Condition: Familial adenomatous polyposis

Inheritance:
Autosomal Dominant**.**

Genetic etiology:
Mutation in APC gene.

Frequency:
1/30,000.

Clinical features:
The hallmark of familial adenomatosis polyposis if the development of multiple polyps in the colon, as well as elsewhere in the GI tract, including the stomach and small intestine. The polyps are precancerous, but there is a very high risk of one or more eventually becoming malignant. There may be associated congenital hypertrophy of the retinal pigment epithelium (CHRPE) and, in some patients, osteomas and soft tissue tumors (Gardner syndrome) or tumors of the central nervous system (Turcot syndrome). Attenuated FAP is the occurrence of a smaller number of colonic polyps, but still an increased risk of cancer.

Management:
Screening for gastrointestinal polyps; colectomy when polyps begin to emerge. Surveillance for other tumor types.

Genetic counseling:
Transmitted as autosomal dominant trait with high penetrance; genetic testing for APC mutation is available. If the mutation in a family cannot be detected, individuals at risk should be offered program of regular screening for cancer. There is an autosomal recessive form of polyposis associated with biallelic mutation of the *myh* gene.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.