

Spontaneous Hemothorax in Hereditary Multiple Exostoses- Report of A Rare Case and Review of Literature

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Abstract: We are reporting a case of spontaneous hemothorax in a 24 year old male patient diagnosed with Hereditary Multiple Exostoses (HME), presented with chest pain and dyspnea and CT chest showed pleural effusion on the left side. Pathological diagnosis was made on biopsy of the excised rib specimen.

Key Words: Hereditary multiple exostoses, Hemothorax, Osteochondroma, Chondrosarcoma

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I. Introduction

Hereditary multiple exostoses (HME) is rare autosomal dominant disorder affecting musculoskeletal tissue characterized by the presence of multiple exostoses or osteochondromas from the beginning of early developmental period. HME is defined by the presence of at least two exostoses or osteochondromas of the juxta epiphyseal region of long bones. Incidence of HME is approximately 1:50000 in general population¹. Even though HME has a preponderance towards long bones, it can grow on any bones in the body including ribs and can cause compression. Rib exostoses can cause injury to pleura, diaphragm or pericardium and the presentation varies from patient to patient. Common presentations ranges from asymptomatic to spontaneous pneumothorax and hemothorax. Chest imaging shows pleural effusion or pneumothorax depending on the patient's presentation. Treatment option for exostoses of rib is mainly surgical.

II. Case Report

24 year old male patient, who is a non-smoker and no past history of pulmonary disease, presented to us with chest pain and dyspnoea of 3 days duration. He has no significant occupational exposure. On examination, his vital status were stable and oxygen saturation was 97%. Upon general examination, there were bony swelling in both wrist, knee, ankle and left side of the chest (picture 1-4). When enquired about these swellings, he told that it was present for almost 15 years and consulted orthopaedics department in the past for this and they told that it is a benign growth. Even though he has multiple bony swellings, he didn't has any symptoms in the past. On pulmonary system examination, there was dullness to percussion in the left lung fields. On auscultation, breath sound was decreased in the left lung fields.

He was investigated further. Chest X-ray showed pleural effusion on the left side (figure 5). CT chest showed pleural effusion and exostoses on the left side (figure 6).

Upon the diagnosis of pleural effusion, the patient was posted for ultrasound-guided thoracocentesis^{2,3} and aspirated around 1L of haemorrhagic fluid and he was treated with antibiotics and supportive measures and cardiothoracic consultation was done and decided to surgically excise the rib exostoses. During the procedure haemothorax was present and aspirated 2.5L of haemorrhagic fluid. There were multiple exostoses observed (figure 7) during surgery and portion of ribs with exostoses were surgically excised (figure 8) and sent for pathological analysis. Pathology report came as osteochondroma. Patient was stable clinically and symptomatically following procedure.



