

A Young Man who Complained Nausea and Headache

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1. Case presentation

A young man was admitted to the hospital to evaluate multiple polyps in gastrointestinal tract which was found during evaluation after craniectomy for craniopharyngioma in left posterior fossa. In his family, his grandfather died from colon cancer in young age and his father also died young but suddenly because of unknown cause. He had a mother and a younger brother, but had lost contact with them more than a decade ago. He didn't complain any symptom, and physical examination was unremarkable. Laboratory results were normal.

In esophagogastroduodenoscopy, numerous polyps were found on body and fundus of stomach and multiple biopsies were done. Multiple flat lesions were also found on duodenum and biopsies were taken. In colonoscopy, multiple polyps were also found from ileocecal valve and cecum to proximal rectum area and a 1.5 cm-sized fungating mass was found in proximal rectum, and biopsy was also performed. The biopsy results showed fundic gland polyps in stomach, a tubular adenoma with low grade dysplasia in duodenum, and adenocarcinoma, well differentiated in rectum.

2. Diagnosis

Genetic test showed frameshift mutation in APC and nonfunctioning adrenal adenoma, dental osteoma, fibromatosis were found in extracolonic manifestation evaluation.

3. Therapy and Clinical course

Total colectomy with IPAA was performed for early rectal adenocarcinoma invading SM2 level and Brain RT for remnant craniopharyngioma. However, 4 years later, Multiple masses were found in liver and confirmed to be adenocarcinoma metastasized from colon.

4. Conclusion

It's a rare case of Gardner syndrome who had an early colon cancer, completely resected but recurred as liver metastasis.

Key words: FAP, Gardner syndrome

REFERENCE

1. Gardner's syndrome (familial adenomatous polyposis): A cilia-related disorder; Lancet Oncol. 2009 Jul;10(7):727-35.

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CASE

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- **Chief complaints**

- Abnormal findings in endoscopy

- **Present illness**

- Dizziness, Headache로 타원에서 crano-pharyngioma in left posterior

fossa 진단받고 craniectomy 시행 후 w/u 중 내시경 검사상 GI tract에

수 백개의 polyp 관찰되어 further evaluation 위해 내원 [2013.4.8]

- **Past history**

- HTN / DM / Tb / Hepatitis (-/-/-/)
- Admission / Op hx (+/+)
 - : Retromastoid suboccipital craniectomy [‘13.2]
- Alcohol / Smoking (-/-)
- Medication (-)
- Family hx. (+)
 - » 할아버지 젊을 때 대장암으로 사망
 - » 아버지 젊을 때 급사, 원인 불명확
 - » 어머니 어릴 때 이후 연락 두절
 - » 남동생 1명: 연락 두절

- **Review of system**

Fever / Chilling(-/-)

Headache / Dizziness(-/-)

Anorexia/Vomiting (-/-)

- **Physical examination**

Clear Breathing sound w/o crackle, wheezing

Abdomen Td/RTd (-/-) Pitting edema (-/-)

