# 위장관 용종증 환자의 위내시경 소견

성균관대학교 의과대학 내과 이준행

# Hereditable AD GI polyposis syndromes

- Adenomatous polyp
  - FAP
  - Gardner's syndrome
  - Turcot's syndrome
  - Nonpolyposis syndrome (Lynch syndrome)
- Harmartomatous polyp
  - Peutz-Jegher syndrome
  - Juvenile polyposis
  - Cowden syndrome

# FAP

성균관대학교 의과대학 내과 이준행

# Extracolonic features in FAP

- 위암은 흔히 언급되는 암은 아닙니다.

| Cancers | Other lesions |
|---|---|
| Meduloblastoma | CHRPE |
| Thyroid | Nasopharangeal angiofibroma |
| Duodenal | Osteomas |
| Periampullary | Radiopaque jaw lesions |
| Pancreas | Supernumerary teeth |
| Hepatoblastoma | Lipomas, fibromas, epidermoid cysts |
| Biliary tree | Desmoid tumors, **Gastric adenomas** |
| | **Fundic gland polyps** |
| | Duodenal, jejunal, ileal adenomas |

Feldman: Sleisenger's GI and Liver Disease, 7th:2002

# 2020년 ASGE guideline

**- 너무 aggressive한 것 아닌가 생각됩니다.**

- During screening and surveillance endoscopy, we recommend careful evaluation of polyps including FGPs with random biopsy sampling and **complete resection of polyps >1 cm** for the evaluation of indolent dysplasia and malignant transformation, particularly in the setting of diffuse gastric polyposis and large gastric mounds.
- All antral polyps should be endoscopically removed, given the high probability of adenoma.
- Surgery should be reserved for patients with FGP and adenomas harboring advanced histologic features who fail endoscopic management.

GIE 2020;91:963-982

# 토의 1. Extracolonic features in FAP에 속하지 않은 암도 발견될 수 있다.

| Cancers | Other lesions |
|---|---|
| Meduloblastoma | CHRPE |
| Thyroid | Nasopharangeal angiofibroma |
| Duodenal | Osteomas |
| Periampullary | Radiopaque jaw lesions |
| Pancreas | Supernumerary teeth |
| Hepatoblastoma | Lipomas, fibromas, epidermoid cysts |
| Biliary tree | Desmoid tumors, Gastric adenomas |
| | Fundic gland polyps |
| | Duodenal, jejunal, ileal adenomas |

위암
신장암

Feldman: Sleisenger's GI and Liver Disease, 7th:2002

# 토의 2. 복부 초음파나 CT 검사가 필요하다.

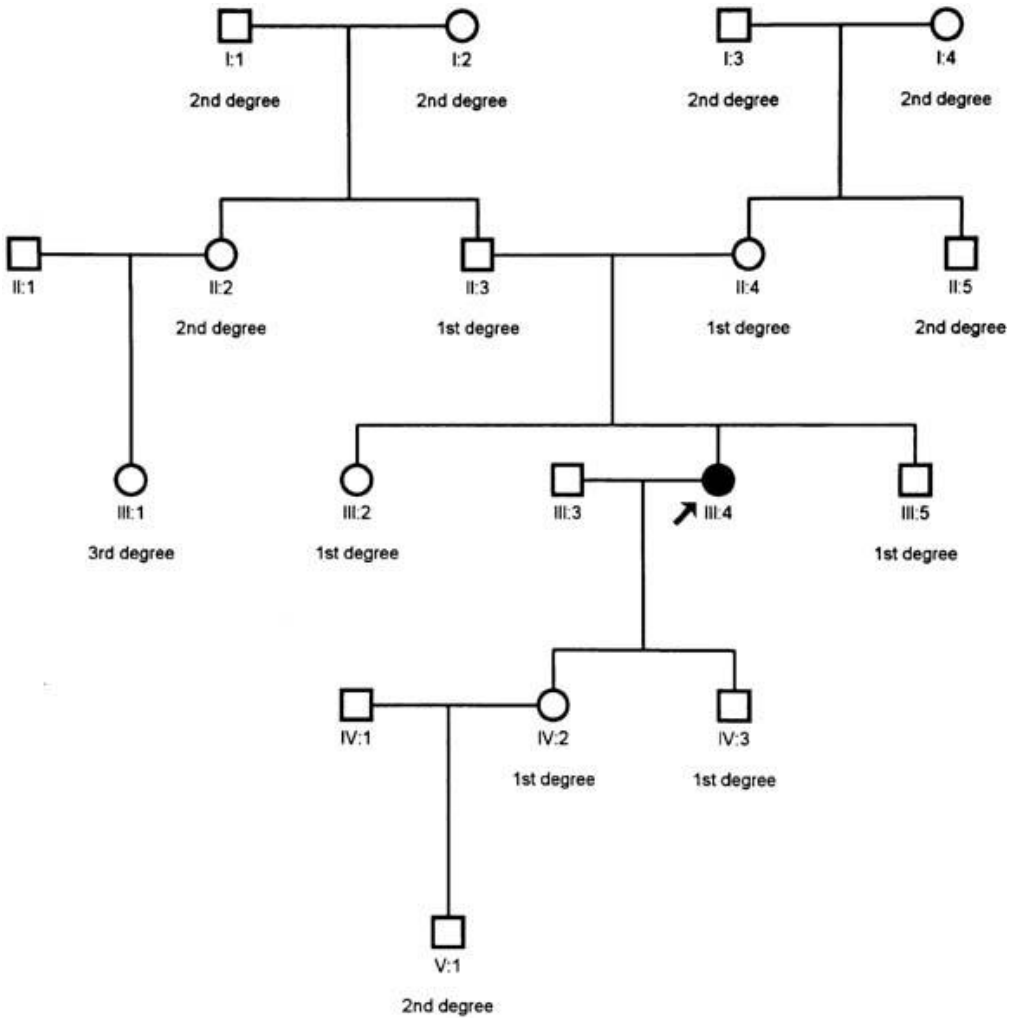
| Syndrome | Lifetime risk[a] | Screening recommendations |
|---|---|---|
| FAP | | |
| Duodenal or periampullary cancer | 5%–12% | Upper GI endoscopy (including side-viewing exam) every 1–3 yr, start at age 20–25 yr |
| Pancreatic cancer | ~2% | Possibly periodic abdominal US after age 20 yr |
| Thyroid cancer | ~2% | Annual thyroid exam, start at age 10–12 yr |
| Gastric cancer | ~0.5% | Same as for duodenal |
| CNS cancer, usually cerebellar meduloblastoma (Turcot syndrome) | <1% but RR, 92[b] | Annual physical exam, possibly periodic head CT in affected families |
| Hepatoblastoma | 1.6% of children <5 yr old | Possibly liver palpation, hepatic US, α-fetoprotein, annually, during first decade of life |

