Myxoid liposarcoma with cartilaginous differentiation showing DDIT3 rearrangement

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Introduction: Myxoid liposarcoma (MLPS) is the second most common histologic subtype of liposarcoma. However, cartilaginous differentiation within MLPS is an extremely rare phenomenon. In this report, we presented a case of MLPS with cartilaginous differentiation in which the FUS-DDIT3 fusion gene was detected by fluorescence in situ hybridization (FISH) analysis and reviewed the literature on MLPS with cartilaginous differentiation.

Case Presentation: A 44-year-old woman had noted a painless mass in the left thigh. The mass had been present for 4 years and had slowly increased in size. Magnetic resonance imaging revealed a 21-cm mass spreading on the lateral muscle component on the axial aspect of the left thigh. The lesion appeared heterogeneously hyperintense and partially hypointense on T2-weighted images. A needle biopsy was performed, and we diagnosed as MLPS. The mass was excised with wide margins, and a sterile portion was submitted for cytogenetic analysis. Histopathological findings showed that a liposarcomatous area was adjacent to the cartilaginous area, which accounted for about 20% of the tumor, had a cartilaginous component with mild cellularity of mature chondrocytes and focally atypical chondrocytes. Based on these findings, the diagnosis of MLPS with cartilaginous differentiation was made. In the cytogenetic analysis, eighteen of the 20 analyzed metaphase cells were characterized by t(12;16)(q13;p11.2). An interphase FISH analysis showed that at least 10% of the cells from both the typical MLPS area and the cartilaginous component area showed a split signal pattern, demonstrating a rearrangement in the DDIT3 gene. The patient developed distant metastasis to the subcutaneous tissue of axilla and lymph node of posterior mediastinum in postoperative 11 months and received continuous chemotherapy.

Discussion: MLPS with cartilaginous differentiation is extremely rare, and there have been only the eight previous cases including our case. In the areas of both typical MLPS and cartilaginous differentiation, the FUS-DDIT3 fusion gene might have the important role for oncogenesis. To date, in the previous reports, it has been suggested that the presence of cartilaginous differentiation with MLPS may be regarded as a good prognosis factor. However, our case developed distant metastasis after operation. Therefore, the relationships between the clinical significance and the presence of cartilaginous differentiation in MLPS are unclear. The mechanism of cartilage differentiation within MLPS and its clinical significance will become clear in the future with examination of more cases.

Keywords : Myxoid liposarcoma, cartilaginous differentiation, cytogenetics, DDIT3 rearrangement

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