A Case of Juvenile and Adenomatous Polyposis Associated with Telangiectasia and Colonic Carcinoma

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Özet: *40 yaşında erkek hastada juvenil ve adenomatöz polipler, herediter hemorajik telanjiektaziler, kolon karsinoması, kısa boy ve kromozomal defektlerle yeni ve ilginç bir kompleksin mevcudiyeti rapor edilmiştir. Çocukluğundan beri sık tekrarlayan burun kanamaları ve hematoşezi nedeniyle incelenen hastada dudak, burun, dil, avuçları ve mide de telanjiektaziler belirlendi. Endoskopik incelemede duodenumda bir tanesi 2 cm çaplı saplı polip ve sayısız sesil polipler, kolonda biri malign görünümlü üç polip izlendi. Kromozom analizinde 5. kromozomda delesyon olduğu belirlendi. Literatürde benzer ancak daha az ögeli raporlarla beraber değerlendirildiğinde geniş bir genetik sendromun muhtemelen aynı mozaiği en geniş biçimde tamamladığı düşünülen bir vaka olarak takdim edildi.*

Anahtar Kelimeler: Palıp, telanjiektazi, kromozom, kolon karsinomu

Summary: *A 40 years old white male patient having both intestinal juvenile and adenomatous polyposis associated with hereditary hemorrhagic telangiectasia, colon carcinoma, short stature and abnormalities in chromosome 2 and 5 has been presented. This combination has not been reported previously. The findings of the patient were consisting of hemorrhagic telangiectasia on his lips, tongue, palms, nasal and stomach mucosa; numerous polyps in duodenum; three colon polyps, which one was neoplasic, and triradiol chromosome at A2 autosome and a deletion on the long arm of 5th chromosome.*

Key Words: Polyps, telangiectasia, chromosome, colon carcinoma

Intestinal polyps which are masses protruding into the lumen may be solitary or a part of a syndrome. A number of syndromes including polyps with other abnormalities have been defined up to date (1).

Polyps together with Osler-Weber-Rendu Syndrome has been described in few case reports. This syndrome is a vascular anomaly characterized by telangiectatic lesions of the skin and mucosa, inherited as an autosomal dominant trait. Rarely involvement of visceral vessels (e.g., pulmonary arteriovenous fistulas) may also be seen (1).

In this paper, a case having a combination of both adenomatous and juvenile polyps, telangiectasia, colon carcinoma and deletion in 5th chromosome is presented.

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CASE REPORT

Patient O.B., a 40-yr-old white man, presented to our clinic with rectal bleeding, malaise and epistaxis complaints in October 2, 1992. At 7 yr of age, oral bleeding and epistaxis which could be controlled medically each time had begun. At 11 yr of age, his first rectal bleeding had occured. A polypectomy had done endoscopically from sigmoid colon when he was 15 years old. After this procedure he hadn't experience any rectal bleeding for 8 years, however his epistaxis continued. His family history revealed his mother and grandmother had colon tumours and both died with extensive metastases.

The physical examination revealed short stature, numerous telangiectatic lesions on his lips, oral mucosa, tongue (Figure 1), nose and palms.

Laboratory tests showed a mild iron deficiency anemia and positive occult blood in stool. Colo-

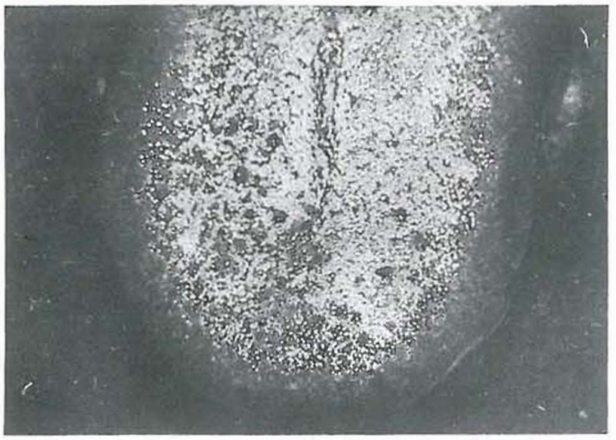

Figure 1: Telangiectatic lesions on the tongue of the patient.

noscopy showed three polyps in the colon, one at the tenth centimeter of rectum, second at splenic flexura and the third which was endoscopically malignant at hepatic flexura. Biopsies revealed mild dysplastic adenomatous polypoidal growth in the distal two polyps (Figure 2), while adenocarcinoma in the polyp at hepatic flexura.

