Rapidly progressive synovial sarcoma with multiple metastases leading to intestinal obstruction

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Abstract

A 34-year-old man complained of radiating pain in the left lower extremity for 1 week,and diagnosed as soft tissue sarcoma of the left hip with pulmonary metastasis.The postoperative pathology report was poorly differentiated synovial sarcoma.Intussusception and liver metastases were found 1 week after surgery,and then rapidly deteriorated and died.

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1 | INTRODUCTION

Synovial sarcoma is a relatively rare soft tissue malignancy, accounting for about 10% of primary soft tissue sarcomas[1]. The occurrence of the disease is mostly located around the proximal joints of the limbs, some of which occur in the head, face and viscera. Multiple metastases other than lung and bone are rare[2]. Today, more than half of the patients live for more than 5 years after treatment[3]. However, a few patients progress rapidly, even almost without treatment opportunity.

2 | CASE REPORT

We report a case of rapidly progressive synovial sarcoma with multiple metastases. A 34-year-old young man complained of radiating pain in the left lower extremity for 1 week. X-ray examination of the hip was performed in another hospital and was misdiagnosed as lumbar disc herniation. Later, in our hospital, the diagnosis was made of soft tissue sarcoma of the left hip with pulmonary metastasis. We performed a biopsy procedure on the patient. The postoperative pathology report was poorly differentiated synovial sarcoma. Intussusception and liver metastases were found 1 week after surgery, and then rapidly deteriorated and died. We performed a high-throughput gene comparison with 2 synovial sarcoma patients of the same age and type with good prognosis. The study found that this patient had a significant genetic mutation compared to patients with a good prognosis.

The patient was previously healthy, denied a history of chronic disease and immune system disease, had no recent renovation of the house, and had no radiation exposure. He denied history of smoking or drinking, but stayed up late frequently. He denied any family history of cancer and sarcoma.

The left hip soft tissue density increased on the X-ray before admission, but it was relatively insidious and there was a significant increase in soft tissue tension in the left hip. Admission laboratory values revealed a anaemia with hemoglobin of 82 g/L and hypoalbuminemia of 23.1 g/L, D-dimer 5.31mg/L. After admission, the patient's first fecal routine suggested occult blood positive. MRI showed multiple irregular occupying

tumor around the left hip with uneven mixed signals and lobulation. CT demonstrated multiple solid nodules in the lungs (Figure 1).

We performed biopsy for the patient. The postoperative pathological results showed that the tumor was poorly differentiated synovial sarcoma. Immunohistochemistry: Vimentin(+++), TLE1(+++), CD99(++), WT-1(++), Bcl- 2(++) , CKpan(-) , CK7(-) ,EMA(-) , S-100(-) , STAT6(-) , CD34(-) , CD31(-) , ERG(-) , SMA(-) , Desmin(-) , CD117(-) , calponin(-) , HBME-1(-) , calretinin(-) , ALK P80(-) , Ki-67(35%+)(Figure 2). One week after the operation, the patient complained of unbearable abdominal pain, and he underwent abdominal CT scan + enhancement. The results showed that the ascending colon was unevenly thickened with intussusception, and there were multiple round lesions in the liver. After consultation with general surgery department, the patient was considered to have surgical indications, and he was transferred to general surgery department for continued treatment. Intestinal obstruction catheter drainage was temporarily administered. After 4 days, the patient's abdominal pain worsened again. His pulmonary and liver nodules were significantly larger than 4 days ago, the progress was rapid(Figure 3).

The patient's family refused to undergo palliative bowel surgery, and the patient died three days after the last CT scan, less than a month from admission to death. In order to explore why the synovial sarcoma of this patient progressed so rapidly, we selected two patients with synovial sarcoma of the same age and gender with good prognosis as controls, performed peripheral blood high-throughput genome sequencing, and performed a preliminary comparative analysis of the results. The results showed that this patient had more significant gene mutations than the 2 patients with good prognosis. The Single Nucleotide Polymorphisms(SNP) showed that the mutation types were mainly homozygous mutations, and the number of mutations was significantly up-regulated in each mutation type(Table 1). The visual scatter plot and volcano plot showed that 820 and 822 genes were up-regulated, and 326 and 225 genes were down-regulated, respectively. The patient and the two controls had a significant intersection on the differential mutation catalog. GO and KEGG results suggest that significantly enriched related pathological pathways include tumor immune examination, transcriptional dysregulation, osteoclast activity, antigen presentation, and T cell function (Figure 4).

3 | DISCUSSION

Synovial sarcoma is generally considered a high-grade aggressive sarcoma, with a poorer prognosis in adults than in children[4]. Prognostic factors also include tumor size, use of radiotherapy and/or chemotherapy, histological subtype, and surgical margin status, among which tumor size >5 cm at the time of treatment is consistently associated with poor prognosis[5]. In addition, synovial sarcoma has a high metastatic potential, and its metastatic rate can reach more than 50%[6]. The most common metastatic site is the lung[7]. Other metastases include lymph, bone, and liver, but patients with gastrointestinal metastases are rare. At present, there were only a few dozen cases of metastatic synovial sarcoma in the gastrointestinal tract had been reported in the literature[8].

In addition to intestinal metastases, the patient was characterized by a particularly rapid progression. It was only 20 days from the time of admission to the patient's death. Although this was related to the delay in the patient's visit to the hospital, the progression of metastases in the patient after admission was very rare. The interval between two CT scans was only 4 days, but the liver and intestinal metastases progressed significantly. Due to the rapid progression, patients lost opportunities for surgery and chemotherapy.

Synovial sarcoma has typical pathological and genomic features including immunohistochemical features and a t(X;18)(p11.2;q11.2) chromosomal translocation in which the SS18(SYT) gene (18q11) is associated with SSX (including SSX1) , SSX2, SSX4, Xp11) gene fusion, resulting in the generation of the SS18-SSX fusion gene, which occurs in most patients[9]. Some RNAs may act in concert with fusion genes to cause rapid tumor progression, but the exact map has not yet been determined[10]. Gene sequencing of this patient showed a significantly higher number of homozygous mutations compared to conventional synovial sarcoma patients. The pathological mechanisms involved in these mutated genes are all involved in immune system exhaustion and tumor transcriptional dysregulation, which may be the reason for the rapid tumor progression in this patient. Humoral and cellular immunity, including the functional expression of T17 and Th cells, may have

certain relevance to the progression and prognosis of synovial sarcoma[11]. Of course, this remains to be verified by further research.

Learning points

1. Abdominal pain in patients with synovial sarcoma should be alert to the possibility of gastrointestinal metastasis leading to intussusception and intestinal obstruction.

2. Genomic differences between rapidly progressive synovial sarcoma and normal patients should be noted by bone oncologist and investigators.

Contributors

X L, Z G and HZ X managed the entire course of the patient's disease. XW Y performed the gene sequencing and literature review. X L drafted and revised the paper.

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Competing interests

None declared.

Deceased Patient Permission

Obtained.

CONSENT

The authors certify that they have obtained all appropriate patient consent forms. The patient's wife and parents agreed to release the details of the patient's medical records and related images. The report of this case does not require approval by the relevant institution. In accordance with the journal's patient consent policy, the patient's family has written informed consent to publish this case report.

DATA AVAILABILITY STATEMENT

All data and images are available inside the paper.

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