Genomic panel testing and treatment attainment in malignant bone and soft tissue tumors

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[Introduction]

In Japan, cancer gene panel tests (CGP tests) will be covered by insurance in 2019. The test plays an important role in diagnosing and treating for various cancers. The purpose of this study was to determine the genetic abnormalities found in CGP tests performed on patients with malignant bone and soft tissue tumors from 2019 to 2023, and to evaluate the attainment rate and efficacy of the treatment.

[Methods]

Between April 2019 and October 2023, we retrospectively collected medical record of 41 patients enrolled in National Registry of Bone and Soft Ulcers of the Japanese Orthopaedic Association with malignant bone and soft tissue tumor: 23 patients with M0 and metastatic recurrence at the initial visit, 18 patients with M1 and N1 at the initial visit. The patients with M0 and no metastatic recurrence at the initial visit were excluded. The survey items included gender, age, diagnosis, stage of the disease, treatment details, stage of the disease at the time CGP testing was performed, CGP test results, genetic abnormality, attainment rate of the treatment, drugs used, treatment efficacy, and the outcome of the treatment.

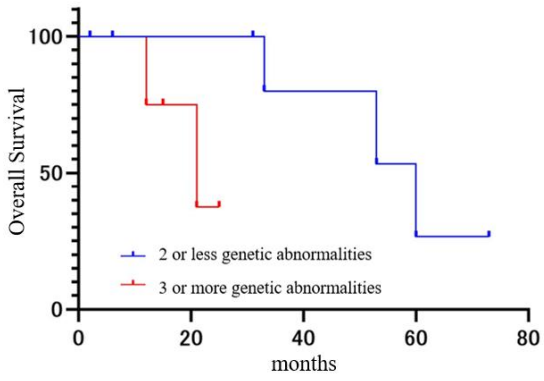
[Results]

CGP testing was performed in 12 cases (29.3%). The mean of genetic abnormalities per case was 6.83. The overall survival rate of 12 patients who underwent CGP testing was divided into 8 patients with 2 or less genetic abnormalities and 4 patients with 3 or more genetic abnormalities. Moreover, the group with 3 or more genetic abnormalities had a significantly worse prognosis ($p<0.05$). The reasons for not implementing CGP testing in 29 cases were: BSC migration in 9 cases, surgery in 6 cases, continued chemotherapy in 7 cases, and others in 7 cases.

[Discussion]

In this study, the prognosis was worse for patients with multiple detected genetic abnormalities, suggesting that oncogene and tumor suppressor gene abnormalities may also affect the prognosis of sarcoma. In addition, 9 of the 29 cases (31.3%) with no CGP testing were BSC migration. This suggests that many cases are difficult to continue active treatment at the end of standard treatment, and CGP testing should probably be performed during standard treatment.

Figure 1. Kaplan Meier curve for overall survival of patients with 2 or less genetic abnormalities and 3 or more genetic abnormalities



Log rank test: $p<0.05$

The patients were divided into two groups by the number of genetic abnormalities: 8 patients with 2 or less genetic abnormalities and 4 patients with 3 or more genetic abnormalities.

Table 1. Baseline characteristics of subject patients

Characteristics	Cases	Percentage(%)
Gender		
Male	27	65.9
Female	14	34.1
Age		
Average	61.15(10-90)	
Diagnosis		
Bone tumor		
Chondrosarcoma	2	4.9
Osteosarcoma	4	9.8
Soft tissue tumor		
Myxofibrosarcoma	4	9.8
Undifferentiated Pleomorphic Sarcoma	6	14.6
Malignant peripheral nerve sheath tumor (MPNST)	5	12.2
Liposarcoma	6	14.6
Leiomyosarcoma	2	4.9
Rhabdomyosarcoma	2	4.9
Malignant soft tissue tumor	2	4.9
Synovial sarcoma	2	4.9
Others	6	14.5
Primary tumor surgery		
Yes	29	70.7
No	11	26.8
Unknown	1	2.5
Rescue chemotherapy		
Yes	9	22
No	32	78

[Significance/Clinical Relevance]

It was suggested that a relationship between sarcoma and genetic abnormalities in prognosis.