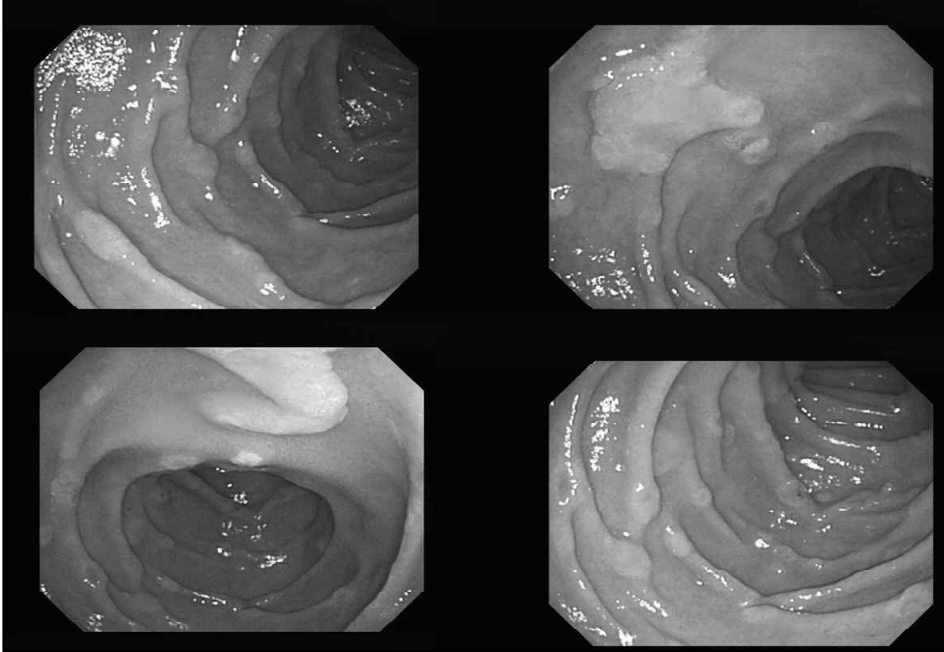
• **Initial Lab findings**

	Hb 14.7 g/dL - WBC 7400 /uL - PLT 169,000 /uL
	Na 142 - K 4.1 - Cl 108 mmol/L
	BUN / Cr 9.9 / 1.01 mg/dL ESR / CRP mm/hr 5 / 0.54 mg/L TSH / ft4 1.76 uIU/mL / 1.17 ng/dL
	Protein / Albumin 6.9 / 4.6 g/dL AST / ALT 19 / 21 IU/L ALP 84 IU/L
	PT 104 %
	CEA 1.2 ng/mL

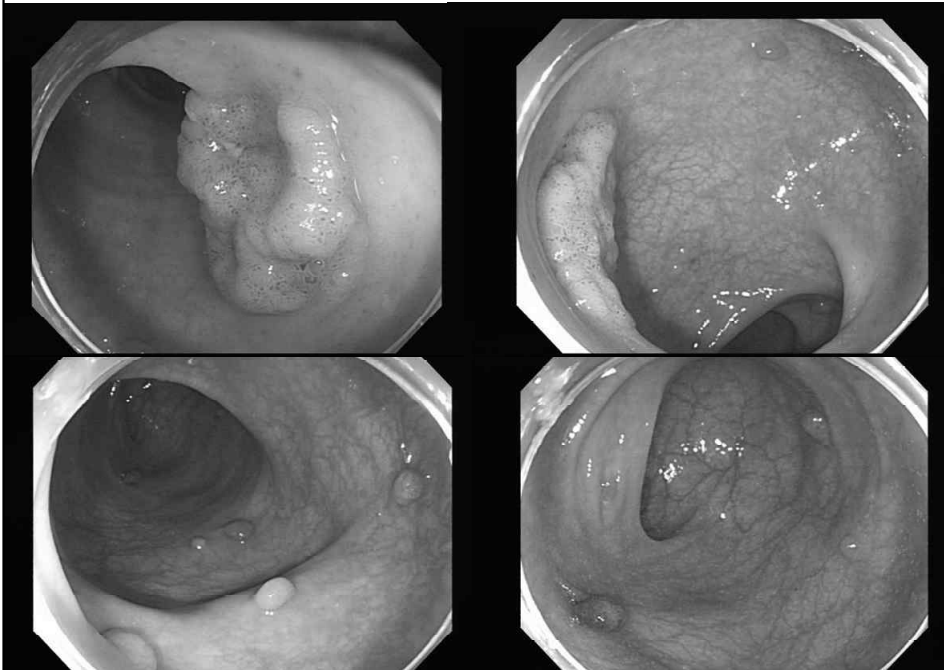
EGD [2013.04.09]



Duodenoscopy ['13.04.09]



Colonoscopy ['13.04.09]



- **Biopsy**

- Stomach, body and fundus, endoscopic biopsy;
 - Fundic gland polyp with no *H. pylori*
- Duodenum, endoscopic biopsy;
 - Tubular adenoma with low grade dysplasia
- Colon, sigmoid, colonoscopic biopsy;
 - Adenocarcinoma, well differentiated in tubular adenoma

- **Positive finding**

- Family hx. of colon cancer
- Brain craniopharyngioma
- Gastric fundic gland polyps
- Colonic polyposis
- Colon adenocarcinoma

