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Aggressive fibromatosis of the neck in a patient with Gardner’s syndrome

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Abstract  We report a patient with Gardner’s syndrome who, in addition to a total colectomy, had multiple excisions of desmoid tumors in both thighs. He presented with left-sided neck swelling and pain. MRI was highly suggestive of desmoid tumors in multiple neck muscles. To our knowledge this is the first description of diffuse fibromatosis of the neck in association with Gardner’s syndrome.

Keywords  Gardner’s syndrome · Aggressive fibromatosis · Desmoid tumors · Head and neck

Introduction

Gardner’s syndrome is familial gastrointestinal polyposis, with autosomal dominant inheritance. The colon is the most common site of involvement, but other parts of the gastrointestinal tract can be involved. Extra-intestinal manifestations include soft-tissue tumors such as sebaceous cysts, fibromas, lipomas, and desmoid tumors, and osteomas of the mandible, skull and long bones.

Case report

A 35-year-old man had been known for some years to have Gardner’s syndrome. His history included a proctocolectomy and multiple excisions of pathologically proven desmoid tumors in both thighs, with postexcision radiation therapy. He presented again with generalized neck pain and swelling on the left side. Contrast-enhanced MRI demonstrated abnormal prominence of several muscles on the left side of the neck: the longus colli and longus capitis were significantly larger than on the right, as were the semispinalis capitis and levator scapulae. The left sternocleidomastoid showed only signal abnormalities, without enlargement. Most of the enlarged muscles gave lower signal than normal on T1-weighted images (Fig. 2) the signal was also predominantly lower than in normal muscles. The left longus capitis and longus colli muscles caused slight distortion of the hypopharynx and larynx and anterior displacement of the carotid artery. Coronal T1-weighted images (Fig. 3) confirmed the prominence of the left longus colli and capitis muscles. There was no significant contrast enhancement (Fig. 4). These MRI findings were felt to indicate desmoid tumors of multiple neck muscles.

Discussion

Gardner’s syndrome is one of the familial polyposis syndromes, inherited in an autosomal dominant pattern with nearly 100% penetrance. The gene responsible has been localized to the long arm of chromosome 5 [1]. The syndrome is characterized by extensive adenomatous polyps in the colon; 100 or more polyps should be identified to establish the diagnosis. The potential for malignant transformation is virtually 100% in untreated cases [1, 2]. Extracolonic manifestations include gastric and small intestinal polyps, perianpillary and biliary involvement, mesenteric fibromatosis, skin and eye involvement, thyroid carcinoma, skeletal and dental manifestations and central nervous system tumors [2]. Desmoid tumors are muscu-
loaponeurotic tumors belonging to a family of fibromatoses characterized by a proliferation of benign fibrous tissue composed of uniform, elongated, fusiform or spindle-shaped cells surrounded and separated by abundant collagen [3]. In Gardner’s syndrome these can be in the abdomen (mesenteric, retroperitoneal) or extra-abdominal (muscoloaponeurotic), usually in the limbs [3]. To our knowledge, this is the first case of presumed multiple desmoid tumors, or aggressive fibromatosis, of the neck in a patient with Gardner’s syndrome.

The characteristic MRI features of desmoid tumors are of a soft-tissue mass with heterogeneous signal intensity and poor margination [4]. They have areas of low signal relative to normal muscle on both T1- and T2-weighted images, corresponding to the fibrous component of the desmoid. The signal intensity of the nonfibrous portion has been reported as higher than that of muscle but less than that of fat on T2-weighted images, in contrast to malignant soft tissue tumors which give higher signal than fat, high signal correlating with less collagenization and high cellularity, and low signal with relative acellularity and abundant collagen [5, 6]. On T1-weighted imaging, both low and isointense signal relative to skeletal muscle has been described [7, 8, 9, 10, 11, 12]. High signal on T1-weighted imaging may be secondary to blood or fat within the tumor [13]. The contrast enhancement pattern ranges from none, through inhomogeneous, to intense homogenous enhancement [7, 8, 11]. In one study, neither the low-signal zones on T1- and T2-weighted images nor the type of contrast enhancement had any significant relationship to the biological behavior of the tumor [7], but in another, increased signal on T2-weighted images was associated with marked growth on follow-up [11].