SAMSUNG MEDICAL CENTER

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SMAD4-associated juvenile polyposis syndrome / hereditary hemorrhagic telangiectasia (JPS/HHT)

성균관대학교 의과대학 삼성서울병원 소화기내과
임상강사 박성빈

Topic review #1: clinical features and diagnostic criteria

Patient phenotype: gastric/colorectal polyposis, retention/juvenile-type polyp, pathogenic SMAD4 variant, epistaxis, telangiectasia, AVM

Clinical features

- Hamartomatous GI polyposis involving colon and stomach
- SMAD4: prominent gastric polyposis/gastropathy and HHT overlap
- HHT phenotype: epistaxis, mucocutaneous telangiectasia, visceral/GI vascular malformation

Diagnostic criteria

- **JPS**: **multiple juvenile polyps** in the GI tract, or colorectal juvenile polyps with **family history**; supported by **SMAD4/BMPR1A** pathogenic variant
- **Curaçao criteria** for **HHT**: recurrent epistaxis, mucocutaneous telangiectasia, visceral AVM, first-degree relative with HHT
- HHT classification: definite ≥3 criteria, possible/suspected 2 criteria, unlikely ≤1 criterion

Refs: Gallione et al. Lancet 2004; Aretz et al. J Med Genet 2007; van Leerdam et al. Endoscopy 2019; Faughnan et al. Ann Intern Med 2020

Topic review #2: treatment and prognosis

Treatment

- Endoscopic resection for clinically significant polyps and surveillance-detected lesions
- Surgery for cancer, severe gastric polyposis, uncontrolled bleeding, or excessive polyp burden
- Manage HHT manifestations: iron replacement/transfusion, local control of epistaxis or GI bleeding, and multidiscip linary AVM care
- Genetic counseling and cascade screening for at-risk family members

Prognosis

- Long-term risk is driven by GI cancer and HHT-related bleeding or shunt complications
- SMAD4-associated disease tends to show more prominent gastric involvement than BMPR1A-associated JPS
- Outcome improves with timely intervention and structured surveillance

Refs: Aytac et al. Br J Surg 2015; Jelsig et al. Endosc Int Open 2022; Faughnan et al. Ann Intern Med 2020

Topic review #3: SMAD4 vs BMPR1A

SMAD4

- Frequent overlap with HHT phenotype
- Marked gastric polyposis / gastropathy is charac teristic
- Upper GI neoplasia, including gastric cancer, is a greater concern
- Upper GI surveillance deserves particular empha sis

BMPR1A

- No typical HHT overlap
- Gastric involvement is usually less prominent
- Gastric cancer concern is lower relative to SMAD 4
- Surveillance still required, but phenotype is ofte n less gastric-dominant

Key message: SMAD4-associated JPS shows more prominent gastric involvement than BMPR1A-associated JPS, and gastr ic cancer is reported more often in SMAD4 carriers.

Refs: Aytac et al. Br J Surg 2015; Jelsig et al. Endosc Int Open 2022; van Leerdam et al. Endoscopy 2019

Topic review #4: surveillance

GI surveillance

- EGD and colonoscopy every 1–3 years, adjusted by phenotype and polyp burden
- Shorter intervals when severe gastric polyposis, dysplasia, or prior neoplasia is present
- Polypectomy and pathology-directed follow-up for suspicious or symptomatic lesions
- Consider small-bowel evaluation when syndrome features or symptoms warrant it

HHT surveillance

- Screen for pulmonary AVM and brain vascular malformations; evaluate liver involvement when indicated
- Monitor hemoglobin, iron deficiency, epistaxis, and GI bleeding over time
- Continue family screening and syndrome-specific counseling

Refs: van Leerdam et al. Endoscopy 2019; Faughnan et al. Ann Intern Med 2020; Jelsig et al. Endosc Int Open 2022

Take-home messages

1. **Do not anchor on cancer alone.**
   대장암처럼 보이는 병변도 영상·표재 생검·절제 병리가 맞지 않으면 vascular lesion을 다시 생각한다.

2. **Recognize the inherited pattern.**
   젊은 나이의 위·대장 다발성 용종, juvenile/retention-type polyp, 가족력은 germline polyposis panel로 이어져야 한다.

3. **SMAD4 changes upper GI surveillance.**
   SMAD4-associated JPS/HHT에서는 위용종증과 위암 위험을 염두에 두고 위 병변의 형태 변화를 적극적으로 확인한다.

4. **Separate evidence from patient-specific judgment.**
   HER2 양성 위암의 근거, HHT/AVM 관련 출혈 위험, 수술 시점은 다학제로 나누어 판단한다.