Abstract. Fibroma of tendon sheath is an uncommon, benign fibroblastic tumor that usually occurs in the upper extremities of young and middle-aged adults. A clonal chromosomal aberration, t(2;11)(q31-32;q12), has been described in one case. We herein present a unique cytogenetic finding of fibroma of tendon sheath arising in the first web space of the right hand of a 38-year-old woman. Physical examination showed a 3.5-cm, firm, mobile, non-tender mass. Magnetic resonance imaging showed a well-defined soft tissue mass with iso- to slightly-low signal intensity relative to skeletal muscle on both T1- and T2-weighted sequences. Contrast-enhanced T1-weighted sequences demonstrated moderate patchy enhancement of the mass. A fibroma or giant cell tumor of tendon sheath was suggested, and the lesion was marginally excised. Histological examination confirmed the diagnosis of fibroma of tendon sheath. Cytogenetic analysis revealed a novel t(9;11)(p24;q13-14) translocation among other karyotypic abnormalities. The postoperative course was uneventful, and the patient is doing well without local recurrence two months after surgery. To the best of our knowledge, this is only the second report of fibroma of tendon sheath with clonal chromosomal abnormalities.

Fibroma of tendon sheath is a rare, benign fibrous soft tissue tumor of unknown pathogenesis. It has a peak incidence in the third to fifth decades of life, with a male predominance (1). Fibroma of tendon sheath typically presents as a small, firm, slow-growing, painless mass in the peripheral extremities, especially the fingers and hands. Surgical excision is curative (2). There is no risk of metastasis or malignant transformation.

Histologically, the lesion is well-circumscribed, lobulated, and consists of spindle-shaped cells embedded in a collagenous stroma. A characteristic feature is the presence of elongated cleft-like spaces lined by flattened cells (1). Cytological atypia is quite uncommon. Immunohistochemically, the neoplastic cells are positive diffusely for vimentin and focally for smooth muscle actin (1).

Fibroma of tendon sheath is a rare, benign fibrous soft tissue tumor of unknown pathogenesis. It has a peak incidence in the third to fifth decades of life, with a male predominance (1). Fibroma of tendon sheath typically presents as a small, firm, slow-growing, painless mass in the peripheral extremities, especially the fingers and hands. Surgical excision is curative (2). There is no risk of metastasis or malignant transformation.
with pearly white color. Microscopically, the tumor was composed of bland fibroblastic spindle cells in an abundant collagenous stroma. Elongated cleft-like spaces lined by flattened cells were also seen (Figure 2). Neither cellular atypia nor mitotic figures were observed. Based on these findings, the tumor was diagnosed as a fibroma of tendon sheath.

Cytogenetic analysis revealed the following karyotype: 46,XX,t(9;11)(p24;q13-14)[10]/46,XX,t(1;20)(p13;q13.1)[2]/46,XX,t(1;2)(q21;q35)[1]/46,XX,t(3;16)(p21;q24)[1]/46,XX[3] (Figure 3).

The postoperative course was uneventful, and the patient is doing well without local recurrence two months after surgery.

Discussion

Only one case of fibroma of tendon sheath has been cytogenetically analyzed to date (3). A t(2;11)(q31-32;q12) chromosomal translocation has been identified. Notably, the same translocation has been also observed in a subset of collagenous fibromas (4-6), suggesting a genetic link between these two entities. Collagenous fibroma, also known as desmoplastic fibroblastoma, is a benign fibrous soft tissue tumor that usually occurs in the subcutaneous tissue of upper extremities. It has a peak incidence in the fifth to seventh decades of life, with a male predominance (7). On MRI, collagenous fibroma typically appears as a well-defined mass with low to intermediate signal intensity on T1-weighted images and low to slightly high signal intensity on T2-weighted images (8). Collagenous fibroma demonstrates mild internal enhancement with rim enhancement (8,9). Histologically, the lesion is hypocellular and consists of spindle to stellate-shaped cells embedded in a collagenous stroma, displaying a similar morphology. Recently, Macchia et al. proposed that FOS-like antigen 1 (FOSL1) is a candidate target gene for 11q12 rearrangements in collagenous fibroma (10). FOSL1 encodes a nuclear leucine zipper protein that can dimerize with proteins of the Jun family, thereby forming the activator protein-1 transcription factor complex (11). The expression of FOSLI is very low in normal cells. Elevated FOSLI mRNA and protein have been detected in a variety of human tumors (11). However, it is uncertain if FOSLI gene rearrangements are involved in fibroma of tendon sheath.

In the current study, we identified a novel t(9;11)(p24;q13-14) translocation among other clonal abnormalities. To the best of our knowledge, chromosomal rearrangements bearing t(9;11)(p24;q13-14) have never been recorded in mesenchymal neoplasms. The breakpoint on the long arm of chromosome 11 was somewhat different from that of a previously reported case. This observation may be explained by the possibility that the previously reported breakpoint is rearranged cryptically.
and not identifiable by G-banding. On the other hand, chromosome 9p24 region contains several important genes, including Janus kinase 2 (JAK2). JAK2 encodes a cytoplasmic tyrosine kinase involved in a specific subset of cytokine receptor signaling pathways (12). JAK2 translocations have been described in hematological malignancies (12, 13).

The differential diagnosis of fibroma of tendon sheath includes giant cell tumor of tendon sheath, as in our case. Despite a distinct morphological appearance, these two lesions can exhibit similar signals on T1- and T2-weighted images. Fibroma of tendon sheath can have a variable enhancement pattern, ranging from no appreciable enhancement to marked enhancement (14). On the other hand, giant cell tumor of tendon sheath typically shows marked enhancement (15). Gradient-echo images of giant cell tumor of tendon sheath may reveal a blooming artifact due to the presence of hemosiderin (16). Cytogenetically, giant cell tumor of tendon sheath is characterized by a translocation t(1;2)(p13;q37), resulting in a collagen type VI alpha-3 (COL6A3)-colony stimulating factor 1 (CSF1) fusion gene (17).

In summary, we reported on a case of fibroma of tendon sheath with a unique 11q rearrangement. Further studies are required to elucidate the significance of this chromosomal alteration in the pathogenesis of fibroma of tendon sheath.

Acknowledgements

This study was supported in part by the Foundation for the Promotion of Medical Science and JSPS KAKENHI Grant Number 25462355.

References


Received May 6, 2014
Revised June 21, 2014
Accepted June 23, 2014