Upper gastrointestinal endoscopy showed telangiectasia localized to stomach and numerous polyps in duodenum which some were sessile (Figure 3) and some pedunculated. Histopathological investigation showed that these were juvenile polyps (Figure 4).

Pulmonary angiography was normal. Chromosomal analysis by bone marrow and peripheral lenfocyte culture showed a deletion at the long arm of 5th chromosome (Figure 5). In the caryo-

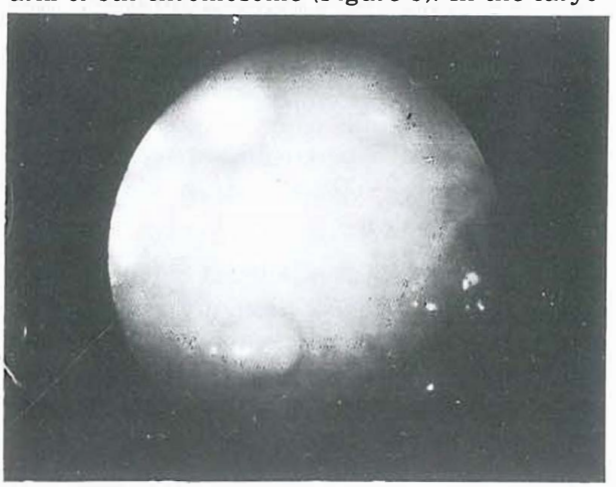

Figure 3: Polyps localized at duodenum.

Figure 2: The polipoid proliferation of the edematous stroma rich of inflammatory cells and fibrous connective tissue with increased number of cryptae lined with dysplastic epithelial cells are seen. (H-E, x32).

type stained by Giemsa showed a triradiol chromosome at A2 autosome (Figure 5). In band staining, a minimal deletion at the fragil area of C9 was also present.

Computerized abdominal tomography revealed a wall thickening and lumen narrowing at the right flexura of colon and a mass at the rectosig-

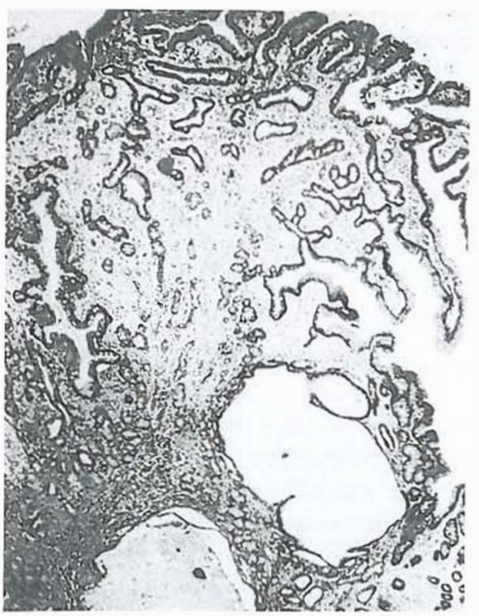

Figure 4: The cryptae and glanduler formation some showing cystic dilatations and have secret-like material within the luminae are seen in the loose, edematous stroma with scattered inflammatory cells. (H-E, x32)

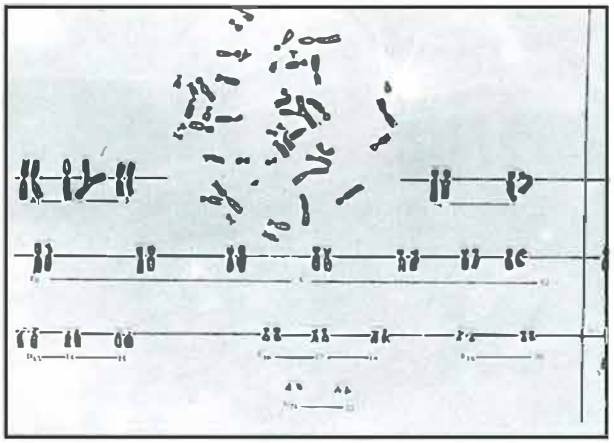

Figure 5: Chromosomal analysis with a caryotype stained by Giemsa showing a triradiol chromosome at A2 autosome and a deletion on the long arm of 5th chromosome.

