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CASE STUDY

Possible role of the collagen type I alpha 1–platelet-derived growth factor beta chain fusion gene in the development of dermatofibrosarcoma protuberans with fibrosarcomatous transformation

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Abstract

Dermatofibrosarcoma protuberans with fibrosarcomatous transformation (DFSP-FS) is a rare variant, with higher rates of recurrence and metastasis than DFSP. Detection of the collagen type I alpha 1 (COL1A1)–platelet-derived growth factor beta chain (PDGFB) fusion gene is useful for the diagnosis of DFSP. In this letter, we report a case of DFSP-FS, focusing on the expression of the COL1A1-PDGFB fusion gene in the lesions. Increased expression of the COL1A1-PDGFB fusion gene may be associated with fibrosarcomatous changes during the pathogenesis of DFSP.

KEYWORDS
collagen type I alpha 1–platelet-derived growth factor beta chain fusion gene, dermatofibrosarcoma protuberans, dermatofibrosarcoma protuberans with fibrosarcomatous transformation

Dermatofibrosarcoma protuberans with fibrosarcomatous transformation (DFSP-FS) is a rare variant, with higher rates of recurrence and metastasis than DFSP.[1] Fibrosarcomatous change was defined as mitotically active cellular areas composed of spindle cells arranged in a clearly fascicular, often herringbone-like growth pattern.[2] Detection of the collagen type I alpha 1 (COL1A1)–platelet-derived growth factor beta chain (PDGFB) fusion gene is useful for the diagnosis of DFSP.[3] In this letter, we report a case of DFSP-FS, focusing on the expression of the COL1A1-PDGFB fusion gene in the lesions.

A 65-year-old man was referred to us because of a reddish tumor on the back. He underwent surgical resection 30 years ago and was diagnosed with DFSP. Local recurrence had not been observed for 15 years after surgery. However, when a recurrent lesion subsequently appeared on the back, he did not visit a hospital. The recurrent lesion proceeded to gradually enlarge over the following 15 years. Upon his first visit to our hospital, a reddish tumor with necrotic tissue was observed on the back (Figure 1A). After the diagnosis of recurrent DFSP via histopathological analysis, the patient underwent tumor resection surgery with a 3 cm margin. Postoperative histopathological findings showed a storiform pattern, partially altered via FS, composed of spindle cells (Figure 1B,C). Immunohistochemical analysis showed that tumor cells expressed cluster of differentiation 34 (CD34; Figure 1D) and Mindbomb E3 ubiquitin-protein ligase 1 (MIB-1; Figure 1E). COL1A1-PDGFB gene fusion was detected via the real-time reverse transcription polymerase chain reaction (Figure 1F). This allowed for the definitive