- **Impression**

- r/o FAP
- r/o Gardner syndrome
- Colon cancer

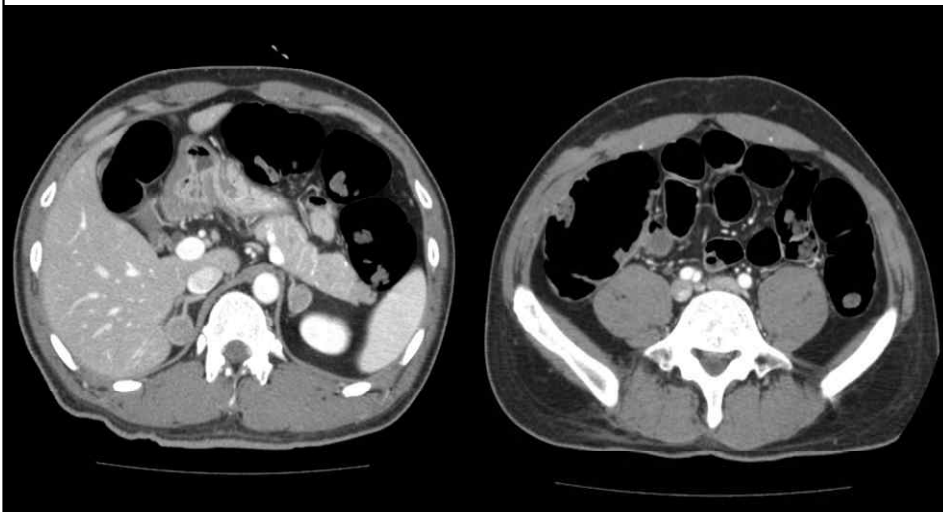
- **Evaluation plan**

- Genetic testing
- Extracolonic manifestation evaluation regarding Gardner syndrome
 - CT scan
 - Ocular, dental evaluation

- **Treatment plan**

- Total colectomy c ileal pouch anal anastomosis
[‘13.04.23]
- Brain RT d/t remnant craniopharyngioma

Abdomen CT [‘13.4.9]



- **Hormone study for adrenal adenoma**

	Cortisol 1.35 ug/dL ACTH 10.1 pg/mL (Basal)
	Epinephrine 8.9 pg/mL Norepinephrine 60.8 pg/mL
	Renin 2.31 ng/mL/hr Aldosterone 3.2 pg/mL

- **Genetic testing**

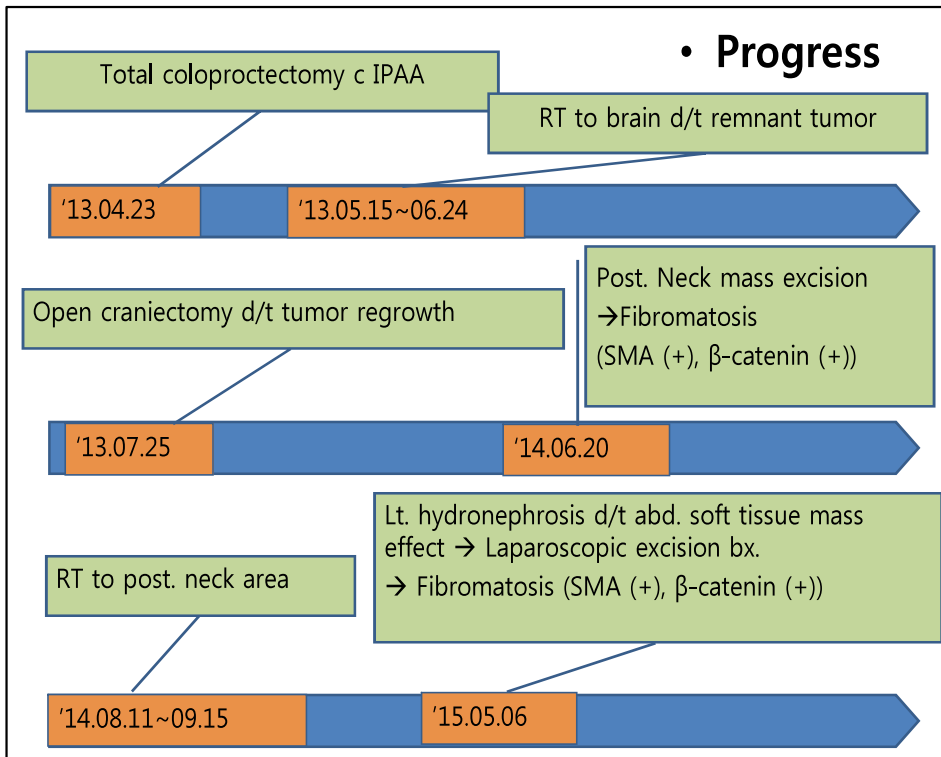
	MLH1 Gene mutation : Not detected
	Frameshift mutation in APC (c.4541_4547delinsTTTCC, p.Pro1514Leufs*18, het)

