Patient Report & Review of Radiologic Features

GARDNER’S SYNDROME
Patient Report: Ms. CB

- Chief Complaint
  - Weight loss >13 lbs in one month

- HPI
  - 38 year old woman with Gardner's Syndrome (diagnosed at age 23) and s/p colectomy (1985) and multiple other small bowel resections secondary to desmoid tumors, now with weight loss.

  - Today on monthly clinic visit, patient weighed in at 93lbs, down 13lbs from her prior visit. She tries to drink 2-4 cans of Ensure daily but is often unable to meet her goal due to early satiety. She also endures abdominal pain, n/v, and increased abdominal girth.
Ms. CB (cont’d)

- **PMH**
  - Gardner's syndrome,
    - s/p colectomy,
    - s/p multiple small bowel resections
    - Small bowel adenocarcinoma
    - s/p dermoid cyst removal (Left ovary)
  - DVT, PE 2001

- **Family History**
  - Father & 6/8 siblings with Gardner's syndrome.
  - Father died at age 42 from polyp blocking pancreatic duct.
  - Sister diagnosed with Gardner’s when attempting to enter marines, led to genetic testing of whole family
What is Gardner’s Syndrome?
Background

In the early 1950s, Dr. Eldon Gardner described a family displaying not only the intestinal signs typical of familial adenomatous polyposis (FAP), but also a number of extra-intestinal lesions such as osteomas, epidermal cysts, desmoid tumors, etc. This combination of FAP-like colonic adenomatosis and extra-colonic lesions came to be known as Gardner’s Syndrome.
Intestinal Lesions

Familial Adenomatous Polyposis

- characterized by thousands of colonic adenomatous polyps extending from the stomach, duodenum, and colon.
- Incidence is approximately 1 case in 8000
- Chance of malignant transformation to colon cancer is 100% without surgical intervention.
It is now known that an autosomal dominant genetic disorder mutation in the adenomatous polyposis coli (APC) gene, the same gene responsible for FAP, is responsible for Gardner’s Syndrome (GS) [1].

The extraintestinal signs of GS have been found in 20% of patients with FAP [2].

Gardner’s syndrome is generally considered a variant of FAP, possibly representing variable penetrance.
ExtraIntestinal Lesions

- Osteomas and dental abnormalities
- Desmoid tumors
- Cutaneous lesions
- Congenital hypertrophy of the retinal pigment epithelium
- Adrenal adenomas
Osteomas & Dental Abnormalities

Skeletal abnormalities are seen in approximately 90% of patients with GS [4]

Osteomas

- most common abnormality. Osteomas precede clinical and radiologic evidence of colonic polyposis; therefore, they may be sensitive markers for the disease.

- Sites include:
  - outer cortex of the skull,
  - paranasal sinuses and the mandible [4]
  - May occur in long bones, but less frequently

Dental abnormalities

- Supernumerary teeth, compound odontomas and/or impacted teeth were seen in 30% of the patients with this disease
Dental panoramic radiography may be useful for early detection of GS, as osteomas (arrows) & dental abnormalities not apparent on physical examination can be detected on routine radiographs in up to 90 percent of FAP patients [5].
Osteoma: CT Detection (Companion Pt #2)

However, given the bidimensional imaging quality and superimposition of bony structures seen with panradiography, CT is superior for localizing and assessing the extent of tumor mass.[6]

Shown here:

**Plain films**: well-defined osteo-sclerotic lesion, with lobulated margins involving the frontal & ethmoidal air sinuses

**CT Scan**: intra-cranial & extra dural extension of the osteoma with compression of the brain parenchyma anteriorly. Incidentally, the brain parenchyma was normal & did not show any focal area of abnormal attenuation.

Dr. Anand Gaikwad [http://www.kem.edu/dept/radiology/case51_03.htm][7]
Desmoid tumors - General Info

- The term desmoid is derived from the Greek word *desmos*, which means “tendonlike.”

- Desmoid tumors often appear as infiltrative, usually well-differentiated, poorly circumscribed fibrous neoplasms originating from the musculoaponeurotic structures. They are also known as “aggressive fibromatosis,” a tribute to their aggressive local behavior. However, they are considered benign as they do not metastasize.

- In the general population, desmoid tumors arise most commonly from the rectus abdominis muscle in postpartum women or from abdominal surgery scars [8].
Desmoid tumor - Microscopic

Shown here: a poorly circumscribed proliferation of spindle cells (fibroblast-like) of uniform appearance and separated from one another by dense bands of collagen. The lesion (*) has infiltrated adipose tissue (arrow) and skeletal muscle (M). The tumors tend to infiltrate adjacent muscle bundles, frequently entrapping them and causing their degeneration.

