Gardner fibroma with localized hypertrichosis without adenomatous polyposis coli gene mutation

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To the Editor: Gardner fibroma (GAF)¹ is usually accompanied by familial adenomatous polyposis (FAP), which is related to mutations in adenomatous polyposis coli (APC).²⁻⁴ Both GAF and FAP present with signs of Gardner syndrome (GS), triad of intestinal polyposis, soft tissue tumor (including GAF and desmoid), and osteomas.²⁻⁴ Here, we presented a man who was diagnosed with sporadic GAF without APC gene mutations. In addition, a tubular adenoma was revealed in his sigmoid. We confirmed the significance of GAF as a powerful sentinel element for gastrointestinal organ and bone problems with or without APC mutations.¹⁴

The 29-year-old Chinese man complained of a painless mass on the right lateral thigh for 5 years. No significant family history was observed. Physical examination [Figure 1A] showed a soft mass with terminal hairs. In serum, the level of neuron-specific enolase was 20.4 (0–16.3) ng/mL. Gray-scale ultrasound analysis showed a superficial, ill-defined hypoechoic nodular area measuring 21 mm × 6 mm. Multiple strips of hypoechogeticity and punctuated hyperechogenicity were observed in the mass. Color power Doppler analysis showed a twinkling artifact in the nodule. Histopathologic examination [Figure 1B] demonstrated the proliferation of haphazardly arranged coarse collagen fibers and an increased number of hair follicles. Immunohistochemically, β-catenin, cyclin-dependent kinases D1 (cyclin-D1), cellular myelocytomatosis (C-myc), cluster of differentiation 34 (CD34), S-100, and fibroblast growth factor 3 were negative. No abnormalities were observed on whole body bone imaging. Intra-abdominal fibroma or desmoid tumors were excluded by abdominal ultrasound. A tubular adenoma was observed in his sigmoid. Peripheral blood was collected and DNA was extracted from the patient and his parents. All 15 exons of the APC gene were amplified by polymerase chain reaction. However, no mutations were identified.

GAF clinically manifests as painless single or multiple subcutaneous masses on the back, paraspinal region, and upper extremities.¹ The diameter of the masses ranged from 0.5 to 2.5 cm. GAF occurs in individuals with or without mutations in APC.²⁻³ Dahl et al.² demonstrated that in general, GAFs with positive APC mutations were too multifocal and too large to be resected. In addition, patients with negative APC testing had a sporadic resectable GAF. GAF is diagnosed histopathologically with formless sheets of thick, haphazardly-arranged collagen bundles with interspersed fibroblasts, and a plaque-like growth pattern with infiltration and entrapment of surrounding structures. Immunohistochemically, β-catenin, cyclin-D1, c-myc, and CD34 reactivity were also noted in GAF.² Over-expression of β-catenin and cyclin-D1 can be caused by an abnormal APC gene.²⁻⁻⁵

Mutations in the APC gene¹⁴ may lead to GS, FAP, or GAF.³ However, in some rare cases, sporadic GAF without APC mutations can develop FAP.² Therefore, GAF may serve as a powerful sentinel element for internal organ and bone problems with or without APC mutations. Taken together, patients with GAF should undergo comprehensive examinations combined with genetic mutation testing. Here, we also highlighted localized hypertrichosis as a special manifestation in our case, which may indicate a relationship between fibrous proliferation and the formation of hair follicles in GAF. Further studies will be required.

Declaration of patient consent

The authors certify that they have obtained all necessary patient consent forms, stating that the patient has consented to the use of his clinical information for

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publication in the journal. The patient understands that his name and initials will not be published, and due efforts will be made to conceal the identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

None

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