Atypical Synovial Sarcoma: Rare Presentation of a New Variant

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Abstract

Medical literature in regards to synovial sarcomas is scarce. This case report displays an atypical presentation of this deadly musculoskeletal pathology. The consequence of death is the result of not recognizing the pattern of disease as described in this paper. Less than 1% of all malignancies are attributed to soft tissue sarcomas, and synovial sarcomas are only 6-8% of this larger category. This patient presentation is unique in numerous regards. Stark differences in the presentation of this patient lends credence to the proposal to the discovery of a new highly invasive subtype of synovial sarcoma. The various subtypes of this form of cancer are sporadically discovered and the recognition is the first step in devising appropriate treatments to save life and limb.

INTRODUCTION

This case report displays an atypical presentation of a deadly musculoskeletal pathology. The consequence of death is the result of not recognizing the pattern of disease as described in this paper. Proper awareness in the orthopedic community is vital is properly treating these patients.

A soft-tissue sarcoma is a malignancy arising from mesenchymal tissues, found throughout the body [1]. They are malignant tumors of extra-skeletal and non-epithelial tissues, such as muscle, fat, fibrous tissue, blood vessels and components of the nervous system. Soft tissue sarcomas are classified on a histological basis according to the adult tissue-form they resemble or are derived from. Synovial sarcomas represents one form of soft tissue sarcomas [2].

The pathogenesis of synovial sarcomas is not completely understood, therefore not many well-established risk factors exist. However, one risk factor has been proven to be of greatest importance in regards to susceptibility to synovial sarcomas, that being predisposition via genetic means, as revealed by family history's. These include a history of polyposis, neurofibromatosis, retinoblastoma, any cancer at young age in first-degree relatives, or prior sarcomas [3].

Soft tissue sarcomas are very rare tumors. They have an annual incidence is around 2-3/100,000, accounting for <1% of all malignancies. Although they account for a very small percentage of total malignancies, they are responsible for 2% of all cancer-related deaths. Therefore, they carry a high rate of mortality [4]. Synovial sarcomas account for only 6-10% of all soft tissue sarcomas. It is apparent that there is a large void in the medical literature regarding this musculoskeletal neoplasm. The lack of knowledge combined with the high mortality rate of this disease process, makes this a very dangerous condition, especially because numerous variants of this disease are sporadically discovered.

The incidence peaks in the 3rd decade (30% of cases occurring in patients less than 20 years of age). Males are affected more often than females (male: female ratio is 1.2:1), as shown in both long-term and short-term studies. Incidence by race reveals 86% of the patients are white, 10% are black, and orientals only account for 1% of the patient population [2, 5].

Analysis of soft tissue sarcomas distribution by anatomic site reveals more than half (59.5%) of the primary lesions are on extremities, but the lower extremities have the greatest preponderance of lesions by far [5]. Of extremity synovial sarcomas, studies have shown the knee region to have the highest incidence of primary tumor, and second highest incidence of bone metastases (second only to foot/ankle synovial sarcomas) [1].

Synovial sarcomas (particularly those of high-grade) can metastases, but do so in less than 1/3 of patients, predominantly via hematogenous spread. Lung metastases are by far the most common site (23% occurrence) and most life-threatening location of neoplastic spread. The most common metastatic route for extremity soft-tissue sarcomas to the lungs is the venous system. Metastases to other sites such as the brain, liver, and soft tissue distant from the primary tumor site are competitively very rare. Liver metastases are found at a considerably lower occurrence [4-6]. Liver involvement by limb or trunk synovial sarcomas is infrequent, with a documented occurrence of 0.5% to 1.1% of diagnosed patients [7].

In a review of 874 patients with soft tissue sarcomas of the extremity, 5.5% had evidence of bone invasion. Of the 5.5%, only 7.5% were from individuals with the synovial sarcoma subtype. Patients with boney invasion more often initially presented to health care providers with larger sized tumors deep in the extremity and more often have metastases at presentation [1].

In extremely rare incidences where metastatic spread or local neoplastic invasion from synovial sarcomas into bone actually does occur, it will commonly causes osteolytic destruction. Destruction of bone in synovial sarcomas has been shown in studies to lead to pathological fracture in just less than half of the metastatic bony lesions [6].

The various symptoms of synovial sarcomas are related to location and tumor mass. The major presenting symptom in both long-term and short-term studies reveals the presence of a mass (64%) to be the most common initial presentation. Pain was only found to be present in 1/3 of patients upon initial presentation [2, 5]. The majority of these tumors are deep seated, imbedded in musculature, ligamentous and tendonous structures. They are therefore buried within the deeper layers of tissue. Previous studies have revealed only 23.4% of patients show evidence of superficial tissue invasion [1].

Pain is often related to involvement of nerves with the tumor mass. Tumors of synovial sarcomas grow slowly, and symptoms may be present for a long time before diagnosis is made, therefore leading to a delayed diagnosis [2].

Limb salvage surgery in combination with adjuvant radiotherapy, is generally recognized as the principal treatment for synovial sarcomas of the extremities. Preservation of the limb allows for highly acceptable functional outcomes in the patients, while still removing the primary tumor. Furthermore, when limb salvage surgery is combined with adjuvant radiotherapy, the risk of tumor recurrence is similar to that reported in patients treated with complete amputation. Despite this fact, amputation remains as a popular option for many physicians [1].

