Gardner Syndrome

Gardner syndrome (GS) is an autosomal dominant form of familial adenomatous polyposis that results from a mutation in the APC tumor suppressor gene and exhibits extracolonic manifestations. These manifestations include osteomas, desmoid tumors, cutaneous lesions, dental abnormalities, congenital hypertrophy of retinal pigment epithelium (CHRPE), adrenal adenomas and nasal angiofibromas as well as an association with malignancies of the thyroid, duodenum/periampullary region, pancreas and liver - in the form of hepatoblastoma developed in childhood.



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Pathophysiology

Familial Adenomatous Polyposis (FAP) + Extraintestinal Benign Tumors

Fat-gardener with Benign-bunny and Tumor-masses-growing-on-ground

Gardner syndrome (GS) is a variant of familial adenomatous polyposis (FAP), which means patients will have multiple colonic polyps. However, unlike FAP, GS also include extraintestinal benign tumors such as osteomas and fibromas.

Autosomal Dominant

Domino-road

This disease demonstrates an autosomal dominant inheritance pattern.

APC Gene Mutation

Apple-PC

Adenomatous polyposis coli, or APC, is a tumor suppressor gene located on the long arm of chromosome 5. When mutated, it may result in Gardner syndrome (GS). This is also the gene mutation responsible for familial adenomatous polyposis (FAP).

Clinical Findings

Desmoid Tumors

Desk-droid with Tumor-guy

Gardner syndrome (GS) predisposes patients to desmoid tumors, which are fibrous lesions that can occur nearly anywhere in the body, but are most commonly found in the abdomen where they begin as plaque-like abnormalities that progress to mesenteric fibromatosis and eventually to desmoid tumors. While these growths are benign, they can grow to cause significant damage to adjacent structures like blood vessels, bone, nerves and bowel.

Nasal Angiofibromas

Angel-fabio with Big-nose

Nasal angiofibromas are benign, vascular growths that have been described in some patients. These most commonly occur in adolescent males and are described as vascular, smooth, submucosal masses located in the posterior nasal cavity. They may result in recurrent and severe epistaxis, unilateral nasal obstruction or invasion of adjacent structures.

Osteomas

Ostrich with Tumor-guy

Osteomas are benign bone growths that can occur anywhere in the body, but most commonly present on the mandible, maxilla, skull and long bones. Panoramic dental radiographs often reveal opaque lesions in the mandible of these patients, even when these lesions are not present on physical exam.

Congenital Hypertrophy of Retinal Pigment Epithelium

Pregnant-woman and Hiker-with-trophy and Pig-with-red-tins-on-eyes-and-E-pick-on-head

Congenital hypertrophy of retinal pigment epithelium (CHRPE) occurs when patches of retinal pigment epithelium are thicker than normal and present on ophthalmological exam as multiple hyperpigmented lesions resembling “bear-tracks”. Despite a very remote malignancy risk, these fundus symptoms

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typically do not cause symptoms or require treatment, but are important clinical findings to note in Gardner syndrome (GS) given their 90% sensitivity and early presentation in the progression of the syndrome.

Dental Abnormalities

Abnormal-tooth and Numerous-teeth-on-ground

Patients with GS may exhibit dental abnormalities in the form of supernumerary and unerupted teeth, odontomas, or dentigerous cysts.

Cutaneous Lesions

Skin-suit with Leeches

Cutaneous lesions associated with Gardner syndrome (GS) include epidermoid cysts, fibromas, lipomas and pilomatriconas. These epidermoid cysts are primarily found on the legs, face, scalp and arms while the fibromas occur on the scalp, shoulders, arms and back. None of the associated cutaneous lesions are malignant.

Adrenal Adenomas

Adrenal-gland-roof and Add-gnome

Adrenal adenomas, the most common form of adrenal tumor, are benign growths that can occur in Gardner syndrome, and are often found incidentally on radiographic imaging when looking for some other abdominal pathology.

Associations

Thyroid Cancer

Thigh-droid with Tumor-guy

Patients have an 8-fold increased risk of developing thyroid cancer over the general population. This is frequently in the form of papillary thyroid carcinoma.

Duodenal / Periampullary Cancer

Dodo-bird-wearing-denim with Tumor-guy on Pear-amp

Patients also have an increased risk of malignancy in the area surrounding the ampulla of Vater.

Pancreatic Cancer

Pancreas with Tumor-guy

Pancreatic cancer is 4 times more likely in these patients.

Hepatoblastoma

Liver with Tumor-guy and gun-blast

GS patients have an 800-fold increased risk of developing hepatoblastoma. This commonly occurs in the first 5 years of life.

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