A young man was found to have desmoid tumors involving multiple sites. This tumor was from the abdominal wall. In addition to multiple desmoid tumors, numerous polyps were identified in the colon, stomach, and duodenum.

A mutation in what gene is likely involved in a patient with desmoid tumors and numerous gastric, duodenal, and colon polyps?

a. TP53
b. RET
c. APC
d. BRAF

(see page 194 for answer)
Answer: c. APC

The APC (adenomatous polyposis coli; OMIM 611731) gene is a tumor suppressor gene on chromosome 5q21-22. It is associated with familial adenomatous polyposis, which is an autosomal dominant condition associated with numerous colorectal and gastrointestinal adenomas and can be associated with prominent extraintestinal manifestations including desmoid tumors, thyroid tumors such as cribriform morular thyroid carcinoma, osteomas of bone, congenital hypertrophy of retinal pigment epithelium, and cutaneous lesions.1,2 Other variants of the disease exist with other tumors (eg, Turcot syndrome and central nervous system tumors). Desmoid tumors occur in approximately 10% to 15% of individuals with familial adenomatous polyposis. Desmoid tumors may occur more frequently after surgery.3 The adenomas in familial adenomatous polyposis have a histology similar to sporadic tubular adenomas.
REFERENCES