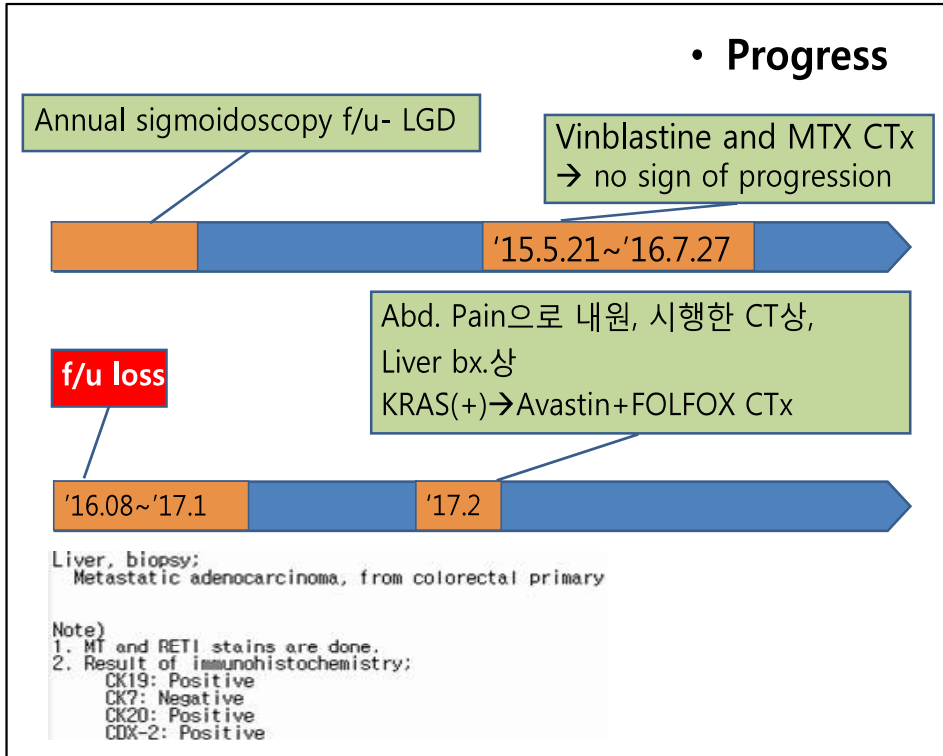
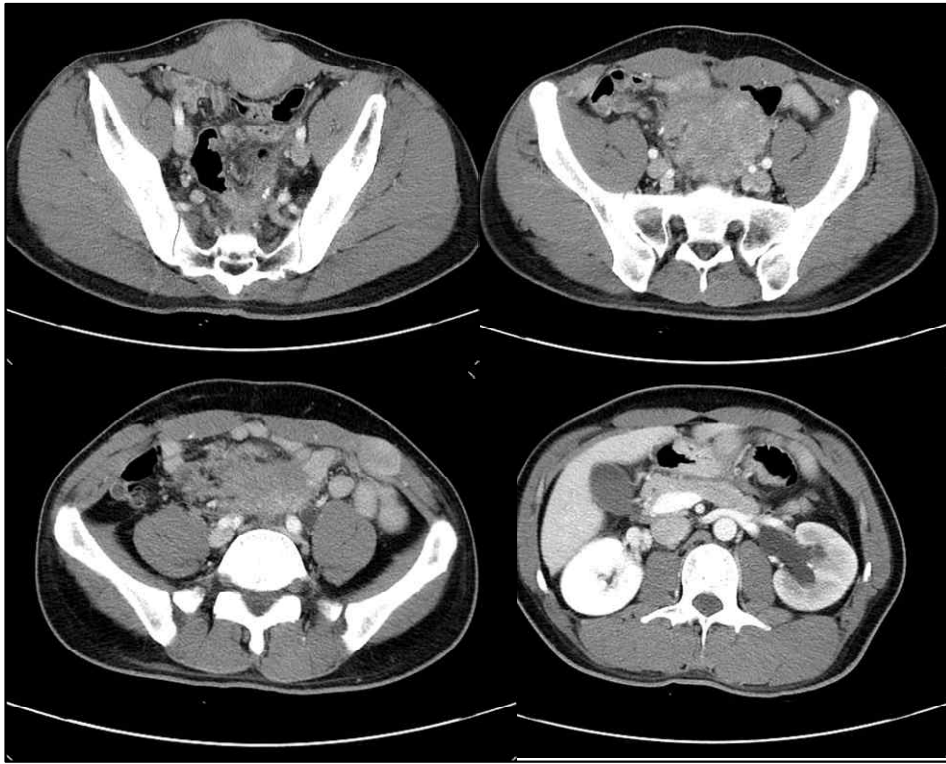
Panorama [‘13.04.09]