Figure 1



Figure 2



Figure 3



Figure 4

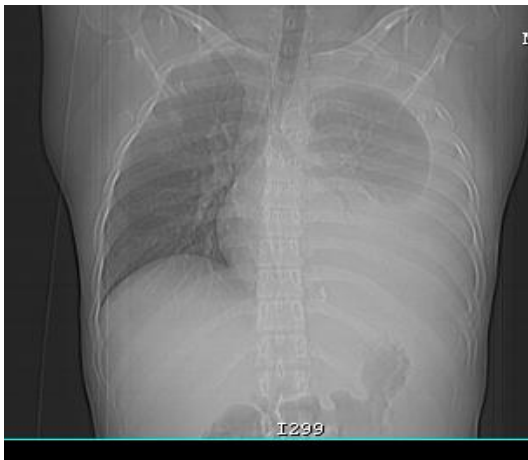


Figure 5

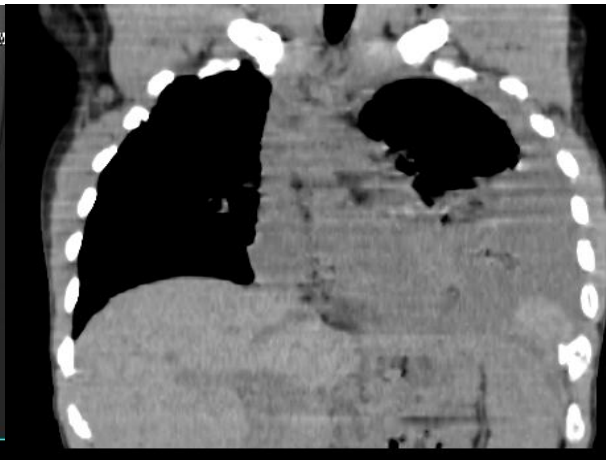


Figure 6

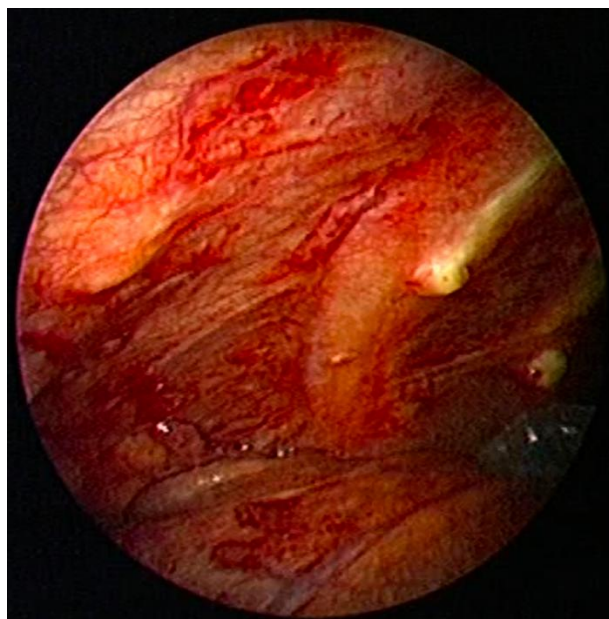


Figure 7

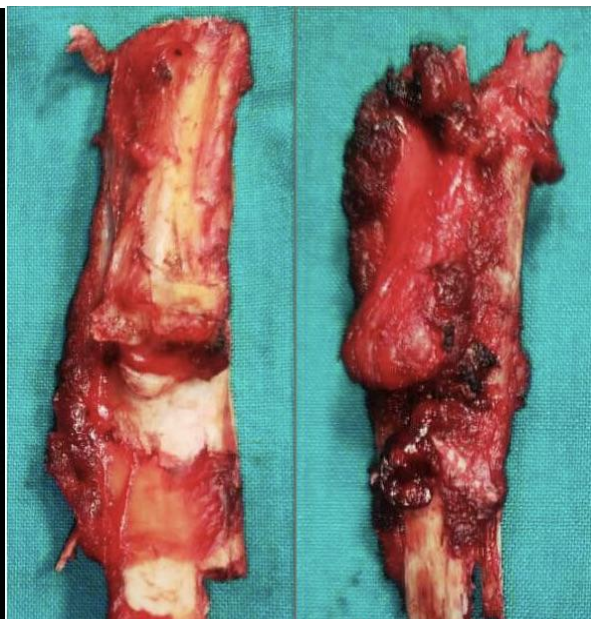


Figure 8

III. Discussion

Hereditary multiple exostoses (HME) is a rare autosomal dominant disorder affecting musculoskeletal tissue. It is characterized by exostoses of long bones usually appearing and extending in first decade of life. Usually there will be negligible or no extension after achieving puberty. The usual site of occurrence of exostoses are proximal portion of humerus and around the knee joint. Facial bones are spared. Even though long bones are affected mostly, it can occur in any bones in the body. Exostoses are mostly asymptomatic but can cause symptoms depending on the location and includes neurological compression, vascular compression, pain and fractures.

HME is associated with loss of function mutations in *EXT1* (8q24) and *EXT2* (11p11) genes⁴. *EXT1* and *EXT2* genes are tumour suppressor genes coding for exostosins 1 and 2, two glycosyl-transferases required for the biosynthesis of heparan sulfate. Individuals with *EXT1* mutation have severe presentation than with *EXT2* mutation and it include higher rate of chondrosarcoma, more exostoses, limb malalignment and more pelvic and flat bone involvement.

Ribs exostoses are described between 44% and 35% of cases, depending on genotype *EXT1* and *EXT2* respectively. Rib exostoses are usually asymptomatic, but can cause pneumothorax⁵, haemothorax⁶, injury to pericardium and diaphragm. In our case, the patient presented with haemothorax.

Diagnosis of HME requires both radiological and pathological analysis. CT and MRI scan are preferred radiological modalities. Radiological features suggesting exostoses are pedunculated or sessile appearance, thin lobulated cartilaginous cap and it may contain calcifications. Pathological analysis shows cartilaginous cap composed of mature hyaline cartilage with overlying fibrous perichondrium.

One of the important complications of HME is malignant transformation to chondrosarcoma⁷ and the incidence is 5%-10%. Proximal lesions are more likely to undergo malignant transformation than distal lesions. Cartilaginous cap of >2cm has more chance towards malignant transformation.

Treatment options of HME varies depending on the patient's presentation. If asymptomatic, observation alone is needed. Surgical excision is done in case of complications. In HME with spontaneous haemothorax, surgical excision of the affected rib can be done. Local recurrence after surgery is rare. In our patient, excision was done and he was discharged without any complications. Surprisingly there were no recurrence during the follow up of 2 years.

Literature review of some of the surgically treated intra thoracic exostoses are given below

Reference	Year	Age	No. of exostoses	Procedure	Outcome
[8]	2015	18	1	Thoracoscopy	Good
[9]	2014	15	1	Thoracotomy	Good
[10]	2014	16	2	Thoracoscopy	Good
[11]	2013	5	1	Thoracoscopy	Good
[12]	2012	25	1	Thoracotomy/Thoracoscopy	Good
[13]	2011	14	2	Thoracotomy	Good

Table 1

IV. Conclusion

In conclusion, HME can occur at any sites. Rib exostoses in HME can be presented as unusual cause of spontaneous haemothorax. Surgical excision of rib exostoses is definitive treatment with minimal chance of recurrence. Change in size of exostoses or change in symptoms can be a concern of malignant transformation to chondrosarcoma and should be investigated further.

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