Gastroenterology 2000;119:837

# Indications of gene testing for FAP

- Affected with FAP (more than 100 colorectal adenomas)
- First-degree relatives of FAP patients
- More than 20 cumulative colorectal adenomas (suspected AFAP)
- First-degree relatives of AFAP patients

# First degree relative는 1촌이 아니다.




The arrow indicates the proband, III-4, who has hereditary colorectal cancer. The proband's first degree relatives are her parents (II-3, II-4), her siblings (III-2, III-5), and her children (IV-2, IV-3). The proband's second degree relatives are her grandparents (I-1, I-2, I-3, I-4), paternal aunt and maternal uncle (II-2, II-5), and her grandson (V-1). The proband's third degree relative is her paternal first cousin (III-1).

# Peutz-Jegher syndrome

성균관대학교 의과대학 내과 이준행

# Peutz-Jegher syndrome

- STK11 (LKB1) gene의 germline mutations (AD)
- Incidence : 1:8000-1:200,000
- 특징
  - 위장관의 multiple hamartomatous polyps
  - Mucocutaneous pigmentation (95% 이상)
- Cancer risk : 37-43%, 평균 42세
  - GI : 대장암, 췌장암
  - 위장관 외 : 유방암, 자궁경부종양

# Juvenile polyposis syndrome

성균관대학교 의과대학 내과 이준행

# Juvenile polyps

- Distinctive hamartomas that usually are solitary.
- Located principally in the rectum
- In children and occasionally in adults
- Smooth surface and covered by normal colonic epithelium

# Juvenile polyposis

- **A diagnosis of exclusion**
- The PTEN hamartoma syndromes (Cowden's disease and Bannayan-Ruvalcaba-Riley syndrome) should be excluded.

# Criteria for JPS (any 1)

- 5 or more juvenile polyps of the colon and rectum
- Juvenile polyps throughout the GI tract
- Any number of juvenile polyps in the GI tract with a family history of juvenile polyps

# Extracolonic manifestations of JPS

- CNS – macrocephaly, hydrocephalus
- Thorax – coarctation of aorta, ASD, TOF
- Urogenital – undescended testes, bifid uterus and vagina, unilateral renal agenesis
- GI – Meckel's diverticulum, malrotation

# Abnormalities associated juvenile polyposis

- Intestinal malrotation
- Meckel diverticulum
- Umbilical fistula
- Mesenteric lymphangioma
- Hydrocephalus
- Ganglioneuromas
- Cardiac lesions
- Hydrochephalus
- Amyotonia congenitum
- Hypertelorism
- Porphyria
- Undescended testis
- Supermumerary toes
- Pulmonary AVM
- Hypertrophic osteoarthropathies
- Retroperitoneal fibrosis
- Villous adenoma

# Cowden syndrome

## - A not-so-rare hereditary cancer syndrome

성균관대학교 의과대학 내과 이준행

# Cowden syndrome

- Multiple harmatomatous polyps of the skin and mucous membranes including GI polyps, facial trichilemmomas, oral papilloma and keratoses of the hands and feet
- Etiology: mutation of PTEN gene (85%)
- Autosomal dominant inherited disorder
- Multiple tumor-like growths called hamartomas and an increased risk of certain forms of cancer

# Clinical diagnostic criteria

**TABLE IV. Cowden Syndrome Clinical Diagnostic Criteria**

| Pathognomonic criteria | Major criteria | Minor criteria |
|---|---|---|
| Mucocutaneous lesions:<br>• Facial trichilemmomas<br>• Acral keratoses<br>• Papillomatous papules<br>• Mucosal lesions | Breast cancer<br>Non-medullary thyroid cancer<br>Macrocephaly<br>Endometrial cancer<br>Lhermitte-Duclos disease | Benign thyroid lesions (goiter/nodules)<br>Mental retardation<br>Hamartomatous intestinal polyps<br>Lipomas<br>Fibrocystic breast disease<br>Fibromas<br>Genitourinary tumors or malformations |
| Operational clinical diagnostic criteria for an individual: | | |
| Pathognomonic mucocutaneous lesions alone if:<br>• Six or more facial papules, at least three of which are biopsy-confirmed trichilemmomas, or<br>• Cutaneous facial papules plus oral mucosal papillomatosis, or<br>• Oral mucosal papillomatosis plus acral keratoses, or<br>• Six or more palmoplantar keratoses | | |
| Two or more major criteria, one of which must be macrocephaly or Lhermitte-Duclos | | |
| One major plus three minor criteria | | |
| Four minor criteria | | |

Mester and Eng. JSO 2014