

A Case of Maffucci's Syndrome With Pleural Effusion: Ten-year Follow-up

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Abstract

Introduction: Maffucci's syndrome (MS) is a congenital non-hereditary mesodermal dysplasia characterised by numerous mesenchymal neoplasias in the form of enchondromas with secondary bone deformities and multiple soft tissue haemangiomas that may have phlebolitis. **Clinical Picture:** A 23-year-old male patient presented with non-productive cough, dyspnoea, chest pain and back pain. Chest X-ray showed unilateral pleural effusion and multiple enchondromas of the ribs. On physical examination, there were mobile, multiple, bluish-coloured lesions probably cavernous haemangiomas on bilateral chest walls. In addition, there were multiple nodular lesions on the extremities especially accumulated on the fingers. The patient was diagnosed as Maffucci's syndrome according to computed tomography (CT) of the thorax, conventional radiography of the skeletal system, magnetic resonance (MR) imaging, Th₄-Th₁₁ intercostal and right upper extremity angiography and physical examination findings. **Treatment:** As the patient rejected any diagnostic intervention, he was monitored with CT. **Outcome:** During the last 6 years of follow-up, the lesion that was detected on the rib adjacent to the basal segments of the left lung showed significant progression and was accepted as chondrosarcoma. **Conclusion:** To our knowledge, this is the first case of Maffucci's syndrome with pleural effusion. In this case report, the probable mechanism of pleural effusion was discussed.

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Key words: Chondrosarcoma, Dischondroplasia, Enchondroma, Haemangioma

Case Report

A 23-year-old male patient was admitted to Ataturk Chest Disease Hospital with non-productive cough, dyspnoea, chest pain and back pain. These complaints had begun 15 days before he was admitted. He was also suffering from gingival bleeding for the past 1 year.

In his medical history, no physical or mental pathology had been noted until he was 7 years old. At the age of 7, he started to complain of swellings on his hands and feet. The swellings were either soft or hard and did not cause any pain. As a result of minimal trauma, he had fracture of the arm at the age of 13. There was no family history. His vital signs were normal. His height was 145 cm and he weighed 47 kg.

Disseminated gingivitis and gingival hyperplasia was present. There were mobile, multiple, bluish-coloured lesions probably cavernous haemangiomas on bilateral chest walls. Breathing sounds were decreased in the left hemithorax and were absent below the scapula.

Shortness of stature and deformities of the upper and lower extremities, particularly on the right side, was present. In addition, there were multiple nodular lesions on the extremities specially accumulated on the fingers (Fig. 1). They were similar to the ones on the right thoracic wall and matched cavernous haemangiomas. The nodular lesions on the left thoracic wall were probably of bony origin. No abnormal finding was present in the other systems.

Complete blood count and routine biochemical analysis of blood and urine were normal. The electrocardiogram was normal. Respiratory function tests revealed a moderate restrictive type ventilation defect. Pleural fluid was exudative and haemorrhagic and the fluid haematocrit was 18%.

Chest X-ray revealed displacement of the mediastinum to the right. A uniform opacification was present at the left hemithorax suggesting pleural effusion. The concave medial border started from the second front rib extending upward

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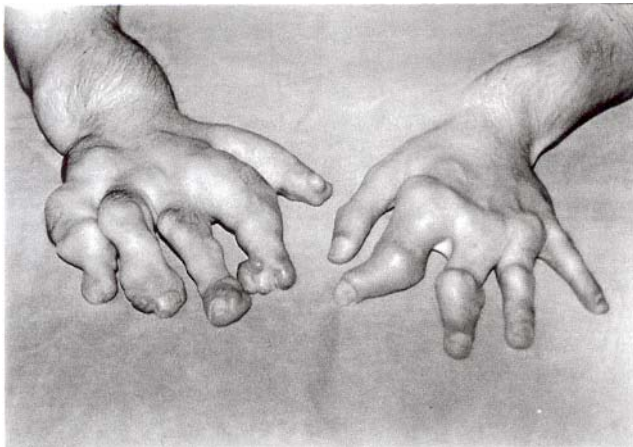


Fig. 1. Multiple soft nodules on the hands.

