Biopsy findings provide the confirmatory diagnosis, illustrating a lymph filled multi-locular cavity. Use of magnetic resonance lymphangiography may prove valuable, where available^[7,8].

Differential diagnoses include metastatic disease, histiocytosis X and Gaucher’s Disease. Lymphangiomas are closely linked to Gorham massive osteolysis, also known as phantom bone disease, the difference lying in the incongruous pattern of lesions noted in systemic lymphangiomas^[1].

Surgery is unlikely to be curative in the systemic form of lymphangiomas owing to its infiltrative properties and the high likelihood of recurrence post operation, quoted as 60- 90% in some series^[9]. Hence, management usually takes a more supportive, and at times, a palliative role. Radiation therapy to skeletal lesions has been noted to provide only temporary remission. Medical therapies may be utilised, such as interferon alpha 2b and bisphosphonate therapy^[1]. Many other experimental treatments are being explored in hope of

achieving a positive result^[10,11].

It is arduous to state a clear prognostic factor or an expected survival gauge in cases of systemic lymphangiomas. However, what is known, is that this is highly dependent on the extent of the condition; and when there is thoracic involvement, as with this patient case, the prognosis worsens significantly^[4,9].

CONCLUSION

Systemic lymphangiomas are a progressive congenital maldevelopment of the lymphatic vessels affecting multiple organ systems, most commonly the skeletal. Diagnosis is highly reliant on imaging studies that depict well defined osteolytic changes of the bone. Management has a supportive role and there is no cure currently for this condition.

ACKNOWLEDGMENTS

Special thanks to Dr. Rao of the pathology department at the Eric Williams Medical Sciences Complex.

CONFLICT OF INTERESTS

There are no conflicts of interest with regard to the present study.

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