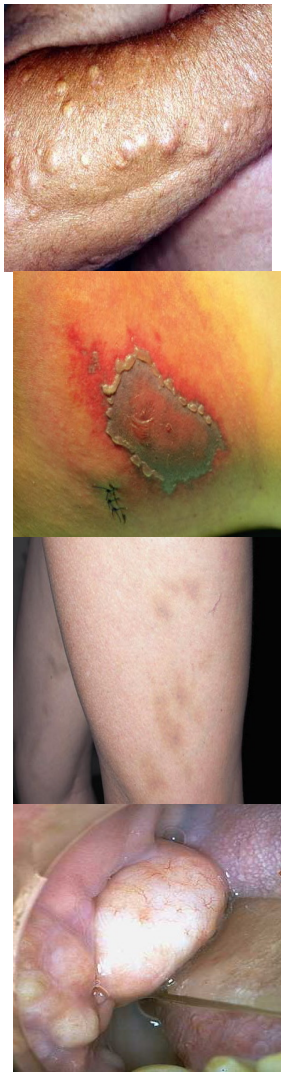

Gardner Syndrome

Gardner syndrome, a variant of familial adenomatous polyposis (FAP),[1] is an autosomal dominant disease characterized by GI polyps, multiple osteomas, and skin and soft tissue tumors. Cutaneous findings

[2]

of Gardner syndrome include epidermoid cysts, desmoid tumors, and other benign tumors. Polyps have a 100% risk of undergoing malignant transformation; consequently, early identification of Gardner syndrome is critical.
[3]

Gardner syndrome is genetically linked to band 5q21, the adenomatous polyposis coli locus.[4] F AP and Gardner syndrome are believed to be variants of the same condition. The wider spectrum of abnormalities found in Gardner syndrome may represent variable penetrance of a common genetic mutation.

One person per million population is diagnosed with Gardner syndrome. The incidence of FAP is 1 case per 8000 people. The most common cutaneous finding in patients with Gardner syndrome is epidermoid cysts (50-65%).

Unless surgical transection is performed, GI polyps may progress to malignancy in almost 100% of Gardner syndrome patients (rates vary from 58-100% in studies).

Although colonic polyps begin to form in puberty, the average age at Gardner syndrome diagnosis is 22 years. Osteoma formation precedes polyposis. Usually, progression to malignancy is observed in patients aged 30-50 years. The average age by which malignancy is diagnosed is 39.2 years.

- Many skin findings of Gardner syndrome are evident on full body examination; however, the patient's history of the age at onset and whether lesions are present in family members is important.
- Cysts in Gardner syndrome patients are usually asymptomatic, but they may be pruritic and/or inflamed.
- More than half the patients with Gardner syndrome have dental anomalies.[5] Previously undiagnosed Gardner syndrome may be detected when the patient is evaluated for multiple impacted and unerupted teeth.

A full body skin examination for skin tumors and epidermal inclusion cysts is necessary in Gardner syndrome.

- Several factors differentiate cutaneous cysts associated with Gardner syndrome from ordinary cysts.
- Epidermoid cysts of Gardner syndrome occur at an earlier age (around puberty) than ordinary cysts and in less common locations, such as the face, the scalp, and the extremities.

- Gardner syndrome cysts tend to be multiple and are present in the multiple form in 50-65% of patients.
- Similar to ordinary epidermal inclusion cysts, cysts in Gardner syndrome are usually asymptomatic; however, they may be pruritic and/or inflamed, and they may rupture.

- Other skin signs in Gardner syndrome include the following:
- Fibromas
- Lipomas
- Leiomyomas
- Neurofibromas
- Pigmented skin lesions

- Noncutaneous features of Gardner syndrome include the following:
- Desmoid tumors occur as swelling in the anterior abdominal wall and are often preceded by surgical trauma. The incidence of desmoid tumors in FAP is 8.9%.
- Osteomas are required to make the diagnosis of Gardner syndrome. The mandible is the most common location. They may be widespread in the jaw. [6] However, osteomas may occur in the skull and the long bones. Osteomas precede clinical and radiologic evidence of colonic polyposis; therefore, they may be sensitive markers for the disease.

- Colonic adenomatous polyps have a 100% risk of transformation to colonic adenocarcinoma.
- Multifocal pigmented lesions of the fundus are seen in 80% of patients and may present shortly after birth. These lesions can be the first marker of disease.
- Dental abnormalities (eg, unerupted teeth, supernumerary teeth) may occur.

- Other associated neoplasms in Gardner syndrome include the following:
- Periampullary carcinoma (ampulla of Vater; reported in 12% of patients with FAP, usually after colectomy)
- CNS tumors, such as medulloblastoma, glioblastoma, and craniopharyngioma (found in FAP subgroup in Turcot syndrome)
- Thyroid carcinoma (especially in female patients)
- Osteosarcoma
- Chondrosarcoma
- Hepatoblastoma
- Liposarcoma

The cause of Gardner syndrome is genetic, with autosomal dominant inheritance.

Treatment

Treatment of the cutaneous manifestations of Gardner syndrome depends on the symptomatic or cosmetic nature and the location of the cysts. Treatment is similar to that used for ordinary cysts and involves excision or use of intralesional steroids if the cysts are inflamed.

- Colectomy is recommended for Gardner syndrome patients if 30 or more polyps are detected on colonoscopy or if biopsy results reveal dysplasia or malignant degeneration.

- Preserving the rectum results in a 25-59% chance of rectal carcinoma occurring in Gardner syndrome patients; therefore, rectal mucosal resection is recommended.
- Generally, cutaneous findings do not require treatment.
- Osteomas may require excision if they are severely deforming or if they interfere with function.

Also see eMedicine's General Surgery article Gardner Syndrome