

The Utility of Clinical Genomic Profiling in the Diagnosis and Treatment of Sarcomas

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INTRODUCTION:

The utility of comprehensive genomic profiling (CGP) test was not fully analyzed in patients with sarcoma. We examined the detection rate of actionable mutations and the corresponding treatments in patients who underwent comprehensive genomic profiling tests at our hospital.

METHODS: From 2019 to 2022, 84 patients with sarcoma underwent CGP at our hospital. We investigated the detection rate of actionable gene mutations, corresponding clinical trials/treatments for insurance coverage, and germline finding associated with hereditary diseases in these patients.

RESULTS:

The histological types included leiomyosarcoma in 19 patients, dedifferentiated liposarcoma in 13, osteosarcoma in 8, and other sarcomas in 44 patients. Actionable gene mutations were found in 38 (46%) patients, among which domestic clinical trials were available in 27 cases (33%). In 2 patients with inflammatory myofibroblastic tumor, a fusion mutation of *ALK* gene was identified and tumor shrinkage was observed with the ALK inhibitor. Microsatellite instability was detected in one patient with leiomyosarcoma, and tumor shrinkage was observed with an immune checkpoint inhibitor. Although histological diagnosis of extraskeletal myxoid chondrosarcoma was difficult, the *NR4A3-EWSR1* fusion gene, found in the comprehensive genomic profiling tests, contributed to the final diagnosis. Germline findings was found in 8 patients (10%); one patient with leiomyosarcoma, in whom *BRCA1* pathogenic variants were detected, was found to have breast cancer and pancreatic cancer in the patient's family, and further genetic testing revealed the germline *BRCA1* pathogenic variants.

DISCUSSION AND CONCLUSION:

The actionable gene mutations were found in approximately half of the patients with sarcoma by the CGP. These tests also contributed to the identification of the suspected germline findings. The comprehensive genomic profiling test appears to be an important tool in the diagnosis and treatment of sarcomas.