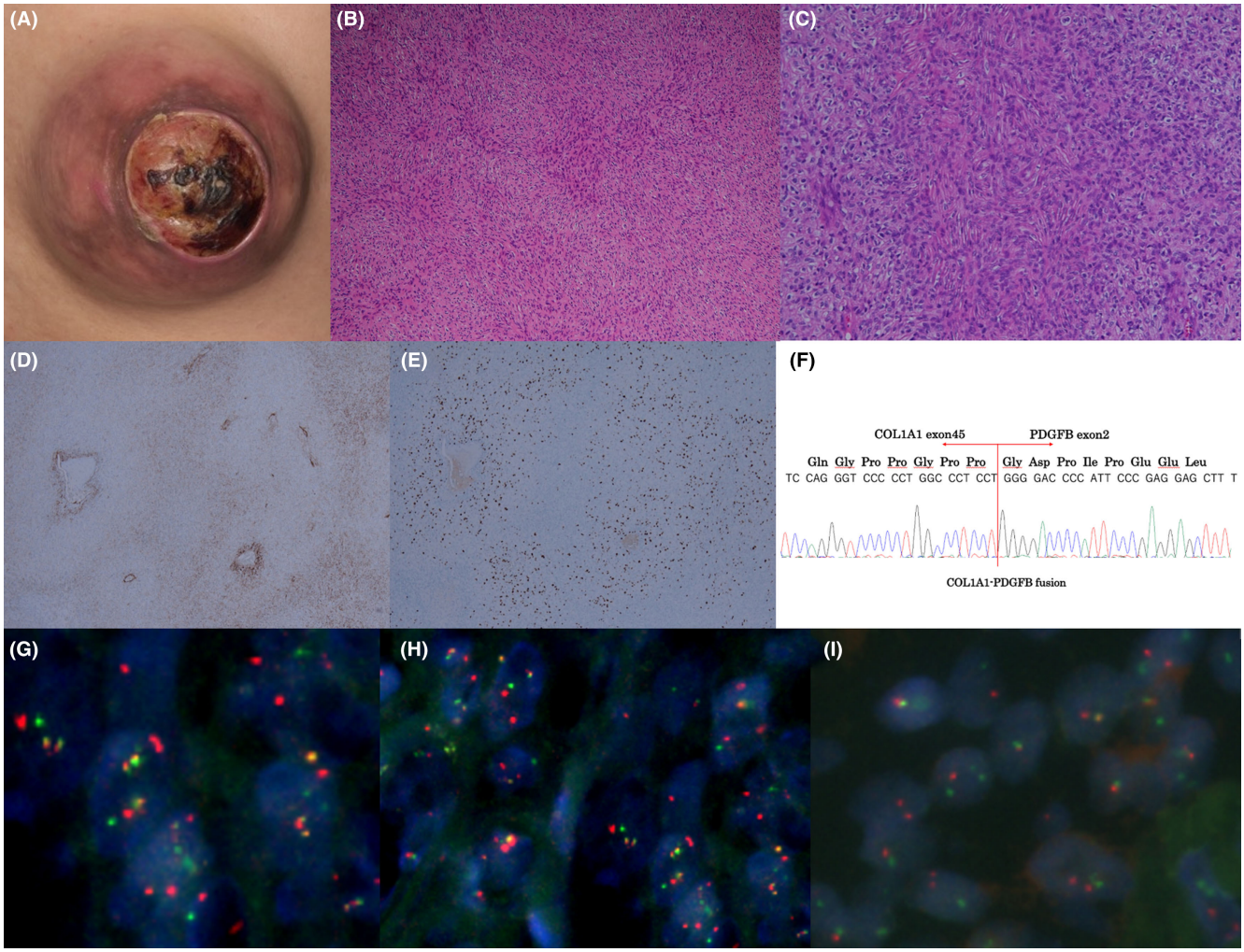

FIGURE 1 (A) Reddish tumor with necrotic tissue on the back. (B) Histopathology shows a storiform pattern, partially altered via fibrosarcomatous transformation (H&E staining, 12.5× magnification). (C) Spindle cell composition of the storiform pattern (200× magnification). (D) Tumor cells expressing cluster of differentiation 34 (40× magnification). (E) Tumor cells expressing Mindbomb E3 ubiquitin-protein ligase 1 (40× magnification). (F) Collagen type I alpha 1 (COL1A1)–platelet-derived growth factor beta chain (PDGFB) gene fusion detected via a real-time reverse transcription polymerase chain reaction. (G) COL1A1-PDGFB fusion genes detected via fluorescence in situ hybridization (red: COL1A1; green: PDGFB). (H) COL1A1-PDGFB fusion genes in the fibrosarcomatous area. (I) COL1A1-PDGFB fusion genes in the non-fibrosarcomatous area.

diagnosis of DFSP-FS. No recurrence has been observed for over a year after surgery.

CD34 was expressed in DFSP areas (Figure 1D). In contrast, MIB-1 was expressed in DFSP-FS areas (Figure 1E), which had a higher MIB-1 labeling index than DFSP areas. These immunohistochemical findings were consistent with those of a previous study.[4]

Dermatofibrosarcoma protuberans is genetically characterized by translocation *t*(17;22)(q22;q13), resulting in the fusion of COL1A1 and PDGFB.[3] The expression of PDGFB prevents the regulation of upstream inhibitory factors and leads to the mass production of COL1A1-PDGFB chimeric mRNA upon initiation of the COL1A1 sequence, a crucial factor in the development of DFSP.[5] However, the function of the COL1A1-PDGFB fusion gene remains to be fully elucidated. In this case, fluorescence in situ hybridization was performed to analyze the expression of the COL1A1-PDGFB fusion gene in the lesions. The COL1A1-PDGFB fusion gene was observed in both DFSP and DFSP-FS areas (Figure 1G). However, the fusion gene was highly expressed in multinucleated, nucleus-deformed, and highly heteromorphic DFSP-FS areas (Figure 1H), but poorly expressed in less heteromorphic DFSP areas (Figure 1I). These results suggest that increased expression of the COL1A1-PDGFB fusion gene may be associated with fibrosarcomatous changes during the pathogenesis of DFSP. However, more case studies are needed to clarify the underlying pathomechanisms.

CONFLICT OF INTEREST

The authors declare no conflicts of interest.

ETHICS STATEMENT

Approval of the research protocol: No human participant was involved in this study.

Informed Consent: The patient has provided informed consent for the publication of the images submitted with this article.
Registry and the Registration No.: N/A.
Animal Studies: N/A.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author, Koji Kamiya, upon reasonable request.

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