Figure 6: An irregularly shaped glandular formation formed by a typically malignant epithelial cells is seen (arrow) in the submucosal layer of the mucosa within the pedunculated polipoid structure. (H-E, x32).

moidal junction protroding into the lumen were noted. Other intraabdominal organs were within normal ranges.

An ileoproctostomy and duodenal polypectomy have been performed to the patient. Histological analysis confirmed the endoscopic diagnosis of adenocarcinoma at the right flexura of colon (Figure 6). He is well now and waiting for the second session of septal dermoplasty operation that had been planned for his epistaxis. The control colonoscopic examination showed a normal rectum being followed by a normal ileum with no polyps.

DISCUSSION

In this case, intestinal and adenomatous polyps with telangiectasia, colon adenocarcinoma and deletional abnormality of chromosome 5 have been demonstrated as a new combination.

Multiple congenital polyposis of the colon is transmitted as autosomal dominantly, and thus is present in the other members of the affected family. The polyps are not present at birth but appear in childhood and adolescence giving rise to diarrhea or rectal bleeding (1). As another entity, juvenile polyposis may be seen in colon, but occurs mainly in small bowel. They don't show malignant degeneration, while familial colonic polyposis has 100% malign transformation (1).

Our patient had both juvenile and adenomatous polyps, localized in duodenum and colon, respectively. Histological examination showed juvenile polyps in duodenum, adenomatous and carcinomatous polyps in colo-rectum. His family history was reinforcing that this combination may be related to a genetic defect. His telangiectatic lesions on skin and mucous membranes which are characterized in Osler-Weber-Rendu syndrome had been described as an autosomal dominantly inherited disorder.

Unlike Cronkhite-Canada syndrome, our patient didn't have any ectodermal abnormality as alopecia, hyperpigmentation or onychodystrophy. He didn't have any melanosis which could suggest Peutz-Jeghers syndrome, either.

In a report, three cases had been described with mixed polyps associating juvenile and adenomatous polyps with pulmonary arteriovenous malformation and hypertrophic osteoarthropaty (2,3). In these cases there were no telangiectatic lesions nor carcinoma.

Also in a report about a 28 years old mother and her daughter with generalized juvenile gastrointestinal polyposis, arteriovenous malformations of the lung, severe digital clubbing and telangiectatic lesions on their lips had been presented (4). They didn't have any adenomatous polyps, malignant features and any mention about their chromosomal analysis.

In a report about a brother and his sister having juvenile polyposis and A-V malformations and colonic adenocarcinoma in their father had proposed that these lesions are inherited concurrently as the pleitropic manifestations of an autosomal dominant gene (5).

Abnormalities of chromosome 5 have been demonstrated and reported in patients with colorectal carcinoma and familial polyposis (1,6,7). The familial polyposis gene (FAP) is localized in chromosome 5 and in about 20% of patients with sporadic colon cancer one of the alleles is lost (1,6). In a report, three patients, with one having polyposis of the colon had constitutional interstitial deletions of the long arm of chromosome 5. Polyposis coli was together with a deletion in the region 5q21-q22, which is a gene involved in familial adenomatous polvps and sporadic colon cancer (6,7). Abnormalities of chromosome 8 and 14 have been shown to precede histological evidence of invasiveness and these findings may tend to link polyp with carcinoma (1). We couldn't detect any abnormality in these chromosomes.

Since studies imply that colon cancer may develop from polyps, early and periodic surveillance of family members is important. Our patient could be detected more earlier regarding his family history. In our patient association of juvenile, adenomatous polyps, colon carcinoma, hereditary hemorrhagic telangiectasia might be accepted as a new combination. Other cases in previous reports (2,3,4,5) seem to carry some parts and features of the same mosaic, but not completely.

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