Gorham’s Disease

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Abstract

Gorham’s disease is a rare disorder characterised by proliferation of vascular channels that result in destruction and resorption of osseous matrix. A case of Gorham’s disease presenting as chylothorax and osteolysis of ribs is presented.

Case Report

A 9 year old female child presented to our department with complaints of breathlessness on exertion, loss of weight and loss of appetite of four months duration. No history of cough or fever. History of occasional mild right sided chest pain present. Her parents noticed a slight deformity of right side of chest 3 years back and she was shown to a local general practitioner and advised vitamins in the absence of any major disease. She was born as full term normal delivery to non consanguineous parents and fully immunised to the age. On examination she was 24 kg, anaemic, afebrile, pulse rate 90/mt, BP 102/70mm of Hg, drooping of right shoulder present, scoliosis with convexity to left, flattening of right chest and ribs on the right side was not palpable. Dull percussion notes were present on both lower chest. Breath sounds were diminished on both lower chest with diminished vocal resonance on those areas. Her blood examination showed Hb 11.5 g%, TC 8900/cumm, N62 L3 E2, ESR 12/hr. X-ray chest demonstrated bilateral pleural effusion. Her X-ray chest taken one year ago showed normal bony cage of chest and recent X-ray chest demonstrated absence of multiple ribs on right side, Ultra sound confirmed pleural effusion on both sides and normal liver, kidneys, spleen and pancreas. She had normal renal and liver functions, serum calcium and phosphorus were normal.

Pleural aspiration was done and aspirated 300 ml of milky fluid on the right side and 200 ml on the left side. Pleural fluid was chylous (cholesterol 140 mg%, triglyceride 186 mg/dL) with mature lymphocytes on cytology and negative cultures. A CT Chest showed bilateral pleural effusion and marked osteolysis of ribs on the right side. Patient expired on the seventh day of admission following acute onset of breathlessness and respiratory failure. A true cut lung biopsy done soon after her death on histopathology showed fibrocollagenous tissue without any evidence of malignancy or infection.

Discussion

Gorham’s disease is a rare
disorder characterised by proliferation of vascular channels that result in destruction and resorption of osseous matrix. Gorham’s disease is first described by Jackson in 1838 and Gorham and Scout described the main pathologic features of this disease as ‘Vanishing bone disease associated with intravascular changes’. Gorham’s disease occur at any age but it is most often recognised in children and young adults. There is no clear sex predilection or inheritance pattern, which is important because an inheritance pattern suggest a diagnosis of other familial osteolytic disorders. The process may affect the appendicular or the axial skeleton. The shoulder and the pelvis are the most common sites of involvement, however various locations such as the humerus, scapula, clavicle, ribs, sternum, pelvis and femur can be affected by Gorham’s disease. Diseases of the ribs, scapula or thoracic vertebra may lead to the development of chylothorax from direct extension of lymphangectasia in to the pleural cavity or via invasion of the thoracic duct. Without surgical intervention, patients with Gorham’s disease who develop chylothorax have a high rate of morbidity and mortality. Signs and symptoms of the disease are related to the area and extent of involvement. The most common of these include pain, cough and dyspnoea. Some patients present with a relatively abrupt onset of pain and swelling in the affected extremity, whereas others present with history of insidious onset of pain, limitation of motion and progressive weakness in the involved limb; this may be accompanied with soft tissue weakness and atrophy.

Laboratory analysis is uniformly unhelpful. Occasional reports of transient mild elevation of serum alkaline phosphatase level as well as eosinophilia have occurred, but their significance is unknown. The hallmark of massive osteolysis of Gorham is the radiographic findings. Radiographically the earliest changes are foci of intramedullary and subcortical lucency resembling osteoporosis. Concentric reduction results in tapering of involved long bones. This is followed by complete resorption of the involved bones in severe cases. Biopsy specimen of affected bone may show initial angiomatosis that is later replaced by vascular fibrous tissue. Lymphangiomatosis and haemangiomatosis have been described and suggested to represent different manifestations of the same abnormality, believed to represent a developmental endothelial defect. Histologic examination shows irregular wide spaces in cortical bone and enlarged marrow spaces with thin walled capillary like vessels with or without blood present. CT scan is useful in the delineation of the soft tissue extension and it enables biopsy guidance. Lymphadenopathy has been used to assess the thoracic duct in patients with chylothorax. The lymphatic vessels and nodes have a normal appearance although altered lymphatic flow can lead to obstruction and oedema. MR images show morphologic disappearance of bone and areas of increased or decreased signal intensity, which may represent haemorrhage at different stages. The differential diagnosis of Gorham’s disease includes skeletal angioma, angiosarcoma, essential osteolysis and hereditary osteolysis.

Due to the rarity of this disease entity, there is no standard therapy available. The medical treatment for Gorham’s disease include radiation therapy, anti osteolytic medication (biphosphonates) and alpha 2b interferon. The principal treatment modalities are surgery and radiation therapy. Surgical options include resection of the lesion and reconstruction using bone grafts and/or prosthesis. Definite radiation therapy in moderate doses appears to result in a good clinical outcome with few long term complications. Chylothorax is a rare complication and is usually associated with shoulder girdle or thoracic vertebral bony osteolysis. The chylothorax is a potentially dangerous complication and result from invasion of the thoracic duct or penetration of the lymphatic dysplasia in to the pleural cavity. Various treatment modalities have been employed for the management
of chylothorax in patients with Gorham’s disease including pleurectomy, pleurodesis, thoracic duct ligation, radiotherapy and interferon therapy. The mortality rate is high once pleural effusion is developed. Our patient died due to respiratory failure prior to any specific treatment.

References