Collagenous Fibroma (Desmoplastic Fibroblastoma) with Trisomy 8 as the Sole Cytogenetic Abnormality

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Abstract. Collagenous fibroma (desmoplastic fibroblastoma) is a benign soft tissue tumor that usually occurs in the subcutaneous tissue or skeletal muscle of adults. Recent cytogenetic studies have revealed clonal rearrangements of the chromosomal band 11q12. We present a unique case of collagenous fibroma arising in the right shoulder of a 63-year-old female. Magnetic resonance imaging showed a solid soft tissue mass deeply relative to the deltoid muscle, with low-to-intermediate signal intensity on T1-weighted sequences and low-to-slightly high signal intensity on T2-weighted sequences. Contrast-enhanced fat-suppressed T1-weighted sequences demonstrated heterogenous internal enhancement with rim enhancement. Following an open biopsy, marginal excision of the tumor was performed. Histological examination confirmed the diagnosis of collagenous fibroma. Cytogenetic analysis displayed a simple karyotypic change with trisomy 8. The postoperative course was uneventful, and the patient is doing well without local recurrence two months after the surgery. To the best of our knowledge, this is the first case of collagenous fibroma with trisomy 8 as the sole cytogenetic abnormality.

Collagenous fibroma, also known as desmoplastic fibroblastoma, is a rare, benign soft tissue tumor first described by Evans (1) in 1995. It belongs to the fibroblastic/myofibroblastic tumor group according to the 2013 World Health Organization Classification of Soft Tissue Tumors (2). Collagenous fibroma has a peak incidence in the fifth to seventh decades of life, with a male predominance. Few cases have been reported in children (3, 4). The most common presentation is of a firm, slow-growing, painless, subcutaneous mass arising in the upper extremities (5). Simple excision is curative, and local recurrence has not been documented (2). It is important to distinguish collagenous fibroma from intermediate and malignant soft tissue tumors to avoid unnecessarily extensive surgery.

Histologically, the lesion is hypocellular and consists of spindle to stellate-shaped cells embedded in a collagenous or myxocollagenous stroma. Mitotic figures are uncommon. Immunohistochemically, the neoplastic cells are diffusely positive for vimentin and focally positive for smooth muscle actin. Occasional cases may exhibit scattered cells that stain for cytokeratin (6). Immunostains for desmin, S-100 protein, and CD34 are typically negative.

Only 10 cases of collagenous fibroma have been cytogenetically characterized (7-12). The long arm of chromosome 11, in particular 11q12, is involved in all cases except for one. Notably, an identical reciprocal translocation, t(2;11)(q31;q12), has been detected in two cases (7, 8). In the present report, we describe the first case, as far as we are aware, of collagenous fibroma with trisomy 8 occurring in the right shoulder of a middle-aged woman.

Case Report

A 63-year-old Japanese woman presented with a palpable mass in the right shoulder. There was no history of antecedent trauma. Physical examination revealed a 5-cm, elastic-hard, immobile, non-tender mass. Neurological and vascular examinations were unremarkable. Magnetic resonance imaging (MRI) demonstrated a well-defined soft tissue mass, deep in relation to the deltoid muscle. The mass exhibited low-to intermediate signal intensity on T1-weighted sequences (Figure 1A) and low-to-lightly high signal intensity on T2-weighted sequences (Figure 1B). Contrast-enhanced fat-suppressed T1-weighted sequences demonstrated heterogenous internal enhancement with rim enhancement (Figure 1C). Our preoperative differential diagnosis included collagenous fibroma and desmoid-type fibromatosis.
An open biopsy was performed, and the tumor was histologically diagnosed as a collagenous fibroma. A marginal excision of the tumor was carried out. Microscopically, the excised tumor was composed of scattered spindled and stellate-shaped cells in an abundant collagenous stroma (Figure 2). Neither cellular atypia nor mitotic figures were observed.

Conventional cytogenetic analysis revealed the following karyotype: 47,XX,+8[5]/46,XX[15] (Figure 3). The postoperative course was uneventful, and the patient is doing well without local recurrence two months after the surgery.

Discussion

Cytogenetic studies strongly suggest that the 11q12 rearrangement is characteristic of collagenous fibroma, with the presence of an identical t(2;11)(q31;q12) translocation (7-12). It is of interest that the same translocation has been identified in a case of fibroma of tendon sheath (13). Recently, Macchia et al. (12) proposed that the functional outcome of 11q12 rearrangement is a deregulated expression of FOS-like antigen-1 (FOSL1) in collagenous fibroma. In the present study, we recorded the occurrence of trisomy 8 as the sole anomaly in 25% of the analyzed cells, although the primary anomaly may not be recognizable. To the best of our knowledge, this is the first reported case of isolated trisomy 8 in collagenous fibroma.

Trisomy 8 is a non-random aberration in a subgroup of benign fibrous lesions (14). Notably, this numerical chromosomal aberration has been detected at the cytogenetic or molecular cytogenetic level in a significant subset of desmoid-type fibromatoses (14-19), with a similar histological appearance to collagenous fibroma. These findings suggest that trisomy 8 may contribute to aberrant cell proliferation in primary fibrous lesions. On the other hand, trisomy 8 is also a well-recognized secondary abnormality in a variety of mesenchymal neoplasms, such as myxoid liposarcoma (20), clear cell sarcoma (21), and Ewing’s sarcoma (22).

The differential diagnosis of collagenous fibroma includes a number of benign and intermediate soft tissue tumors, such as fibroma of tendon sheath, tenosynovial giant cell tumor, solitary fibrous tumor, and desmoid-type fibromatosis. These tumors can exhibit similar signals on T1- and T2-weighted sequences. The most significant differential diagnosis to be considered is desmoid-type fibromatosis, as in our case. Recently, Yamamoto et al. (23) suggested that the presence of rim enhancement on contrast-enhanced fat-suppressed T1-weighted sequences may be a primary indication of collagenous fibroma. In the present case, we observed a similar enhancement pattern at the periphery of the lesion. On the other hand, desmoid-type fibromatosis would not be as clearly circumscribed as collagenous fibroma and typically exhibits moderate-to-marked enhancement. Prominent low-signal intensity band on all pulse sequences can be seen in desmoid-type fibromatosis (24). The enhanced pattern and the presence of low-signal intensity band may be useful to distinguish between collagenous fibroma and desmoid-type fibromatosis.

In summary, we have reported the first case of collagenous fibroma involving trisomy 8. Further studies are needed to determine the precise role of this trisomy in collagenous fibroma.
Figure 2. Histological finding of collagenous fibroma. The tumor is hypocellular and consists of spindled and stellate-shaped cells in a dense collagenous stroma (hematoxylin and eosin staining, original magnification ×80).

Figure 3. A representative Giemsa-Trypsin-Giemsa (GTG)-banded karyotype of collagenous fibroma showing trisomy 8 as the sole anomaly.
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References


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