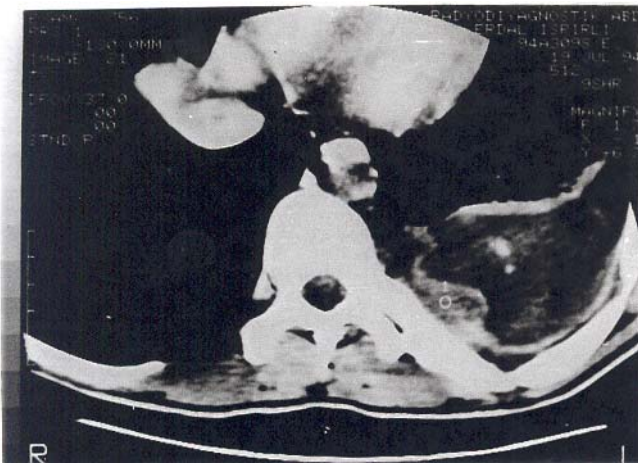


Fig. 3. CT scan on admission showing pleural effusion and pleural thickening. A 64 x 30 mm enchondroma was detected on the rib.

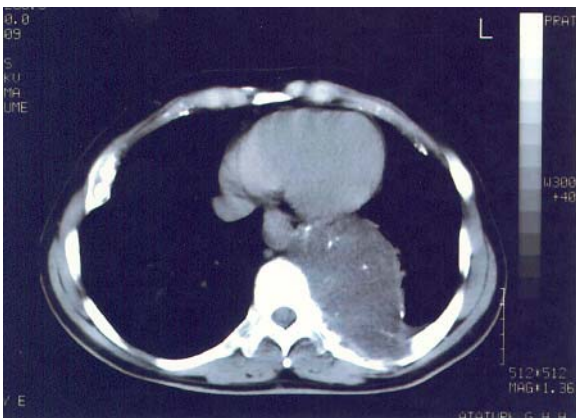


Fig. 5. CT showing significant progression in the lesion that was suggested as chondrosarcoma.

to the axilla, obliterating the diaphragm, costodiaphragmatic and costophrenic sinus. The underlying skeletal structures were irregular. There were also calcified lesions causing expansion of the bones, on the 8th and 9th back ribs, and



Fig. 2. Tc 99m labelled erythrocyte pooling scintigraphy revealing multiple hypervascular lesions due to haemangiomas on the right hand.



Fig. 4. Angiography of the right upper extremity. Haemangiomas in the form of pooling and curls were detected in the brachial, radial and ulnar arteries.

scapulas bilaterally.

Calcified lesions were seen in the feet, hands, pelvis, long bones, scapula and the ribs accumulating on the right side. The bony structures were expanded causing cortical irregularity.

Multifocal activities were present on both hands and feet, especially on the right side. Tc99m labelled erythrocyte pooling bone scintigraphy revealed multiple hypervascular lesions probably matching the haemangiomas on the extremities, especially on the right hand and forearm (Fig. 2).

Multiple expansile mass lesions, matching the enchondromas, were present in the scapula and the ribs bilaterally. There was massive pleural effusion and pleural thickening in the left hemithorax. A 64 x 30 mm enchondroma was detected in the rib adjacent to the basal segments of the left lung (Fig. 3).

There were diffuse parietal and visceral pleura thickening at the level of T₈ vertebral body adjacent to the lesion situated proximally to the left rib. Massive pleural effusion was present in the left hemithorax.

Imaging specific to the haemangioma could not be viewed, at the site of the lesion. In the right upper extremity, angiography showed opacification which matches the haemangioma, in the form of pooling and curls, detected in the brachial, radial and ulnar arteries (Fig. 4).

According to these laboratory findings and imaging procedures, Maffucci's syndrome (MS) was suggested and consultations confirmed the diagnosis. Left thoracentesis was performed and the effusion obtained was haemorrhagic. In order to evaluate the pleura and the lesion situated on the left thoracic wall, thoracoscopy was performed. 2000 L haemorrhagic effusion was drained. Parietal and visceral pleura were found to be thickened. A red-coloured lesion with an irregular surface was seen on the posterior wall of the left hemithorax. Pathological diagnosis of the biopsies taken from the lesion was chronic non-specific pleuritis. Cytology of the effusion was classified as Class II.

When computed tomography (CT) of the thorax and magnetic resonance imaging were evaluated concurrently, no difference could be found in signal intensity and opacification between the other enchondromas or haemangiomas and the lesion on the left posterior rib probably causing the hemothorax. Malignant degeneration was not suggested as no early arterial colouring was seen on the angiogram.