- **Positive finding**
 - Family hx. of colon cancer
 - Brain craniopharyngioma
 - Gastric fundic gland polyps
 - Colonic polyposis
 - Colon adenocarcinoma
 - +
 - Genetic test positive for FAP
 - Nonfunctioning adrenal adenoma
 - Dental osteoma

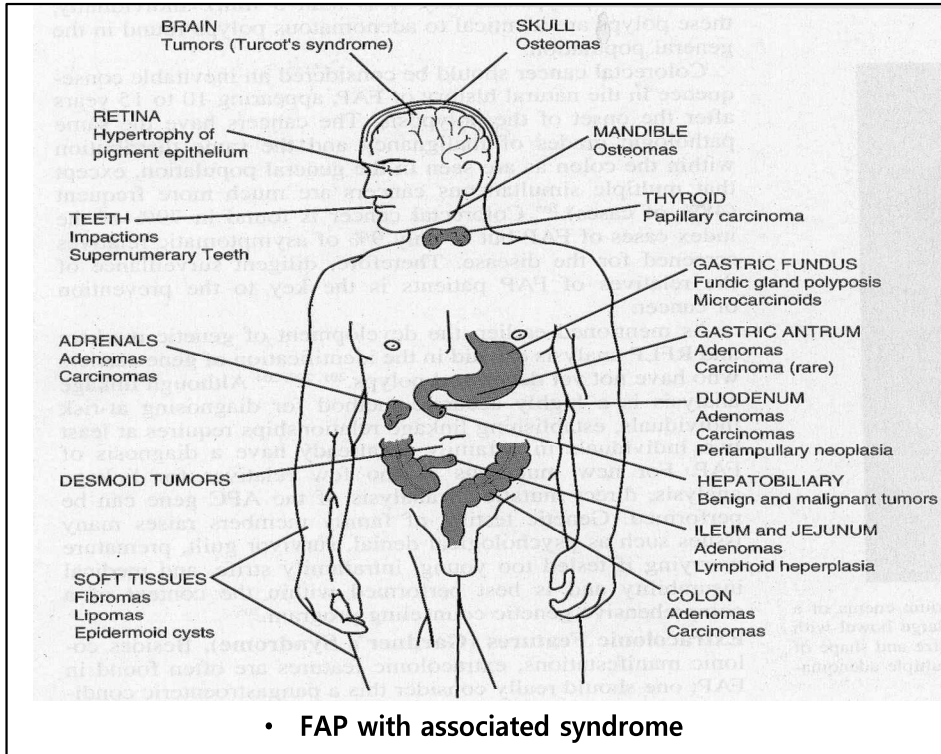
- **Diagnosis**
 - Gardner syndrome





Classification of polyposis syndrome

- Inherited adenomatous polyposis syndrome
 - Classic FAP
 - Gardner’s variant
 - Turcot’s variant
- Familial hamartomatous polyposis syndrome
 - Peutz-Jeghers syndrome
 - Juvenile polyposis
- Non-familial multiple polyposis syndrome
 - Cronkhite-Canada syndrome
 - Lymphoid polyposis



Gardner's syndrome

- Familial adenomatous polyposis
- Extraintestinal manifestation
 - Bone
 - Osteoma of mandible(90%), skull, and long bones
 - Exostoses
 - Dental abnormalities (Supernumerary teeth, impacted teeth)
 - Congenital hypertrophy of the retinal pigmented epithelium (CHRPE)
 - Mesenteric fibromatosis (Desmoid tumors)

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