Desmoid Tumor- Macroscopic

Tumors range from 5-20 cm in diameter and have a firm, gritty texture. The cut surface reveals a glistening white, trabeculated tissue.

The tumors lack a true capsule.

Desmoid Tumor- Differential Diagnosis

There are no specific imaging features to distinguish desmoid tumors from other solid masses. Thus, the diagnosis of desmoid tumor should be considered in patients with an abdominal mass, a history of abdominal surgery or injury, or Gardner syndrome.

<table>
<thead>
<tr>
<th>Differential Diagnosis of Desmoid Tumors</th>
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<tbody>
<tr>
<td><strong>Malignant</strong></td>
</tr>
<tr>
<td>Fibrosarcoma, rhabdomyosarcoma</td>
</tr>
<tr>
<td>synoviosarcoma, liposarcoma, fibrous</td>
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<tr>
<td>histiocytoma, lymphoma, and metastases</td>
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<tr>
<td><strong>Benign</strong></td>
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<tr>
<td>Neurofibroma, neuroma, and leiomyomas</td>
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<tr>
<td><strong>Hematoma</strong></td>
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<tr>
<td>Rectus sheath, chest wall, mesentery,</td>
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<tr>
<td>retroperitoneum</td>
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Adapted from Casillas, J. RadioGraphics 1991 11: 959-968
Desmoid Tumors – Gardner’s Syndrome

- Peak incidence of desmoid tumor in GS is between 28 and 31 years [1]
- About ½ of abdominal desmoids occur intra-abdominally while the other ½ are found in tissues of the abdominal wall
- Mesentenic desmoids tend to develop 1 or 2 years after resection of the intestinal tract. They may even be accompanied by an anterior abdominal wall tumor arising from the surgical scar.
- However, a few have been noted to arise before surgery and even before the onset of polyposis [1].
- Clinically, desmoids may be asymptomatic or may cause pain or intestinal obstruction as a result of impingement.
Desmoid Tumor - Imaging

**Ultrasound**
- variable echogenicity, with smooth, well-defined margins.

**CT**
- On contrast-enhanced scans, the tumors are generally high attenuation (relative to muscle) & may have well-defined margins.

**MRI**
- T1: low signal intensity relative to muscle and
- T2: variable signal intensity on T2-weight
Enhancing ST density material extends diffusely along the intra-abdominal mesentery, tethering the SB loops. Within the right anterior abdominal wall, there is an 8.6 x 4.2 cm heterogeneously-enhancing ST mass measuring 8.6 x 4.2 cm. The proximal loops of SB are dilated, with multiple air-fluid levels.
(a) Axial T2-weighted image shows a large heterogeneous mass (arrow) in the left abdominal wall containing regions of intermediate to low signal intensity. 

(b) Sagittal gadolinium-enhanced T1-weighted image shows an enhancing tumor (arrow) that involves fascial layers of the left rectus abdominis muscle.
The main morbidity & mortality of desmoid tumors is due to their ability to engulf & eventually strangle blood vessels, nerves, ureters, & small bowel. Mortality is as high as 10 to 50 percent in patients who have a desmoid tumor [1]. However, progression is often gradual and the 10 year survival rate is 63 % [1].
EXTRA-COLONIC MALIGNANCIES

- Thyroid (12 percent)
- Duodenal and periampullary (3 to 5 percent)
- Pancreatic (2 percent)
- Gastric (0.6 percent)
- Central nervous system (<1 percent)
- Hepatoblastoma (1.6 percent)
- Small bowel distal to the duodenum
- Adrenal
Thyroid cancers

- The mean age of diagnosis is 33 years [1].
- The thyroid should probably be subject to physical examination and ultrasound annually, starting at age 10 to 12 years [1].

Top panel: A sonogram of the left lobe of the thyroid gland that shows a hypoechoic nodule that is surrounded by a "halo."
Lower panel: Doppler image shows that the "halo" is vascular.
N: nodule; L: thyroid lobe

Blum, M. http://uptodateonline.com/online/content/topic.do?topicKey=thyroid/22414
The follow-up and management of patients with Gardner's syndrome requires a collaboration of gastroenterologists, general surgeons, oral surgeons, radiologists, endocrinologists, neurologists, ophthalmologists, and dermatologists. Radiologists are in a unique position to initially diagnose this disorder.
Acknowledgements

- Jay Catena, MD
References

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