No fluid accumulation was detected after thoracoscopy. The patient was followed-up regularly, with CT of the thorax performed every 6 months. During the first 3 years, there was no progression of the lesion. A slow progression was observed in the fourth year. As the patient rejected any diagnostic interventions, he was monitored with CT. During the last 6 years of follow-up, the lesion showed significant progression (Fig. 5). Consultations were made with orthopaedic surgeons and oncologists. The lesion was diagnosed as chondrosarcoma and surgery was offered, but the patient refused surgery. At the end of the tenth year, follow-up showed progression of the lesion.

Discussion

MS is a non-hereditary congenital mesodermal hyperplasia characterised by enchondromas, secondary bone deformities and mesenchymal neoplasias.¹ Mesenchymal neoplasias are seen in the form of soft tissue haemangiomas and may be associated with phlebolitis. In addition to these pathologies, lymphangiomas, lymphangiectasia, phlebectasia and pigmentary macules have also been reported.² The syndrome was first coined by Maffucci in 1881 and, to date, approximately 200 cases have been reported.^{3,4}

The aetiology is unknown. It can occur in all races and there is no sex predilection.⁵ The patients are generally normal at birth. The average age at which symptoms

develop is 5 years, but there are reported cases where symptoms occur at the age of 1.³ The progression of bone and vascular lesions generally ceases to precede in the second decade.⁶

Skeletal changes seen are believed to be caused by a congenital defect in the enchondral ossification. Consequently, the defect affects the growing ends of the bones and irregular growth. Dischondroplasia occurs. Islands of cartilage proliferates and cartilagenous enchondromas develop at the surface of the bone.⁵ The lesions most commonly affects metacarpals, hand phalanges and bones of the feet, tibia, fibula, radius and ulna. Vertebra, ribs, scapula, pelvis and the cranium may also be involved. Enchondromas are generally asymmetric in distribution.⁷ In our case, multiple enchondromas were noted in long bones, hand and feet bones, pelvis, scapula and ribs. The patient noted that these lesions started to occur at the age of 7 and there had been no progression of the lesions in the last 4 years.

Along with the enchondromas, superficial and deep haemangiomas are also seen. Soft tissue lesions generally develop along with the bone lesions. Vascular lesions seen in this syndrome are subcutaneous capillary and cavernous haemangiomas in various dimensions. Haemangiomas may be situated in the brain, eyes and gastrointestinal tract. The distribution of these lesions may not be associated to the distribution of the skeletal lesions.^{5,6,8}

Sarcomatous transformation is seen in bone and soft tissue lesions in MS but the risk is more often in enchondromas, the incidence being 15% to 57%. The overall risk of malignant transformation in this syndrome is 25%.^{1,6,8-10}

Pathological fractures are commonly seen with an incidence rate of 26%. Other complications include haemorrhages from the haemangiomas, shortness of stature and cranial nerve paralysis produced by the compression of the enchondromas.

Though there were multiple haemangiomas, most of them were situated on our patient's hands. He had no haemorrhagic complaint and his stature was below normal standards.

Pleural effusion associated with MS has not been reported until now. The unilateral haemorrhagic pleural effusion detected in our case may be related to 3 causes.

1. Enlarging enchondromas causing pleural irritation and lymphatic obstruction conventionally may result in a reactionary pleural effusion.
2. In literature, gingival biopsies performed in patients with MS showed abnormal capillary proliferation. In the angiography performed in our patient, there was increased vascularisation. Pleural effusion may be

caused by vascular instability because of microtrauma.

3. Haemorrhage and irritation of an enchondroma by a pathological fracture may be the cause of the pleural effusion.

The patient has been followed-up for 10 years. Though the lesion has shown significant progression, pleural effusion has not recurred. It is known that in MS, chondromas have the potential for malignant transformation into chondrosarcomas.¹¹ During the last 10-year of follow-up, the lesion revealed a slow progression beginning from the fourth year. Presently, there is no reasonable scientific explanation for the undulating growth rate. Surgical options and radiotherapy will be considered in the event of further increase in size.¹¹

Clinical signs and natural history of the lesion favoured the diagnosis of chondrosarcoma in our patient.

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