■ Sat-036 ■

A case of hereditary hemorrhagic telangiectasia presented with upper gastrointestinal bleeding

강릉아산병원 내과

*이종윤, 박종규

Hereditary hemorrhagic telangiectasia (HHT) also known Osler-Weber-Rendu syndrome, is a rare autosomal-dominantly inherited disorder. The most common clinical manifestations with HHT patients are epistaxis and gastrointestinal bleeding. Gastrointestinal bleeding usually begins in the fourth or fifth decade and occurs in 13-33% of HHT patients. Telangiectasia is found more common in the stomach and duodenum than in the colon. We report here the case of a 62 year-old female patient who presented melena and coffee grounded vomitus. She had a history of treatment with recurrent spontaneous epistaxis for five years. Family history revealed that her mother also had recurrent episodes of epistaxis. Physical examination revealed multiple tongue telangiectasias (Fig. 1). Esophagogastroduodenoscopy showed multiple angiodysplasia without current bleeding on antrum, body and fundus (Fig. 2). There was epistaxis during endoscopy. Image reconstruction of abdomen CT angiography revealed hepatic artery hypertrophy and early enhancement of portal vein and hepatic vein, suggesting arterio-portal and arterio-venous shunt (Fig 3). On the basis of these findings, the patient was clinically diagnosed with HHT based on Curaçao clinical criteria. The Curaçao criteria consist of four criteria of (1) spontaneous recurrent epistaxis, (2) multiple mucocutaneous telangiectasia at lips, oral cavity, finger and nose, (3) visceral lesions including gastrointestinal telangiectasias, pulmonary, hepatic, cerebral and spinal arteriovenous malformation, and (4) an affected first-degree relative. These define “definite HHT” where three criteria are present. Our patient had all the four criteria present. If a patient presented with gastrointestinal bleeding has recurrent episodes of spontaneous epistaxis and multiple angiodysplasia on endoscopy, family history taking of bleeding episodes and radiologic evaluation is necessary to differentiate HHT.

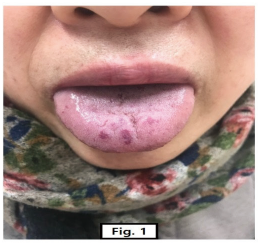
Fig. 1

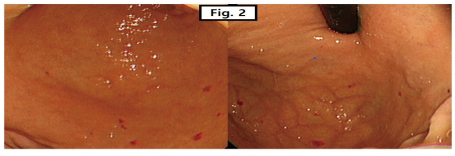
Fig. 2

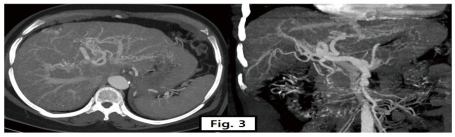
Fig. 3