CASE REPORT

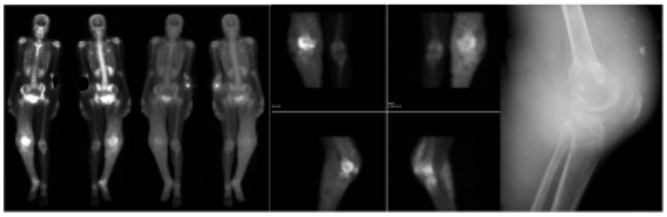
The patient is a 29-year-old female, Nigerian native, presenting to medical professionals with complaints of decreased range of motion and pain about the right knee. At initial presentation, gross examination revealed no mass or deformity at the knee. Post initial orthopedic consultation, the patient was deemed to have infrapatellar syndrome, and subsequent arthroscopic surgery was performed. She underwent a synovectomy and shavings were taken from the cartilage at the right knee during the arthroscopic procedure. These samples were sent for pathological review and clinical data concluded the diagnosis of a Stage 3 High Grade Biphasic Variant Synovial Sarcoma of the soft tissue about the right knee joint. The right knee at the time was determined to be the sole location of neoplastic growth in the body and the primary source. Lost to non-compliance, the patient emerged 3 years later to a different orthopedic surgeon, with the inability to fully extend or flex the right knee at the joint secondary to mass effect of the enlarged tumor. MRI of the right knee revealed a multi-lobulated heterogeneous enhancing mass in the anterior and posterior aspects of the knee. The tumor mass completely encased the femoral neurovascular bundle. At this time, the patient refused surgical intervention, but agreed to chemotherapy. The neoplastic process did not seem to respond in any significant way to the prescribed chemotherapeutic treatment over the course of 1 year.

Progression of the disease lead to extensive lytic lesions of the distal femur, contributing to a supracondylar comminuted pathological fracture. Destructive neoplastic changes of the distal femur, patella, proximal tibia and proximal fibula we all noted at this time. MRI of the right knee revealed a large enhancing multilobulated right thigh and leg soft tissue mass grossly measured at 20.2 cm in the anterior-posterior plane, 20 cm in the transverse plane, and 33 cm in the cranio-caudal dimensions. The femoral neurovascular bundle was encased at multiple levels by the neoplastic lesion. Range of motion evaluation revealed the patient was unable to perform any active or passive range of motion at the right knee. She had chronic pain of the right lower extremity and she was very guarded in terms of any

type of motion. The knee at this time was noted to have "massive swelling…with deformity from tumor growth". An open wound on back of knee was draining a small amount of serous material. Scintigraphy via technetium 99m revealed axial and appendicular skeleton involvement (Image 1). A CT guided core biopsy of pulmonary lesions revealed a Stage 4 High Grade Biphasic Synovial Sarcoma, with metastasis to the chest and throughout the body.

Figure 1

Scintigraphy and Plain film Imaging



The debilitating orthopedic sequela of the sarcoma made the patient dependent on the care of others. Based on the patient's age being so young, the family and patient reportedly wished to pursue aggressive treatment at this time, although the patient died from complications related to the malignancy before further treatment was received.

DISCUSSION

Medical literature in regards to synovial sarcomas is scarce. This is in part due to the rarity of occurrence of this disease process. Less than 1% of all malignancies are attributed to soft tissue sarcomas, and synovial sarcomas are only 6-8% of this larger category. In conclusion, synovial sarcomas are exceedingly rare and remarkably invasive.

This patient presentation is unique in numerous regards. The patient in this case report did not have a family history of any neoplastic process at all, much less those listed as risk factors. Most of the patients effected are white (86%) males (1.2: 1), in stark contrast to this African female.

The neoplasm only metastasizes in 1/3 of patients, most commonly to the lungs (23%), but it occurs in the liver in .5-1.1% of patients. Interestingly, this patient had metastatic infiltration at both sites. Boney invasion in soft tissue sarcomas only occurs at a rate of 5.5%, and is rarer in synovial sarcomas (in 7.5% of the 5.5% of soft tissue sarcomas with invasion). Only 1/3 of patient present with pain as their initial symptom, as was seen in this case. The patient also displayed superficial invasion of tissue, occurring in only 23% of documented cases, and pathological boney fracture, seen in less than half of patients. Synovial sarcomas are much more commonly purely deep structures, but this neoplastic process displayed characteristics of both, which adds to the evidence of the severe invasive nature of this individual's unique neoplastic subtype.

CONCLUSION

Syndromes or particular pathologies are characterized by a similar constellation of signs, symptoms, and epidemiological factors. Stark differences in the presentation of this patient, lends credence to the proposal of the discovery of a new highly invasive subtype of synovial sarcoma. The various subtypes of this form of cancer are sporadically discovered and the recognition is the first step in devising appropriate treatments to save life and limb.

This case displays a large combination of highly rare presentations for a pathology which is already very seldom seen in itself. The statistical probability of all of these rare presentations occurring together is tremendously small. These facts could very likely be attributed to a strain of synovial sarcoma the scientific community is unfamiliar with. As previously stated, synovial sarcomas are not an extensively studied entity, and this case report calls for more research in this field. It is the role of the astute orthopedic surgeon to recognize the constellation of abnormal signs and symptoms in the patient with synovial sarcoma, and intervene surgically when appropriate. The high level of clinical knowledge and surgical capabilities of the well trained orthopedic surgeon must be applied to successfully treat patients and avoid debility, and ultimately death.

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